

Genexus™ Software 7.0

USER GUIDE

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B	13 March 2026	Software security information added to the following topics: • Software compatibility and requirements • Network configuration and security • Data backup and storage • Antivirus software • Create a single sample • Create multiple samples • Create multiple samples with a sample definition file • Manage software updates • Download and install software updates from the App Store • View system services Added technical clarifications and corrections, and/or information related to custom Ion AmpliSeq™ or Ion AmpliSeqHD™ assays to the following topics: • Prepare a library batch • Plan a Sample to Result run • Run plugins after a sequencing run is complete • Copy an assay • Edit the QC parameters of a locked assay • Create a copy number baseline for use with Ion AmpliSeq™ libraries • Create a copy number baseline for use with Ion AmpliSeq™ HD libraries • Create a copy number baseline for low-coverage whole-genome libraries • Sequence variant baselines (manager/administrator) • Create a sequence variant baseline • Consensus sequence summary statistics • Import or add a custom panel • Configure email notifications • Customer Support Archive • Create a library batch • Create samples • Create BAM samples • Create a sample tag by display name • Custom panels • Download result files • Review sampleID plugin results in Genexus™ Software • Get the authenticator token • Get a list of Ion Reporter™ Software accounts • Get a list result files • Get a list of variants • Get instrument status • Get library batches

Revision	Date	Description
B (continued)	13 March 2026	• Get a list of report templates • Get a list of run plans • Get a list of samples • Get a list of signed-off samples • Get a list of variants • Get a list of variants in Ion ReproSeq™ assays • Get analysis results • Get result details for a sample • Get sample status • OncoPrint™ filter • Non-targeted fusion detection • Results files • System-installed filter chains • Update samples • Variant identifiers for OncoPrint™ panels • System-installed filter chains
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Software overview

Genexus™ Software supports the Ion Torrent™ Genexus™ Integrated Sequencer workflow for research purposes, from sample preparation through library preparation, template preparation, and sequencing. During and after sequencing, the software generates base calls, trims reads, and determines quality values (that is, primary analysis), then aligns reads, calls variants, and generates reports (that is, secondary analysis).

The software also supports run plans for nucleic acid isolation on the Ion Torrent™ Genexus™ Purification System that are followed by sequencing on the Genexus™ Integrated Sequencer.

Instructions and explanations for the following features and functions are included in this documentation.

Organizations

Genexus™ Software provides the ability to create organizations for different application categories, and to assign members to organizations based on their work assignments. An organization determines which application categories are available to members, and gives members access to specific software components, such as assays, presets, and results, that apply to an application category. Members do not have access or visibility to other organizations, application categories, or components.

Application categories

Application categories are used to group the various types of research that can be done in Genexus™ Software. Application categories consist of groups of assays and software capabilities to provide output that is relevant to the research needs for the application. Examples of application categories are **Oncology**, **Reproductive** and **Infectious/Microbial**.

Entering sample information

Information that represents data about a physical sample, samples that will be purified or sequenced, or libraries can be entered into the software in the following ways.

- With a Excel™ spreadsheet that contains sample information. A template file to define sample information is provided in the software.
- Individually in the software screen.
- With a BAM file or a CRAM file that contains sample reads. These sample files are either generated from a previous run on the same sequencer, or from BAM or CRAM files generated on a different sequencer then uploaded to Genexus™ Software.

Run plans

Run plans created in Genexus™ Software include the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes. Run plans are used to track samples, consumables, and chips throughout the library preparation, templating, sequencing, and data analysis workflow. Run plans are based on the selected template. Run plans can be used to purify samples only, to sequence samples with previously prepared libraries, to sequence samples of purified nucleic acid, to sequence samples starting with purified nucleic acid samples and followed by sequencing, or analyze samples from BAM or CRAM sample files. When a run is planned, a Excel™ spreadsheet template that provides flexibility for rearranging samples in the sample plate of the Genexus™ Integrated Sequencer is available. Samples are automatically loaded into the desired well positions based on the information in the spreadsheet.

Assays

Assays are reusable experimental designs that you add to run plans in Genexus™ Software. Assays include settings and parameters for library preparation, templating, controlling the sequencing run, analyzing, and reporting the results. Assays also define the panels, kits, and chips that are used in a run, and specify the reference files and threshold values for quality control and variant detection. The software files that include the assay settings and parameters are packaged in a ZIP file called an assay definition file (ADF). You can create new assays to meet your needs from system-installed assay templates, or copy and edit assays that are previously created or edited in the software.

Presets

Annotation sets, genome references, gene lists, filter chains, report templates, baseline files, primer sets, plugins, and reference files can be added to assays in Genexus™ Software to refine, filter and present sample results to meet your needs.

Dashboard

Status and details for connected instruments and up to five recent sequencing or purification runs are shown in the **Dashboard**. Progress is also shown for each phase of a run including purification, quantification, library preparation, templating, sequencing, analysis and plugin runs. Access to run and sample results for runs that are complete, by run name, sample name or by assay name is available in the **Dashboard**.

Results

You can view sample results, including detailed information about variants, and key findings for samples. You can also view QC metrics, results from plugins that were used during the sequencing run, and reports that are associated with the sequencing run. Sample results are based on the filter chain or gene list, or both, that are associated with the assay, and those variants are selected by default the first time that sample results are reviewed. You can further refine sample results if you change or apply a filter chain or gene list or deselect or select variants. All selected variants are included in reports.

You can download results files for each sample. Results files include the sequencing data, results from the analyses, such as variant files, and audit and log files.

Table preferences for sample results

You can customize the view of sample results to reflect the information that is most relevant to your research and reports.

You can create a table preference to customize the columns that are shown in the Variants tab. The table preference that is applied to the sample results can be associated with an assay, or applied only to the results that have the same library chemistry and research application, or can be associated with a combination of library chemistry and research application, or applied later to results that you view.

Fusion analysis pipeline changes

Changes in the fusion algorithm include small bug fixes and specific improvements to targeted fusions, non-targeted fusions and Exon-tiling imbalance detection. The algorithm retains its primary alignment and analysis components, and compatibility with a sample-to-result workflow in Genexus™ Software. Several new parameters were introduced in the software to improve the specificity of novel fusion detection (non-targeted method), and to limit reporting of syntenic (same chromosome) fusion isoforms between two genes at close genomic proximity. Such fusions have a higher background of fusion reads. Additionally, changes in the Exon-tiling imbalance algorithm were made with the goal of improving imbalance specificity by using normalized exon-tile read coverage and by flagging likely expression of short alternative transcript (ALK-ATI) in the ALK gene.

Parameter name	Description	Value Type, range	Example value
Syntenic Fusions Max Distance	Maximum genomic distance (bp) below which, a pair of genes in a fusion isoform is considered syntenic.	Integer >0	500000
Syntenic Fusions Minimum Read Counts	Minimum number of valid reads aligned to a syntenic fusion isoform sequence to call the isoform as Present .	Integer >0	1000
Syntenic Fusions Normalized Read Counts	Minimum normalized read counts threshold to call a syntenic fusion isoform as present.	Float 0-1.0	0.001
Minimum Normalized Read Counts for Non-Targeted Fusions	Minimum normalized read counts threshold to call a non-targeted fusion as present.	Float 0-1.0	0.001

SDK to create user-defined plugins

Using design specifications provided in the Genexus™ Software help or software user guide, you can create a plugin that can be used in Genexus™ Software 7.0.

APIs

Genexus™ Software provides two APIs to integrate laboratory information management systems (LIMS) to help with sample tracking and variant reviews.

- Genexus™ Software Service API can be installed with the **Software Update** feature and is provided with Genexus™ Software 7.0. The service can be updated independent of software updates.
- Genexus™ Software API – Uses a set of RESTful APIs included in the software that connects to the LIMS with an HTTP call to the URL of an API, and sends a user name and a password in the HTTP header.

Reanalysis

If a sequencing run fails to meet one or more QC parameters defined by the assay, you can adjust the assay parameters and reanalyze a run or a sample from the run. You can reanalyze a sequencing run with the same assay or with a different assay in Genexus™ Software. Reanalysis can start from the alignment, basecalling, or signal processing steps. When you reanalyze a run, the reanalysis is applied to all samples in the assay.

Audit records

You can review and download audit records for changes made in the software. Each audit record lists the change that was made, and include a timestamp for the change, the old and new values, and the user who made the change.

Software updates

Updates to the ClinVar annotation source, assay definition files (ADFs), configuration files, oncology content update packages, and security patches, can be installed in the software.

Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Software 7.0 or later. Version-specific information is provided in the software release notes for your version of the software. The software version is shown in the bottom footer of each screen in the software.

Genexus™ Software is supported on Google Chrome™ browser version 90 and later and is best viewed with 1440 × 900 screen resolution. Google Chrome™ browser is recommended for use with the software.

English must be the primary language that is set in the web browser in which you use Genexus™ Software when you download a template file for sample creation.

The operating system of the sequencer is Ubuntu™ 18.04.1 LTS. The operating system has an Ubuntu™ Pro license that includes expanded security maintenance coverage.

It is recommended that Genexus™ Software be used only on computers that are maintained by facility system administrators and IT and security personnel, and include security software, including but not limited to firewalls, antivirus software, encryption-based passwords systems.

To upload sample results from a sequencer to an Ion Reporter™ Software to further analyze the results, you must configure a connection in Genexus™ Software to Ion Reporter™ Software 5.16 or later. For more information, see “IonReporterUploader plugin” on page 423.

Software anomalies

For Genexus™ Software anomalies, see the release notes for your version of the software available at <http://www.thermofisher.com/ngssoftwaresupport>.

Network and password security requirements

Network configuration and security

The network configuration and security settings (for example, firewalls, antivirus software, network passwords) of your laboratory or facility are the sole responsibility of facility system administrators and IT and security personnel. Genexus™ Software does not provide any network or security configuration files, utilities, or instructions. The Genexus™ Integrated Sequencer has a firewall enabled by default. Only ports required for the operation of the instrument and software are open.



CAUTION! Data can be lost or corrupted if you operate a sequencer that does not have sufficient security controls in place.

If external or network drives are connected to Genexus™ Integrated Sequencer for archiving or backups, it is the sole responsibility of IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss.

If a Laboratory Information Management System (LIMS) connection is set up, you can use the LIMS API to obtain the list of files generated for the assay. For more information, see Appendix B, “Genexus™ Software Service API” and Appendix A, “Genexus™ Software API”.

Data backup and data storage

You must have an established setup for regularly backing up and archiving your data. Data backup is solely your responsibility. In the event your system needs repair, Thermo Fisher Scientific is not responsible for data backup or any loss of data.

Note: A paid data migration service is available if needed. Contact your Thermo Fisher Scientific representative for more information.

For more information about data storage compatibility, contact your local support team.

Password security recommendations for Genexus™ Software

Thermo Fisher Scientific strongly recommends that you maintain unique passwords for all accounts in use on this product. Passwords must be reset upon first sign in to the product. Change passwords according to the password policies of your organization.

It is the sole responsibility of your IT personnel to develop and enforce secure use of passwords.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

Antivirus software

For recommendations for antivirus software that is compatible with Genexus™ Software, see the current version of the Genexus™ Software release notes.

Installation and verification run

The Genexus™ Integrated Sequencer and the Genexus™ Software provide the option to perform a performance qualification (PQ) verification run.

Notifications are sent when the PQ run is expired. Contact your Field Support Engineer (FSE) or Technical Support if you have questions about an expired PQ status.

Reagent management

Put in place a reagent management system to inventory and track reagent use.

Proper use of a reagent management system enables optimal use of kits, chips, and reagents.

System tracking

The system tracks and checks auditable systems and user activities for samples, assays, QC metrics, and information related to reagents.

If the software detects an error at any step—for example, a scanned barcode is inconsistent with the information given for the run plan—the software alerts the user and does not proceed with the run.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

User-access levels

Users at this level	Can perform these functions
Reporter	• Access the Sample Results and Run Results screens to view results. • Generate, view, and sign variant reports. • Send notifications. • Download results files. • View and edit notes for the sample result. • View the audit trail for sample results. • View notifications.
Operator	Reporter functions, plus the following functions. • Add, import, and export sample files. • Edit sample files. • Prepare library batches. • Plan, save, audit, and delete runs. • Monitor runs. • View results and reports. • Upload sample results files to Ion Reporter™ Software. • View the audit trail for samples, library batches, runs, and assays. • Reanalyze runs and run plugins.
Manager	Operator functions, plus the following functions. • Create, edit, and obsolete sample attributes. • Create and import assays. • Create presets for annotation sets, filter chains, copy number baselines, sequence variant baselines, exon tile assay baselines, and report templates. • Edit and delete sample files. • Add or edit Thermo Fisher Scientific accounts. • Manage gene lists. • Restart a stalled or failed run. • Manage reference sequences and panel, hotspot, and other sequence files. • Access services information. • Create and manage user-specific organizations.

(continued)

Users at this level	Can perform these functions
Organization administrator	Operator and manager functions, plus the following functions. • View, export, and print audit records. • Configure network settings. • View and manage software updates. • Install and manage plugins. • Configure backup settings and restore runs. • Manage sequencer files and software log files. • Add and manage user accounts. • Create and manage user-specific and application-specific organizations.
System administrator	Reporter, operator, manager, and organization administrator functions, plus the following system-level functions. • Create and manage organizations. • Configure system-wide email settings and notifications. • Backup and restore the system. • Configure and manage logs. • Configure Oncomine™ Reporter settings. • Configure and manage system services. • Install software updates. • Link Thermo Fisher Connect account to devices. • Configure system-wide user management policies. • Configure audit rules. • Manage SSH keys • Sign off verification results. • Import assays (factory shipped).

Genexus™ Software concepts

The following terms are used in Genexus™ Software.

For more definitions, see “Genexus™ Software definitions” on page 593.

Term or concept	Description	Example
Sample	The information that describes the material from a single source. Each sample is defined by a set of attributes, which are categories of information that describe important aspects of each sample, such as date of sample collection, gender, or a sample type. In the software, a sample can be isolated nucleic acid, or a sample can be a specimen that requires nucleic acid purification before sequencing.	Data from a biopsy or blood draw.
Sample name	A unique identifier for a specific sample.	Sample1
Assay	A collection of settings and parameters for library preparation, templating, sequencing, and sequencing data analysis.	Oncomine™ Precision Assay GX
Library	A sample associated with an assay, including barcode information and sample input quantity. Libraries are associated with an assay during library batch creation.	DNA from a non-small cell lung cancer specimen that is sequenced with the Oncomine™ Precision Assay GX.
Library batch	A collection of libraries that are prepared and sequenced in the same run under the same conditions on the Genexus™ Integrated Sequencer.	A library batch that consists of one library of sample DNA and another library of sample RNA (from the same sample).
Library batch ID	A unique identifier for a library batch.	DNA_B1_Lib123456_Samples123
Download	Term used to describe downloads of files from Genexus™ Software for the following file types: • Audit record files • Sample results and run results files • Parameter files • Template files	Download and edit a template file to create a new file. Download parameter files from the Manage Assay screen.

(continued)

Term or concept	Description	Example
Export	Term used to describe downloads of files from Genexus™ Software for the following file types: • Variant files • Panel files • Sample files • Baseline files • Assay files (before imports of an assay to another Genexus™ Integrated Sequencer).	You can export the information for all variants, or for selected variants.
Analysis pipeline version	The version for a collection of algorithmic modules that analyze data and identify variants including SNPs, INDELs and fusions, and biomarkers, and can include the annotation of variants. Each assay in the software is associated with an analysis pipeline version. The annotation sets that are available in the software when you create or edit an assay are compatible with the same version of that analysis pipeline as the assay.	5.14, 5.16, 5.18, 6.8, 7.0
Archive	The process that occurs when data for runs on the Ion Torrent™ Genexus™ Integrated Sequencer are moved to the external storage and is no longer available for review on the sequencer.	N/A
Backup	The process of data being copied to an external storage device or system from the Genexus™ Integrated Sequencer. Backups of data are scheduled based on backup settings, and data that is backed up is not deleted from the sequencer. When data are backed up, the same data remains on both the sequencer and on the external storage system.	N/A

Workflows

Workflow diagrams provide high-level views of the steps involved in Genexus™ Software to prepare a purification run or a sequencing run.

Type of workflow	More information
Sample to Result	"Sample to Result workflow" on page 28
Nucleic Acid to Result Library to Result	"Nucleic Acid to Result or Library to Result workflow" on page 30
BAM/CRAM to Result	"BAM/CRAM to Result workflow" on page 32
Purification Only	"Purification Only workflow" on page 33

Sample to Result workflow

This is a workflow for a **Sample to Result** purification and sequencing run that is planned in Genexus™ Software and run on the Genexus™ Purification System and the Genexus™ Integrated Sequencer.

Sample to Result workflow

Create and manage assays (page 233)

System-installed assays that are specifically configured for each sample type are available in the software. You can use the system-installed assays in a run without change, copy the system-installed assay that best represents the experiment, then edit the assay settings in an existing assay, or create a new assay.

Manage samples and library batches (page 41)

Enter samples in software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



Plan a Sample to Result run (page 89)

Runs created in the software include the settings that are used in purification, library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

Prepare samples and load the purification instrument

Load reagents on to the Genexus™ Purification System for purification of DNA, then start the run.

For more information, see the *Genexus™ Purification System User Guide*, (Pub. No. [MAN0018475](#)).



Review purified samples for a Sample to Result run (page 94)

You can review samples before sequencing.



Load the sequencer and start a run (see user guide for the instrument or assay)

Follow the step-by-step instructions about the sequencer touchscreen to load the sample plate and consumables into the sequencer.



Sample to Result workflow

Monitor the Dashboard (page 114)

Use the **Dashboard** to view and monitor instrument status and details for recent and current runs.

Review results (page 118)

Review data and results in the software.

You can also analyze data in Ion Reporter™ Software using an Ion Reporter™ analysis workflow.



Nucleic Acid to Result or Library to Result workflow

This is a workflow for a **Nucleic Acid to Result** sequencing run or a **Library to Result** sequencing run that is planned in Genexus™ Software and run on the Genexus™ Integrated Sequencer.

Nucleic Acid to Result or Library to Result workflow

Create and manage assays (page 233)

System-installed assays that are specifically configured for each sample type are available in the software. You can use the system-installed assays in a run without change, copy the system-installed assay that best represents the experiment, then edit the assay settings in an existing assay, or create a new assay.

Manage samples and library batches (page 41)

Enter samples in the software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



Plan a Nucleic Acid to Result run (page 97) or Plan a Library to Result run (page 103)

Runs created in the software include the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

Dilute samples and load the sample plate (see the instrument or assay user guide)

Quantify and dilute the nucleic acid samples, then load the sample plate.



Load the sequencer and start a run (see the instrument or assay user guide)

Follow the step-by-step instructions about the sequencer touchscreen to load the sample plate and consumables into the sequencer.



Nucleic Acid to Result or Library to Result workflow

Monitor the Dashboard (page 114)

Use the **Dashboard** to view and monitor instrument status and details for recent and current runs.



Review results (page 118)

Review data and results in the software.

You can also analyze data with an analysis workflow in Ion Reporter™ Software.

BAM/CRAM to Result workflow

This is a workflow is for a **BAM/CRAM to Result** analysis run that is planned in Genexus™ Software and run on the Genexus™ Integrated Sequencer.

BAM/CRAM to Result workflow

Create and manage assays (page 233)

Use a system-installed assay or create a user-defined assay.

Create BAM and CRAM samples from a sample definition file (page 50)

Upload a BAM or CRAM file to enter samples in the software. Then define sample names and provide other information such as sample collection date, gender, sample type, and application category.



Plan a BAM/CRAM to Result run (page 106)

Plan a **BAM/CRAM to Result** run, then save and launch it in the software. Runs created in the software contain the assay settings that are used for the analysis of samples.

Review results (page 118)

Review data and results in the software.



Purification Only workflow

This is a workflow for a **Purification Only** run that is planned in Genexus™ Software and run on the Genexus™ Purification System.

Purification Only workflow

Create and manage assays (page 233)

System-installed assays that are specifically configured for each sample type are available in the software. You can use the system-installed assays in a run without change. To modify any assay settings, copy the system-installed assay that best represents the experiment, then edit the assay settings if needed.

Manage samples and library batches (page 41)

Enter samples in the software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



Plan a Purification Only run (page 95)

Runs created in the software contain the settings that are used in purification, library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

Prepare samples and load the purification instrument

Load reagents on to the Genexus™ Purification System for purification of DNA, then start the run.

For more information, see the *Genexus™ Purification System User Guide*, (Pub. No. [MAN0018475](#)).



Review purified samples (page 96)

You can review samples before sequencing.



Load the sequencer and start a run (see user guide for the instrument or assay)

Follow the step-by-step instructions on the sequencer touchscreen to load the sample plate and consumables into the sequencer.



Purification Only workflow

Monitor the Dashboard (page 114)

Use the **Dashboard** to view and monitor instrument status and details for recent and current runs.

Review results (page 118)

Review data and results in the software.

You can also analyze data in Ion Reporter™ Software using an Ion Reporter™ analysis workflow.



Review purified samples (page 96)

Review samples in Genexus™ Software that were purified on a Genexus™ Purification System.



Get started with Genexus™ Software

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Sign in to a new user account

Users must request a new user account from administrators before they can sign in to Genexus™ Software. Administrators receive the request and create the user account. After the administrator creates a new user account, the software sends an email to the new user with a user name and temporary password. The user name and password must be unique and must not be shared with other users.

1. Obtain your user name and temporary password that is supplied by your administrator in an email message.
2. Browse to the Genexus™ Software sign in page (for example, <https://<your server address>>), then enter your user name and the temporary password.
3. Press **Enter**, or click **Sign In**.
4. In the **End User License Agreement** that appears at first sign on, read the agreement, then click **Accept**.
5. In the **Change Password** screen, enter your temporary password, type a new password, confirm the password, then click **Change**.

The software opens to the **Dashboard**.

Sign in

Before you can sign in to Genexus™ Software, you must have a registered account. See “Sign in to a new user account” on page 35.

1. Browse to the Genexus™ Software sign in page (for example, *<https://<your server address>>*).
2. (Optional) Click **Switch Language**, then select the preferred language from the list.
3. Enter your user name and password, then press **Enter** or click **Sign In**.

If a message appears that indicates that there is a problem with the security token, refresh the browser, then enter the user name and password again.

The software opens to the **Dashboard**.

Genexus™ Software Dashboard

The screenshot displays the Genexus Software Dashboard interface. At the top, a navigation bar includes links for Dashboard, Samples, Runs, Results, Assays, and a user profile icon. The main content area is divided into several sections:

- Recent Runs (1):** A table listing recent runs with columns for run name and status. The selected run is **Cpy_OCAv3_DnF_47**, which is **Analysis Completed**.
- Instruments (2):** A section showing connected instruments. **GNXP-0017** is listed with a status of **Run Completed**. **System1** is also shown with a status of **Templating Completed**.
- Run Details (3):** A detailed view of the selected run **Cpy_OCAv3_DnF_4778439**. It shows a progress bar for the run stages: Purification, Quantification, Library, Templating, Sequencing, Analysis, and Plugins. The **QC Status** is **5 Pass**.
- Results (4):** A table showing the results for the selected run, including sample names and their status. The samples are **SRTestSample01**, **SRTestSample02**, **SRTestSample03**, **SRTestSample04**, and **SRTestSample05**, all of which are **✓** (Completed).
- System1 Details (5):** A detailed view of the **System1** instrument, showing its status as **Templating Completed** and a progress bar for the run stages: Templating, Sequencing, Analysis, and Plugins.

- ① Up to five recent runs for your organization are listed in the left pane.
 - Click the row of a run of interest to view details for the run in the right pane.
 - Click the name of the run to leave the **Dashboard** and view the run results or sample results for the selected run.
- ② Instruments that are connected to Genexus™ Software are shown. A string of asterisks (*) for the run name indicates that the instrument is in use by another organization.
- ③ In the details pane on the right, place the pointer over the status for a run for your organization to view the progress of the run.
- ④ For the recent run that is selected in the left pane, each assay for the run is shown. Click the name of an assay to leave the **Dashboard** and open the results for the run and view assay metrics.
- ⑤ For the recent run that is selected in the left pane, each sample in the run is shown. Click a sample name to leave the **Dashboard** and open the sample results.

Genexus™ Software menus

Genexus™ Software provides menus to help you add, select, and manage samples, libraries, runs, and assays. You can also view and manage sample and run results, monitor Genexus™ Integrated Sequencer runs in progress, and manage software settings.



- ① **Dashboard**—View recent run history, and current purification or sequencing run status.
- ② **Samples**—Add new samples, import samples, prepare library batches, import library batches and manage attributes.
- ③ **Runs**—Plan a run starting from a sample, a nucleic acid sample, samples created from BAM or CRAM files, or from a library. Use the Purification Only run plan to purify samples on the Genexus™ Purification System and the Sample to Result runs plans for use with nucleic acid isolation on a Genexus™ Purification System, followed by sequencing on a Genexus™ Integrated Sequencer. View, edit, and manage runs.
- ④ **Results**—View sample results, run results, purification results, and verification results.
- ⑤ **Assays**—Manage, create, and import assays. Manage assays, presets, panels, and gene lists. Assay presets include annotation sets, filter chains, copy number baselines, sequence variant baselines, exon tile baselines, and report templates.
- ⑥ **Notifications**—Receive alerts and messages for password expiration, system critical service failures, system backup failures, available software updates, and full disk space.
- ⑦ **Settings**—Access audit records and run logs, configure network settings, manage backup settings, restore runs, link to Connect user accounts and Ion Reporter™ Software accounts, check for software updates, manage plugins, manage data archiving, disk space, users, and organizations. Field Service Engineers (FSEs) access verification templates during installation of the sequencer.
- ⑧ **Profile**—Access the software help system, manage and edit user profile settings, configure an SSH key (system administrator only), and sign out.

View notifications

Any user can view notifications that are sent for their review in Genexus™ Software. For example, a notification might be sent to one or more users about software updates, backup events, electronic signature notices, or file uploads, downloads, or imports.

1. In the menu bar, click  (**Notifications**).
The number of unread notifications is shown in the upper right of the notification button.
2. View the list of notifications.
3. To delete a notification, click  (**Remove**).
Notifications are retained until you manually delete notifications in the software.

Edit your user profile

After your user profile has been established, you can view your user profile information, change your password, change your email contact information, change your electronic signature type, and set your organization.

1. In the menu bar, click  **(Profile)** ▶ **My Profile**.
2. In the **Profile** screen, view the profile information.
3. Click **...** (**More Options**), then select an option to edit your user profile.

To	Do this
Change your password	a. Click Change Password. b. In the Change Password dialog box, enter the current password and a new password. c. Confirm the password, then click Change.
Change your email address	a. Click Edit Email. b. In the Set Email dialog box, enter a new email address, and a password, then click Save.
Edit your security questions and answers	a. Click Edit Security questions and answers. b. In the Edit Security questions and answers dialog box, select a security question from the list, then enter an answer. c. Enter the password for the account that is associated with the profile. d. Click Submit.

The changes are applied immediately. After the password is changed, you must sign in with the new password.

Access the End User License Agreement (EULA)

A link to the End User License Agreement (EULA) is located at the bottom of the screen.

1. Scroll to the bottom of the screen.
2. Click the **EULA** link to open a PDF file of the EULA.

Contact Support

You can access Genexus™ Software technical support from the software.

1. Scroll to the bottom of the screen.
2. Click **Support** to open <https://www.thermofisher.com/contactus>, where you can request help and technical support.

3

Manage samples and library batches

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Before you plan a run in Genexus™ Software, you must first enter sample information in the software to assign sample names and provide other information.

About samples and library batches

In Genexus™ Software, *sample* refers to the information in the software that represents data about a physical sample or samples that will be sequenced, or samples that are created after sequencing from BAM or CRAM files.

Sample type	Description
Sequencing sample	The data about a specimen sample that requires nucleic acid isolation on a connected Genexus™ Purification System before sequencing, or data about the purified nucleic acid that is ready for library preparation, sequencing, and analysis on a Genexus™ Integrated Sequencer. These samples can be analyzed in Genexus™ Software with a Nucleic Acid to Result , Sample to Result , or Library to Result run plan.
BAM or CRAM sample	Sample data in the software that is created from a BAM file or a CRAM file that contains sample reads. These sample files are either generated from a previous run on the same sequencer, or from BAM or CRAM files generated on a different sequencer then uploaded to Genexus™ Software. In the software, the file that contains this data is called a <i>sequence file</i>. BAM samples are identified in the list of samples with BAM after the sample name and CRAM samples are identified with CRAM after the sample name. These samples can be analyzed by the software with a BAM/CRAM to Result run plan.

Before you can plan a run to sequence or analyze a sample, you must enter the information, or attributes, that characterizes each sample. For example, each sample must have a unique name and be assigned to an application category.

A library batch is a group of samples from a previous sequencing run that need to be resequenced. A library batch can also be samples that are prepared with a manual or automated library preparation protocol. You can use a **Library to Result** run for these samples. You can create library batches from existing samples. When you prepare a library batch, you enter information about the barcode adapters that were used to prepare the libraries. After you create a library batch, you can plan a **Library to Result** run to sequence and analyze the samples that are contained in the library batch.

No-template and positive controls

You can add a no-template control (NTC) or a positive control when you assign samples in run planning, or when you create a library batch in Genexus™ Software. The configuration of each control is based on the assay that is used. For more information, see the user guide for the panel that you use.

View a sample summary

Role—System administrator, organization administrator and manager, operator.

You can view a summary of the details for a sample. The sample summary lists sample attributes, such as gender and the date that the sample was created in the software. For samples added to the software from BAM/CRAM files, any associated sequencing files are also shown.

Notes for a sample are not shown in the sample summary pane. For more information, see “View notes or add a note to a sample” on page 68.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Sample Name** column, click the sample name of interest.
Details for the sample are shown in the sample summary pane.
The sample attributes that are shown in the sample summary pane are based on the application category for the sample. For more information about sample attributes, see “System-installed sample attributes” on page 583.
3. (Optional) Click ... **(More Options)** to perform more actions, such as copy, edit, assign tags, add notes, or audit the sample. The options that are available depend on whether the sample is assigned to a run plan.
4. When you finish viewing the sample summary, click another sample name to view details for another sample, or click **✕ (Close)** to return to the **Manage Samples** screen.

The actions for each sample are also available in the **Manage Samples** screen when you place the pointer over the row of a sample.

View library batches

Role—System administrator, organization administrator and manager, operator.

You can view library batch information in Genexus™ Software.

You can filter, search, and sort the list of libraries batches. For more information, see “Find library batches” on page 81.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, use the following options to view library batches.

Option	Description
View more library batch information, such as the list of samples included in the library batch.	Click a Library Batch ID in the library batches list. Alternatively, in the Total Samples column, click the number of samples in the library batch. In the pane that shows details for the library batch, you can click ... (More Options) to perform more actions, such as edit, audit, or add notes to the library batch.
View all library batches.	In Filter Library Batches By , select All .
View libraries that are not in a run plan.	In Filter Library Batches By , select To Be Planned .
View multiple pages of libraries.	Click the page numbers and arrows at the bottom of the screen.
View notes for a library batch.	Place the pointer over the row of a library batch of interest, then click Notes .

Enter or import samples in Genexus™ Software

You can add samples in Genexus™ Software for use in purification on a connected Genexus™ Purification System before sequencing, or for library preparation, sequencing, and analysis on a Genexus™ Integrated Sequencer.

Add these samples in the **Create Sample** screen in the following ways.

- Use the **Single** tab to individually enter information for each sample into the software.
- Use the **Multiple (.xls, .csv, .tsv)** tab to import a file that defines sample attribute information for multiple sequencing samples. A file template is provided in the software. For more information, see “Download a template file for sample creation” on page 52. You can also individually enter and edit information for the imported samples in this tab.

Sample data in the software can be also created from a BAM file or a CRAM file that contains sample reads.

- To add these samples, use the **From Sequence Files (.bam, .cram)** tab to do one of the following.
 - Individually enter sample attribute information into the software, and then associate the sample information with an imported sequence file that contains sample reads in a BAM or CRAM format. For more information, see “Create BAM and CRAM samples” on page 48.
 - Or, upload a file that defines sample attribute information, and then associate that sample information with the BAM or CRAM sequence file. For more information, see “Create BAM and CRAM samples from a sample definition file” on page 50.

If you have both BAM and CRAM sample files, each single sample must contain files in either only BAM format, or only CRAM format. If you have a combination of file types, convert all files to the preferred file type, then import the files successfully. You can convert files to BAM or CRAM using **Samtools**, a set of utilities to interact with and perform post-processing of short DNA sequence read alignments in SAM (Sequence Alignment/Map), BAM, and CRAM file formats. For more information, see <https://www.htslib.org/doc/samtools.html>.

Create a single sample

Role—System administrator, organization administrator and manager, operator.

Note: Genexus™ Software encrypts data to protect the information that you enter for a sample.

Administrators, managers and operators can individually enter information for a sample into Genexus™ Software for purification of samples with a connected Genexus™ Purification System before sequencing, or for library preparation, sequencing, and analysis of samples on a Genexus™ Integrated Sequencer.

If you have many samples to add, see “Create multiple samples” on page 45.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Samples / Create Sample** screen, click the **Single** tab, then enter a name for the sample. The name of the sample cannot be *NTC*, *Control Sample*, or *Positive Control*.
3. In **Application Category**, do one of the following.
 - Select a system-installed or existing user-defined application category.

Only application categories that are associated with the organization for which you are signed in are available for selection.

- The fields for sample attributes change based on the **Application Category**. Attributes identified with a red asterisk (*) are required.

Note: It is recommended that user-defined attributes such as "Specimen Collection Site" and "Specimen Type" (for example, resection, CNB, and FNA) be added. For information about adding custom attributes, see “Create a user-defined sample attribute” on page 57.

- If the application category of interest is not listed, select **Other** to add an application category that you can use with the sample creation, then enter a name in the **Application Category** text box.

4. If you use the **Other** or **Generic Sequencing** application categories, select an application subcategory.
Subcategories are used to further define the sample attributes.
Sample attributes are shown in the **Create Sample** screen, based on the selected application category and subcategory, if applicable.
5. Enter sample attribute information in the fields of the table. For information about system-installed system attributes, see “System-installed sample attributes” on page 583.
6. Click **Save**.

The new sample is listed in the **Manage Samples** screen and is available for use in the software.

Create multiple samples

Role—System administrator, organization administrator and manager, operator.

You can individually enter information for multiple samples into Genexus™ Software for purification of samples with a connected Genexus™ Purification System before sequencing, or for library preparation, sequencing, and analysis of samples on a Genexus™ Integrated Sequencer.

Alternatively, you can import a file that defines the sample attribute information for multiple samples. A template for the file is provided in the software. For more information, see “Create multiple samples with a sample definition file” on page 46.

Note: Genexus™ Software encrypts data to protect the information that you enter for a sample.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Samples / Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, do one of the following.
 - Select a system-installed or existing user-defined application category.

Only application categories that are associated with the organization for which you are signed in are available for selection.

 - If the application category of interest is not listed, select **Other** to add an application category that you can use with the sample creation, then enter a name in the **Application Category** text box.
4. If you use the **Other** or **Generic Sequencing** application categories, select an application subcategory.
Subcategories are used to further define the sample attributes.
Sample attributes are shown in the **Create Sample** screen, based on the selected application category and subcategory, if applicable.
5. (Optional) Click **Columns** to show or hide sample attributes in the **Multiple (.xls, .csv, .tsv)** tab.
 - Select the checkbox next to each attribute to show in the table.
 - Deselect the checkbox next to each attribute to hide in the table.

6. Enter sample attribute information in fields for the first row in the table. For information about system-installed system attributes, see “System-installed sample attributes” on page 583.
The name of the samples that you create in the software cannot be **NTC**, **Control Sample**, or **Positive Control**.
7. Click **Add Row**, then select the row and enter the information in the new row for the new sample.
8. Repeat step 7 for each new sample.
9. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column heading row.

IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you create.

10. Click **Save**.

The new samples are listed in the **Manage Samples** screen and are available for use in the software. Samples are listed in the sample plate numerically and alphabetically. If you add 01, 02, 03, and so on as sample name prefixes according to the order you need, samples are automatically arranged in the desired order in the sample plate.

Create multiple samples with a sample definition file

Role—System administrator, organization administrator and manager, operator.

You can create multiple samples in Genexus™ Software for purification of samples with a connected Genexus™ Purification System before sequencing, or for library preparation, sequencing, and analysis of samples on a Genexus™ Integrated Sequencer.

Note: Genexus™ Software encrypts data to protect the information that you enter for a sample.

When you want to create multiple samples in Genexus™ Software, you can create a sample definition file that contains the information for a group of samples, and then import the file. You can import sample definition files in TSV, XLS, XLSX, or CSV file formats. You can use a template file to create a sample definition file in Excel™.

You must create user-defined attributes before importing sample and run information for the attributes to be propagated through to output files. All attributes that are included in the file that you use to import samples must be either system-installed attributes or user-defined attributes that exist in the software. Other file content is not transferred with the sample.

The sample definition file can be exported from an external software such as a LIMS system. Before you import a file that originates in external software, you must map all sample attribute names in the file that are named differently in the external software from the attribute used in Genexus™ Software. For more information, see “Map sample attributes” on page 61.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab to import sample data files in TSV, XLS, XLSX, or CSV file formats.

3. In **Application Category**, do one of the following.

- Select a system-installed or existing user-defined application category.

Only application categories that are associated with the organization for which you are signed in are available for selection.

- If the application category of interest is not listed, select **Other** to add an application category that you can use with the multiple sample creation, then enter a name in the **Application Category** text box.

4. If you use the **Other** or **Generic Sequencing** application categories, select an application subcategory.

Subcategories are used to further define the sample attributes.

Sample attributes are shown in the **Create Sample** screen, based on the selected application category and subcategory, if applicable.

5. Import a sample definition file in one of the following ways.

Download a template file to which you add information for the samples.	- Click Download Template. For more information, see “Download a template file for sample creation” on page 52. - Enter sample information into the file to edit the file. - Click Upload Sample Definition to upload the edited template file.
Use a sample definition file that is on your computer.	- Click Upload Sample Definition. - Navigate to the file on your computer. - Select the file and click Enter.

The data contained in the file populates the samples table in the screen.

Note: The name of the samples that you create in the software cannot be **NTC**, **Control Sample**, or **Positive Control**.

6. (Optional) In the **Multiple (.xls, .csv, .tsv)** tab, edit the sample table and data.

Option	Description
Remove an attribute column.	Click Columns , then deselect the column name.
Add an attribute column to the table.	Click Columns , then select the column name.
Edit sample data.	Click the sample row of interest, then edit the text field or dropdown list for the sample.
Add more samples to the table.	Click Add Row , then enter the sample data.
Remove a sample from the table.	Select the checkbox for the row, then click Remove Row .
View information about an error for sample data.	Place the pointer over  (alert).

7. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column heading row.

IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you intend to create.

8. Click **Save**.

The new samples are listed in the **Manage Samples** screen and are available for use in the software.

Create BAM and CRAM samples

Role—System administrator, organization administrator and manager, operator.

Sample data in the software can be created from a BAM file or a CRAM file that contains sample reads. The file is called a sequence file in Genexus™ Software.

BAM and CRAM sample files are generated in the following ways.

- From a previous run on the same sequencer.
- From BAM or CRAM files that are generated on a sequencer and uploaded to the software.

BAM and CRAM sample files can be analyzed by the software with a **BAM/CRAM to Result** run plan.

There are two ways you can create new BAM or CRAM samples in Genexus™ Software.

- You can individually enter sample attribute information into the software, and then associate the sample information with a sequence file.
- Or you can upload a file that defines sample attribute information, and then associate that sample information with a sequence file.

Use this procedure to individually enter sample information for each BAM or CRAM sample into the software. For information about how to use a file to enter sample information for BAM/CRAM samples, see instead “Create BAM and CRAM samples from a sample definition file” on page 50.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Samples / Create Sample** screen, click the **From Sequence Files (.bam, .cram)** tab.
3. In **Application Category**, do one of the following.
 - Select a system-installed or existing user-defined application category.

Only application categories that are associated with the organization for which you are signed in are available for selection.

- If the application category of interest is not listed, select **Other** to add an application category that you can use with the multiple sample creation, then enter a name in the **Application Category** text box.
4. If you use the **Other** or **Generic Sequencing** application categories, select an application subcategory.
Subcategories are used to further define the sample attributes.
Sample attributes are shown in the **Create Sample** screen, based on the selected application category and subcategory, if applicable.

5. Enter sample attribute information in fields for the first row in the table. For information about system-installed system attributes, see “System-installed sample attributes” on page 583.

Note: The name of the samples that you create in the software cannot be *NTC*, *Control Sample*, or *Positive Control*.

- a. Click **Add Row**, then select the row and enter the information in the new row for the new sample.
- b. Repeat step 7 for each new sample.
- c. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column heading row.

IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you create.

6. (Optional) To create TNA samples, click **Columns** then add the **TNA BAM/CRAM** column then select the **TNA BAM/CRAM** column.
 - Select the checkbox next to each attribute to show in the table.
 - Deselect the checkbox next to each attribute to hide in the table.
7. Click **Browse**, then use one of the following options to add the sequence file to the dropdown lists in the software.

Note: Sequence files that are used to define samples must be in either BAM or CRAM file format. When you upload multiple sample files, you can select a combination of files in both BAM or CRAM formats. However, when you define the individual samples, you must use either BAM format only or CRAM format.

Option	Description
Upload the file from the instrument.	Click  Server , then navigate to the file on the Genexus™ Integrated Sequencer. Select the sequencing files from the folders that are shown for the organizations of which you are a member and click Select .
Find the file on the computer that is used to access Genexus™ Software.	a. Click  My Computer, then in the Browse dialog box, navigate to the file on your computer, and then click Select. b. Wait for the file upload to complete. c. A popup message shows the progress of the file upload and prompts you to click Save. When the message goes away, go to step 5.

The sequence file is available for selection in the dropdown menus for **DNA BAM/CRAM**, **RNA BAM/CRAM**, and **TNA BAM/CRAM** columns.

8. Select the sequence file or files for the samples from the dropdown menus in the **DNA BAM/CRAM**, **RNA BAM/CRAM**, and **TNA BAM/CRAM** columns.
9. Click **Save**.
The new samples are listed in the **Manage Samples** screen and are available for use in the software.

Create BAM and CRAM samples from a sample definition file

Role—System administrator, organization administrator and manager, operator.

You can upload a file to define the sample attributes for multiple BAM or CRAM samples to create samples in Genexus™ Software. As part of this procedure, you associate that sample information with a sequence file.

To create samples from a BAM/CRAM file that is part of sequencing run, you must first download the BAM/CRAM file from the sample results screen, and then upload the BAM/CRAM file when you create new samples, then define the samples. During this process, you can enter the same or different values for the sample attributes that are associated with the file.

If the sequence file is not on the Genexus™ Integrated Sequencer, you can upload a file from the computer that is used to access Genexus™ Software. You can upload the file directly from the computer or from a USB drive connected to the computer.

Note: Sequencing files that are used to define samples must be in either BAM or CRAM file format. When you upload multiple sample files, you can select a combination of files both BAM or CRAM formats. However, when you define the individual samples, you must use either BAM format only or CRAM format.

IMPORTANT! Do not use processed BAM/CRAM files or consensus BAM/CRAM files. Use only the mapped or unmapped BAM files that are created during a sequencing run in the basecalling or alignment step that can be downloaded from the sample results screen. For more information, see “Download results files for multiple sample results” on page 183 and “Download results files for a sample result” on page 183.

BAM and CRAM files can be analyzed in the software if you use a **BAM/CRAM to Result** run plan. For more information, see “Plan a BAM/CRAM to Result run” on page 106.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Create Sample** screen, click the **From Sequence Files (.bam, .cram)** tab.
3. In **Application Category**, do one of the following.
 - Select a system-installed or existing user-defined application category.

Only application categories that are associated with the organization for which you are signed in are available for selection.

- If the application category of interest is not listed, select **Other** to add an application category that you can use with the multiple sample creation, then enter a name in the **Application Category** text box.

4. If you use the **Other** or **Generic Sequencing** application categories, select an application subcategory.
Subcategories are used to further define the sample attributes.
Sample attributes are shown in the **Create Sample** screen, based on the selected application category and subcategory, if applicable.
5. (Optional) Click **Columns** to show or hide sample attributes in the **From Sequence Files (.bam, .cram)** tab.
6. (Optional) To create TNA samples, click **Columns** then add the **TNA BAM/CRAM** column then select the **TNA BAM/CRAM** column.
 - Select the checkbox next to each attribute to show in the table.
 - Deselect the checkbox next to each attribute to hide in the table.
7. Click **Browse**, then use one of the following options to add each sequencing file to the dropdown lists in the software.

Option	Description
Upload the file from the instrument.	Click  Server , then navigate to the file on the Genexus™ Integrated Sequencer. Select the sequencing file from the folders that are shown for the organizations of which you are a member and click Select .
Find the file on the computer that is used to access Genexus™ Software.	a. Click  My Computer, then in the Browse dialog box, navigate to the file on the computer, and then click Select. b. Wait for the file upload to complete. c. A popup message shows the progress of the file upload and prompts you to click Save. When the message goes away, proceed to next step.

A sample file name is shown in each dropdown list in the **DNA BAM/CRAM File**, **RNA BAM/CRAM File**, and **TNA BAM/CRAM File** columns.

8. Select the sequence file or files for the samples from the dropdown list in the **DNA BAM/CRAM**, **RNA BAM/CRAM**, and **TNA BAM/CRAM** columns. For multiple file uploads, select only one file format type for this step.
9. Import a sample definition file in one of the following ways.

Download a template file to which you add information for the samples.	a. Click Download Template. For more information, see “Download a template file for sample creation” on page 52. b. Enter sample information into the file to edit the file. c. Click Upload Sample Definition to upload the edited template file.
Use a sample definition file that is on your computer.	a. Click Upload Sample Definition. b. Navigate to the file on your computer. c. Select the file and click Enter.

The data contained in the file populates the samples table in the screen.

Note: The name of the samples that you create in the software cannot be **NTC**, **Control Sample**, or **Positive Control**.

10. (Optional) In the **Multiple (.xls, .csv, .tsv)** tab, edit the sample table and data.

Option	Description
Remove an attribute column.	Click Columns , then deselect the column name.
Add an attribute column to the table.	Click Columns , then select the column name.
Edit sample data.	Click the sample row of interest, then edit the text field or dropdown list for the sample.
Add more samples to the table.	Click Add Row , then enter the sample data.
Remove a sample from the table.	Select the checkbox for the row, then click Remove Row .
View information about an errors for sample data.	Place the pointer over  (alert).

11. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column heading row.

IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you intend to create.

12. Click **Save**.

The new samples are listed in the **Manage Samples** screen, are denoted with **BAM** or **CRAM** after the sample name, and are available to assign to a **BAM/CRAM to Result** run plan.

Download a template file for sample creation

Role—System administrator, organization administrator and manager, operator.

You can download a template file, add sample information to it, then use the file to import the sample data for the following types of samples Genexus™ Software.

- Samples for use in purification on a connected Genexus™ Purification System before sequencing, or for library preparation, sequencing, and analysis on a Genexus™ Integrated Sequencer.
- Samples in the software that were created from sequence files in BAM or CRAM format.

Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional sample attributes, which are the column headings in the **Sample Details** tab. Use the **Sample Details** tab to enter sample information.

Template files contain both the system-installed and the user-defined sample attributes as column headings.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab, or **From Sequence Files (.bam, .cram)**.

3. In **Application Category**, do one of the following.

- Select a system-installed or existing user-defined application category.

Only application categories that are associated with the organization for which you are signed in are available for selection.

- If the application category of interest is not listed, select **Other** to add an application category that you can use with the multiple sample creation, then enter a name in the **Application Category** text box.

Sample attributes are shown in the **Create Sample** screen, based on the selected application category.

4. If you use the **Other** or **Generic Sequencing** application categories, select an application subcategory.

Subcategories are used to further define the sample attributes.

Sample attributes are shown in the **Create Sample** screen, based on the selected application category and subcategory, if applicable.

5. Click **Download Template** to download the Excel™ template file.

System-installed and user-defined sample attributes that are associated with the application category that is used by the sample are included as column headings in the file.

6. Click **Save** to your computer.

You can now import the file to Genexus™ Software. For more information, see “Create multiple samples with a sample definition file” on page 46 and “Create BAM and CRAM samples from a sample definition file” on page 50.

Enter or import a library batch in Genexus™ Software

A library batch is a group of prepared libraries that are sequenced in the same **Library to Result** run.

If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Software from samples. You can prepare a library batch manually or import library batch information from a template file. For more information see, “Enter or import samples in Genexus™ Software” on page 43.

Prepare a library batch

Role—System administrator, organization administrator and manager, operator.

A library batch is a group of prepared libraries that are sequenced in the same **Library to Result** run. Before you plan a run, you must first create a library batch in Genexus™ Software from samples that you have added in the software. For more information, see “Enter or import samples in Genexus™ Software” on page 43.

Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Software to prepare the library batch, and verify that the barcode scanner is compatible with the computer.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** list, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.

Note: If the library batch is being created in the same sequencer where the samples were processed initially, in the **Filter Samples by** dropdown list, apply the **All** filter to be able to select the previously processed samples.

For more information about how to filter the list of samples, see “Find samples” on page 74.

3. Select samples in the list by clicking the checkbox to the left of each sample, then click **+ Prepare Library Batch**.
4. In the **Create Library batch** screen, select the assay that you want to use in the run plan from the **Select Assay** list.

The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library preparation type, the **Nucleic Acid Type** automatically fills with the nucleic acid type that is specified by the assay you selected. For example, **DNA, RNA, DNA+RNA, or TNA**.

For assays that include more than one library preparation type, select an item from the **Nucleic Acid Type** list, such as **DNA, RNA, DNA+RNA, or TNA** for the library batch.

5. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.
Library Batch IDs can contain only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-). Required fields are indicated with a red asterisk (*).
6. (Optional) Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.
7. Select the **Include NTC** checkbox to add no-template control sample processing and reporting to the library batch.
8. Select the barcode ID of the adapter used to prepare each library. If appropriate, swap the default barcodes between DNA, RNA, and Fusions by clicking the **Swap Barcodes** swap image.

For example, click **DNA ↔ Fusions** to swap barcodes.

Each library in a library batch must have a different barcode ID. When preparing the physical libraries, best practice is to swap barcodes between DNA and RNA libraries in consecutive sequencing runs to prevent carryover contamination. The barcodes that are listed in the **DNA**

Barcode or **RNA Barcode** list belong to the barcode set that was selected when the assay was created.

IMPORTANT! Ensure that the barcodes that you used to create the libraries match the barcodes that you enter in the **Create Library batch** screen.

9. (Optional) Enter the **Input Quantity=** of each **DNA, RNA, DNA+RNA, or TNA** library.
10. Use the connected barcode scanner to scan the barcode from the control kit into the fields in the **DNA Kit Barcodes, RNA Kit Barcodes, or TNA Kit Barcodes** columns for the positive controls and the no-template controls, and for each sample, if applicable.
If needed, you can enter the barcodes manually.
11. Click **Submit** to save and submit your selections.
The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

Import a file to prepare a library batch

Role—System administrator, organization administrator and manager, operator.

You can import library batch information in the form of an XLS or XLSX file. The import file must include all required library and kit information.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, click  **Import Library Batch**.
3. In the **Import Library Batch** dialog box, click **Click here** to select an assay for which the libraries are prepared, and to download an example file for import.
If the assay includes more than one library preparation type, a library preparation type dropdown list appears.
4. Select the library preparation type, such as **DNA, RNA, DNA+RNA, or TNA** for the library batch.
5. Select an assay from the list, then click **Download**.
The assay name is automatically populated in the Excel™ template file that downloads to your computer.
6. In the template file, enter or confirm the library batch information.
Use the connected barcode scanner to scan the barcode from the library kit, panel kit, control kit, and extraction kit into the fields in the DNA Kit Barcodes, RNA Kit Barcodes, or TNA Kit Barcodes columns for the positive controls and the no-template controls, and for each sample, if applicable.

If needed, you can enter the barcodes manually.

Template item	Description
Reagents tab	
Assay Name	Auto-populated when assay is selected in step 4 (required).
Extraction Method Type	Auto-populated when assay and library prep type, if applicable, is selected in step 5 (required).
Library Batch ID	Must be 50 or fewer alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) characters (required).
Library Kit Barcode	For example, Genexus™ Library Strips 1 and 2-AS barcode (optional).
Panel Kit Barcode	For example, Oncomine™ Comprehensive Assay v3 DNA GX barcode (optional).
Libraries tab	
Sample Name	Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required). To Include a no-template control, add a row with NTC for the Sample Name .
Barcode	Barcodes used for each sample and control prepared library (required).
Nucleic Acid Type	DNA, RNA, or TNA (required).
Input Quantity	Library input quantity (optional).
Control Kit Barcode	The barcode for the Positive Control and NTC kits used for Sample to Result and Nucleic Acid to Result runs (optional).
Extraction Kit Barcode	The barcode for the kit used for nucleic acid extraction of a sample (optional).

IMPORTANT! For DNA and Fusions assays, the DNA library and RNA libraries must be listed in sequential order per pool for each sample. For example, for a 1-pool DNA and Fusions assay, order should be DNA, RNA for sample 1, DNA, RNA for sample 2. For a 2-pool DNA and Fusions assay, library order should be DNA, RNA (pool 1), DNA, RNA (pool 2) for sample1, then DNA, RNA (pool 1), DNA, RNA (pool 2) for sample 2.

7. Save the file.
8. In the **Import Library Batch** dialog box, click **Select Library batch file**, navigate to the saved file, then select it.
9. Click **Upload**.
A progress bar followed by an import report displays. If the import process fails, click **View errors** to review the reason for failure. For more troubleshooting support, see “Library batch import fails” on page 588.

Sample attributes

Role—System administrator, organization administrator and manager.

Samples contain attributes that are relevant to genetic research. For example, attributes can include sample information such as gender, cancer type, the nucleic acid type such as DNA, RNA, or cfDNA, or a sample type such as blood, urine, or swab.

In Genexus™ Software, administrators and managers can add user-defined attributes and add or change information for those attributes. For more information, see “Create a user-defined sample attribute” on page 57 and “Edit a user-defined sample attribute” on page 59.

User-defined and system-installed sample attributes can be viewed in the **Attributes** table.

For more information, see “System-installed sample attributes” on page 583.

Create a user-defined sample attribute

Role—System administrator, organization administrator and manager.

Administrators and managers can create user-defined sample attributes. Sample attributes, such as the date of collection and **Application Category**, characterize and define a purification sample, nucleic acid sample, or a sample from a BAM or CRAM file. A sample attribute can be required, in which case you are required to enter the attribute information for each new sample. User-defined sample attributes can be used to further characterize samples.

Genexus™ Software supports following types of sample data.

- **Text**—Accepted values: general text.
- **Date**—Accepted values: provides a calendar tool to select a date.
- **Integer**—Accepted values: integer numbers. Examples: 0, 9, -44.
- **Decimal**—Accepted values: decimal numbers Examples: 0.1, 9.0, -44.3.
- **List**—Creates a list of options that you define with preset terms.
- **Boolean**—Creates a list of these two choices: True, False.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** screen, click **+ Add New Attribute**.
3. In the **Add New Attribute** dialog box, in **Attribute Name**, enter a name for the attribute.
Attribute names are limited to 50 or fewer alphanumeric characters (0–9 and Aa–Zz), period (.), underscore (_), or hyphen (-).
4. In **Application Category**, select an applicable application category or application categories to add application categories that are appropriate to the new attribute.
The application category is associated with the sample attribute and is shown in the list of available attributes during sample definition, when users select an application category.

IMPORTANT! You cannot create user-defined sample attributes for the **Generic Sequencing** application category. Sample attributes shown for the **Generic Sequencing** application category are based on the subcategory selection.

5. In **Data Type**, specify the data type to create.
6. Click the toggle switch to designate the attribute as **Required**. When an attribute is required, users must enter a value for the attribute when defining samples.
7. Click the toggle switch to designate the attribute as **Unique**, if needed.
When enabled, the **Unique** setting allows you to require a unique attribute value for samples. For example, you can create a unique attribute called **Sample number**. Then, when each new sample is created, you must enter a unique number for each sample in the user-defined **Sample number** attribute field.
8. For **List** and **Boolean** data types, complete the following options.

Data type	Steps
List	a. In Vocabulary List, enter a list of values separated by commas, then click Add. The first option that you enter is designated as the default option. b. Change the default option, if needed. Deselect Use default if you want the new attribute to be shown without a default preset. Select a different option to change the value that is shown as the default option. Click  (Delete) to remove an attribute list value.
Boolean	Select True or False to designate one option as the default value for the attribute.

9. Click **Save**.

The new sample attribute is added in the **Manage Attributes** screen and is available for use in the software.

You can now add information for the user-defined sample attribute to when you create new samples for the application category that is associated with the sample attribute. In addition, you can enter information for the attribute to existing samples that were created before the user-defined attribute was created. For more information, see “Enter or import samples in Genexus™ Software” on page 43 and “Edit a sample” on page 65.

Example of sample attribute creation

You can create a **List** data type sample attribute that has two values and shows how to set, or remove, a default value for the sample attribute. All samples that are added to Genexus™ Software after the sample attribute is created, and have the same application category, are associated with the default value, unless the default value is changed when the sample is created.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** screen, click **+ Add New Attribute**.

3. In the **Add New Attribute** dialog box, in **Attribute Name**, enter **Grade** as the name for the attribute.

Attribute names are limited to 50 or fewer alphanumeric characters (0–9 and Aa–Zz), period (.), underscore (_), or hyphen (-).

- a. In **Application Category**, select **Cancer (Germline)**.

The application category is associated with the sample attribute and is shown in the list of available attributes during sample definition, when users select an application category.

IMPORTANT! You cannot create user-defined sample attributes for the **Generic Sequencing** application category. Sample attributes shown for the **Generic Sequencing** application category are based on the subcategory selection.

- b. In **Data Type**, select **List**.

- c. In **Vocabulary List**, enter **High, Low**, then click **Add**.

The two options are shown below the **Vocabulary List** field. The first option in the list is selected as the default option in the **Create Sample** screen. When you create a new cancer sample with the attribute in this example, **High** is shown as the default **Grade** value. When you add the sample attribute to a sample, you can select **High** from the **Grade** dropdown list to change the value.

4. (Optional) Change or remove the default value for the sample attribute.

Option	Description
Change the default option.	with the Use default checkbox selected, select the value to show as the default. In this example, you can select Low .
Remove the default option.	Deselect the Use default checkbox.

The default value, if selected, is shown in the If the **Use default** checkbox is not selected, a default value is not selected in the software when the sample attribute is viewed.

5. Click **Save**.

After the new attribute is saved, it is available for use when samples are created in the software.

Edit a user-defined sample attribute

Role—System administrator, organization administrator and manager.

Administrators and managers can edit user-defined sample attributes. Some sample attribute settings, such the **Attribute Name**, **Data Type**, and whether or not the attribute is required, cannot be edited.

If desired, you can edit the user-defined attributes to associate the application categories with the appropriate applications.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** table, place the pointer over the row of a user-defined sample attribute that you want to change, then click **Edit**.

3. In the **Edit Attribute** dialog box:

Option	Description
Change the application category that is associated with the sample attribute.	In Application Category , select a different application category.
Remove the application category that is associated with the sample attribute.	In Application Category , deselect the checkbox for application category.
Add to the list of comma-delimited list of values for List type sample attributes.	a. In the Vocabulary List text box, enter the new values, then click Add. b. Select Use default for a default value to be selected for the sample attribute. Deselect Use default to have no default selected for the sample.
Change the default value for Boolean type sample attributes.	a. In the Data type text box, verify that Boolean, is selected. b. Change the default option, if needed.

4. Click **Save**.

Edit Cancer Type and Cancer Stage sample attributes

Role—System administrator, organization administrator and manager.

Administrators and managers can edit the **Cancer Type** or **Cancer Stage** sample attributes for a sample in Genexus™ Software.

IMPORTANT! When the only change that you make to a sample attribute is a change to either the **Cancer Type** and **Cancer Stage** sample attributes, or to both of the attributes, you must select one sample result for which the edited sample information is used. Any other sample results in which the sample is included are marked as **Obsolete**. If you need to review results for an obsolete sample result, you must reanalyze the sample. For more information, see “Reanalysis” on page 218.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to edit, then click **Edit**.
3. In the **Edit Sample** screen, change the **Cancer Type** or **Cancer Stage** sample attributes, as needed.
The **Amend result** dialog box is shown.
4. In the **Amend result** dialog box, select a run name from the list of runs to which the edit will apply, then click **Confirm**.
5. Click **Save**.

The changes to the **Cancer Type** and **Cancer Stage** sample attributes are saved to the selected sample result. Other sample results in which the sample is included are marked as **Obsolete**.

Obsolete a user-defined sample attribute

Role—System administrator, organization administrator and manager.

Administrators and managers can obsolete a user-defined sample attribute so that the attribute is no longer available for use in the software.

When you obsolete a user-defined sample attribute, no change is made to the samples that already have an assigned value for the sample attribute. You cannot obsolete a system-installed sample attribute.

1. In the menu bar, click **Samples ► Manage Attributes**.
The **Manage Attributes** screen opens to the **Manage Attributes** tab.
2. Place the pointer over the row of the user-defined sample attribute that you want to obsolete, then click **Obsolete**.
3. In the confirmation box, click **Yes** to confirm.

The obsolete user-defined sample attribute is listed in the **Manage Attributes** screen but is no longer available for use when you create a sample.

Obsolete user-defined sample attributes can be reactivated.

Reactivate an obsolete sample attribute

Role—System administrator, organization administrator and manager.

Administrators and managers can reactivate an obsolete user-defined sample attribute.

1. In the menu bar, click **Samples ► Manage Attributes**.
The **Manage Attributes** screen opens to the **Manage Attributes** tab.
2. In the **Manage Attributes** table, place the pointer over the row of a user-defined sample attribute that you want to reactivate, then click **Reactivate**.
3. In the confirmation dialog box, click **Yes** to confirm.

The sample attribute is available to use when you create a sample.

Map sample attributes

Role—System administrator, organization administrator and manager.

Administrators and managers can import a file that you have exported from external LIMS software to add samples in Genexus™ Software. Each column heading in the file that you want to import to create samples in the software is a name for a sample attribute. Before you import samples with the file, an administrator or manager must map the names of sample attributes that are used by the LIMS software to the names of the sample attributes recognized by Genexus™ Software.

For example, in Genexus™ Software, the sample attribute for the name of a sample is **Sample Name**. In the file that you want to import, however, the same attribute can be called *Name*. You can map the sample attribute *Name* to **Sample Name** so that the software recognizes *Name* as the name of a sample.

If the attribute settings in the file contain vocabulary that is not recognized in the software, you can map the associated vocabulary so that you can seamlessly import the file to create samples. For more information, see “Map the name of a setting for a sample attribute” on page 62.

1. In the menu bar, click **Samples ► Manage Attributes**.
2. In the **Manage Attributes** screen, click the **Sample Attribute Mapping** tab.
If any attributes have been mapped, a list of sample attributes and any associated attribute names for all application categories is shown. Attributes identified with a red asterisk (*) are the attributes that are required when you create a sample.
3. Click  **Edit Mapping**.

Option	Description
Populate attribute name options from a file, then select the attribute names to add the names in the software.	a. Click Click here, then find and upload the file that you want to import. The attribute names are listed in the right pane. b. In Associated Attribute Name for the attribute of interest, select the attribute name from the list. For sample attributes that include a date, select the proper date format for the LIMS software from the list. c. Select the checkbox of an associated attribute name to map the name to an Attribute Name.
Enter an associated attribute name to add the name in the software.	a. In Associated Attribute Name for the attribute of interest, enter the attribute name used in the file that you want to import. For sample attributes that include a date, select the proper date format for the LIMS software from the dropdown list. b. Select the checkbox of an associated attribute name to map the name to an Attribute Name.
Remove an associated attribute name.	In Associated Attribute Name for the attribute of interest, in the tile for the name that you want to remove, click ✕ (Remove) .

4. Click **Save**.

The external attribute names are mapped to the attribute names used in Genexus™ Software and are listed as associated attribute names in the **Manage Attribute Mapping** list.

You can then import the LIMS software file to create samples with the sample data. For more information, see “Create multiple samples with a sample definition file” on page 46.

Map the name of a setting for a sample attribute

Role—System administrator, organization administrator and manager.

Administrators and managers can import a file that you have exported from external LIMS software to add samples in Genexus™ Software. Each column cell in the file to create samples in the software can contain alternate vocabulary for a sample attribute that are not recognized in the software. Before you import samples, an administrator or manager must map the vocabulary for the sample attributes that is used by the LIMS software to the names of the sample attributes recognized by Genexus™ Software.

For example, in Genexus™ Software, one of the sample attribute settings for **Sample Type** is **Blood (Whole)**. In the file that you want to import, however, the same attribute value can be called **whole blood**. You can map the sample attribute value, **whole blood**, to **Blood (Whole)** so that the software recognizes **whole blood** as **Blood (Whole)**.

1. In the menu bar, click **Samples ► Manage Attributes**.
2. In the **Manage Attributes** screen, click the **Sample Attribute Mapping** tab.
If any attributes have been mapped, a list of sample attributes and any associated attribute names for all application categories is shown. Attributes identified with a red asterisk (*) are the attributes that are required when you create a sample.
3. Click **Manage Vocabulary Mapping**.
By default, sample attributes that are a list type, such as **Sample Type** and **Cancer Stage**, and have vocabulary values defined in the software, are shown in the pane on the left. Any sample attributes that have associated vocabulary mapped to the attribute are also listed.
Click an attribute name in the left pane to view the vocabulary value and associated vocabulary value in the pane on the right.
4. Click  **Edit Mapping**.
5. In **Associated Vocabulary Value** for the attribute value of interest, enter the term of the attribute value used in the file that you want to import, then click **Save**.
To remove a term that you have added, in **Associated Vocabulary Value** for the attribute of interest, delete the text.
6. Click **Save**.

The name for the external attribute value is mapped to the attribute vocabulary value recognized in Genexus™ Software and is listed as an associated vocabulary value in the **Manage Vocabulary Mapping** list.

You can then import the LIMS software file to create samples with the sample data. For more information, see “Create multiple samples with a sample definition file” on page 46.

Review and download audit records for sample attribute mapping

Role—System administrator, organization administrator and manager.

Administrators and managers can review, export, or print the entire history of sample attribute mapping in Genexus™ Software. An audit trail lists the records for each sample mapping attribute to which changes are made. Each audit record in the list shows the name of each sample mapping attribute, and the original attribute settings and any new attribute settings.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples ► Manage Attributes**.
2. In the **Manage Attributes** screen, click the **Sample Attribute Mapping** tab.

3. Click **Audit**.

A list of time-stamped audit records that includes information for each action performed on sample attribute mappings is shown in the **Audit Details** screen.

4. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the sample attribute mapping, such as creation of mapped attributes, or edits, is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attribute mappings Manage Attributes screen.

Manage samples

You can find tools to create, find, copy, sort, edit, delete, export, and view the sample history in the **Samples / Manage Samples** screen.

Copy a sample

You can copy a sample and change the information for the sample, if needed, to create a new sample in Genexus™ Software.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to copy, then click **Copy**.
The **Copy Sample** screen opens. The sample information for the source sample is shown. The **Sample Name** for the new sample is the sample name of the sample that was copied, appended with **_Copy**.
3. In the **Copy Sample** screen, edit the sample information, and change the sample name, if needed.
4. Click **Save**.

The new sample is listed in the **Manage Samples** table and is available for use in the software.

Copy multiple samples

You can copy multiple samples at once in Genexus™ Software and change the information for the samples. To edit the samples at the same time, all samples must be associated with the same application category and, if applicable, subcategory.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
The actions that you can apply to multiple samples are shown at the top of the samples table.
3. Click  **Copy**.
The **Copy Sample** screen opens. The sample information for each source sample is shown. The **Sample Name** for each new sample is the sample name of the sample that was copied, appended with **_Copy**.
4. In the **Copy Sample** screen, edit the sample information, and change the sample names, if needed.
5. Click **Save**.

The new samples are listed in the **Manage Samples** table and are available for use in the software.

Edit a sample

Role—System administrator, organization administrator and manager.

Administrators and managers can edit the information that describes a sample in Genexus™ Software. When you edit a sample, you can change the application category, update sample attributes, and remove attributes that are not required.

To change the **Cancer Type** or **Cancer Stage** sample attributes, you must apply the change to one sample result for which the edited sample information is used. For more information, see “Edit Cancer Type and Cancer Stage sample attributes” on page 60.

You can reuse edited samples in another sequencing run. For example, you might want to edit a sample to reflect the information found in a final report after a run, then reuse the samples.

If you edit a sample after the sequencing run and the sample result is obsolete, you must reanalyze the sample to generate sample results and a new variant report for the sample. For more information, see “Reanalysis” on page 218.

For samples created from BAM or CRAM files, you can add or remove associated sequence files.

Samples can be edited at the same time under the following circumstances.

- The samples are not yet added to a run plan, or a sequencing run for the samples is not in progress.
- Any sequencing runs that include the samples are complete.

When you edit a sample that has a deprecated application category, the sample is assigned to the **Other** application category and the name of the application category is automatically filled with the deprecated value. For example, if you edit a sample that includes the **Allergic Disease** application category, **Other** is selected in the **Edit Sample** screen, and **Allergic Disease** is automatically filled in the editable text box as the application category.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to edit, then click **Edit**.
3. In the **Edit Sample** screen, change the sample information, as needed.

IMPORTANT! A sample created with an application category that was available only in a previous version of the software is assigned to the **Other** application category and subcategory. When you edit samples, **Application category** field shows the name for the deprecated application category. For example, if the sample has an application category of **Allergic Disease**, which is no longer available in the software, **Other** is shown in **Application category**, and the **Allergic Disease** field is automatically filled in the text box as the application category name.

4. (Optional) For samples created from BAM or CRAM files, do one of the following to change the BAM or CRAM files that are associated with the sample.
 - – Click  **Server** to upload a file from the sequencer, then navigate to the file.
 - – Or click  **My Computer** to find a file on the computer that is used to access Genexus™ Software. In the **Browse** dialog box, navigate to the file, and click **Save** when the file upload is complete.
 - Click **My Computer** to find and select a BAM or CRAM file on your computer.
5. Click **Save**.

Edit multiple samples

Role—System administrator, organization administrator and manager.

Administrators and managers can edit multiple samples at the same time in Genexus™ Software.

Samples can be edited at the same time under the following circumstances.

- The samples have the same sample application category.
- The samples are not yet added to a run plan, or a sequencing run for the samples is not in progress.
- Any sequencing runs that include the samples are complete.

You must reanalyze the samples to generate sample results and a new variant report for edited samples and other files associated with the samples. For more information, see “Reanalysis” on page 218.

If you edit the sample after results are generated, then the status for the sample results is changed to obsolete and the report and sample results that are associated with the samples are no longer available.

When you edit samples that have a deprecated application category, the samples are assigned to the **Other** application category. The value for the **Other** application category is automatically filled with the deprecated value. For example, if you edit a sample that includes the deprecated application category **Allergic Disease** application category, **Other** is selected in the Edit Sample screen, and **Allergic Disease** is automatically filled in the editable text box as the application category.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
The options that you can apply to multiple samples appear.
3. Click  **Edit**.
4. In the **Edit Sample** screen, modify the sample information, as needed.
 - a. (Optional) Change the sample name.
 - b. (Optional) In **Application Category**, select a system-installed or existing user-defined application category.
Only application categories that are associated with the organization for which you are signed in are available for selection.

IMPORTANT! A sample created with an application category that was available only in a previous version of the software is assigned to the **Other** application category and subcategory. When you edit samples, **Application category** field shows the name for the deprecated application category. For example, if the sample has an application category of **Allergic Disease**, which is no longer available in the software, **Other** is shown in **Application category**, and the **Allergic Disease** field is automatically filled in the text box as the application category name.

Sample attributes are shown in the **Create Sample** screen, based on the selected application category.

- c. (Optional) If the application category of interest is not listed, select **Other** to add an application category that you can use with the multiple sample creation. If you use the **Other** application category, enter a name in the **Application Category** text box.
- d. (Optional) Make changes to the sample attributes, as needed. For more information, see “System-installed sample attributes” on page 583.
- e. (Optional) If you edit BAM or CRAM samples, you can add or remove the associated sequence files if at least one file is associated with the sample.
 - To select a file that is not listed, click **Browse**, then find the file to associate with the sample.
 - Click  **Server** to upload a file from the sequencer, then navigate to the file.
 - Or click  **My Computer** to find a file on the computer that is used to access Genexus™ Software. In the **Browse** dialog box, navigate to the file, and click **Save** when the file upload is complete.

- f. (Optional) Select a file from the **DNA BAM/CRAM File**, **RNA BAM/CRAM File**, or **TNA BAM/CRAM File** dropdown lists.

5. Click **Save**.

The changes to the samples are saved and the edited samples are available for use in the software.

View notes or add a note to a sample

Role—System administrator, organization administrator and manager, operator.

You can view existing notes or add new notes to a sample in Genexus™ Software. Use notes to capture time-stamped written comments and observations for a sample. A sample can contain multiple notes from different users.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.

Action	Description
View existing notes for a sample.	In the Add Notes dialog box, notes that are associated with the sample are listed by user, date, and time.
Add a new note to a sample.	a. In the Add Notes dialog box, click + Add Note. b. Enter the note, then click Save. The note is added to the sample and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.

Delete a sample note

Role—System administrator, organization administrator and manager, operator.

You can delete notes for a sample in Genexus™ Software.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.
3. In the **Add Notes** dialog box, in the row of the note that you want to delete, click  **(Delete)**.

The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **✕ (Close)** to return to the **Manage Samples** screen.

Manage sample tags

A sample tag is a label that you add to a sample to help you group and track samples in Genexus™ Software. For example, you can assign a tag to each sample in a study or project so that you can easily find that group of samples in the software.

An administrator or a manager first creates a tag, then users can assign that tag to a single sample or a group of samples. You can assign multiple tags to each sample.

You can assign a tag when you create a sample or later add, edit, or remove the tags that are assigned to a sample. You can apply sample tags to multiple samples at the same time. Later, you can filter the table of samples with that tag to list only the samples that have the tag. The tags that you assign to samples are also shown in the list of sample results.

Tags are preserved with the sample when you archive a run that includes a sample with one or more assigned tags.

You can also add tags to sample results. For more information, see “Assign sample tags to a sample result” on page 123 and “Assign sample tags to multiple sample results” on page 123.

Create a sample tag

Role—System administrator, organization administrator and manager.

Administrators and managers can create sample tags in Genexus™ Software that can be used to label and group samples.

You can create nested tags, if desired. For example, if a large project is divided into sections, you can create a tag called **Master project** and a subordinate tag called **New variant**. Nested tags help to sort projects and categories within those projects. When you assign nested tags to a sample, all tags are listed for the sample. Later, when you search or filter for nested tags, the top-level tags are shown to help you group and track the samples.

1. In the menu bar, click **Samples ▶ Manage Samples**.
Alternatively, you can create a tag when you create a sample. For more information, see “Create a single sample” on page 44 and “Create multiple samples with a sample definition file” on page 46.
2. In the **Manage Samples** screen, click  **Manage Tags**.
The **Manage Tags** dialog box opens and shows the tags, if any, that have been created in the software.
3. Click  **Create Tag**.
4. In the **Create Tag** dialog box, enter a name for the tag.
5. (Optional) Create a subordinate tag. Select a tag from the **Parent Tag** list to nest the tag.
6. Click **Save**.

The tag is available to assign to samples. The tag or tags are shown in the **Tags** column in the sample results table. If the **Tags** column is not shown, click **Columns** and add the **Tags** column.

Assign sample tags

Role—System administrator, organization administrator and manager, operator.

You can assign tags to samples in Genexus™ Software to label and group samples.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample that you want to tag, then click **Assign Tags**.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample does not exist in the software, a manager or administrator can create a new sample tag.
4. Select one or more tags that you want to assign to the sample, then click **Save**.

The tag is assigned to the sample and is shown in the **Tags** column in the sample results table. If the **Tags** column is not shown, click **Columns** and add the **Tags** column.

Assign a sample tag to multiple samples

Role—System administrator, organization administrator and manager, operator.

You can assign tags to multiple samples at once in Genexus™ Software. Tags are shown in the sample results table and can help to label and group samples, and sort projects and categories within those projects.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, click the checkbox to the left of each sample of interest, then click  **Assign Tags** above the list of samples.
 **Assign Tags** is not shown until you select at least one sample.
The **Assign Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample does not exist in the software, an administrator or manager can click  **Create Tag** to create a new sample tag.
4. Select one or more tags that you want to assign to the samples, then click **Save**.

The tag is available to assign to samples. The tag or tags are shown in the **Tags** column in the sample results table. If the **Tags** column is not shown, click **Columns** and add the **Tags** column.

Add or remove the tags that are assigned to a sample

Role—System administrator, organization administrator and manager, operator.

You can add or remove the tags that are assigned to a sample. The tags that are assigned are shown in the **Tags** column in the sample results table in Genexus™ Software.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample that you want to add or remove tags, then click **Assign Tags**.
3. In the **Assign Tags** dialog box:
 - Select the checkbox next to each tag that you want to add to the sample.
 - Deselect the checkbox next to each tag that you want to remove from the sample.
4. Click **Save**.

Edit a sample tag

Role—System administrator, organization administrator and manager.

Administrators and managers can rename a tag and change the hierarchy of the tags that are shown in the sample results table in Genexus™ Software. When you edit a tag, the changes are applied to all samples to which the tag is assigned.

When you edit a sample and make a change to the tags associated with the sample, the changes you make are tracked in the software and can be reviewed. For more information, see “Review and download audit records for a sample” on page 79.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, click  **Manage Tags**.
The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click  **(Edit)**.
4. Change the name of the tag and parent tag assignment if needed, then click **Save**.
5. In the confirmation dialog box, click **Yes** to confirm.

All samples to which the edited tag is assigned are updated with the changes. If the name of the tag is changed, the new name is shown for all samples and sample results to which the tag is assigned. Any changes to the hierarchy of the tag are also applied to all samples and sample results that include the tag.

Delete a sample tag

Role—System administrator, organization administrator and manager, operator.

Administrators and managers can delete sample tags in Genexus™ Software. When a tag is deleted, the tag is also removed from any samples to which the tag is assigned. Additionally, when you delete the top-level tag of a nested tag, all tags in the nested hierarchy are deleted.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, click **Manage Tags**.
The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click  **Delete**.
4. In the **Confirm** message box, click **Yes** to confirm the changes.

The tag is no longer shown in the **Manage Tags** list and is removed from all samples and sample results.

Export samples

The **Export** function generates an XLS file that contains details about the selected sample. You might want to use the file to edit the samples, or to import sample information to create multiple sequencing samples that require nucleic acid isolation on another Genexus™ Integrated Sequencer.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to export. To select all samples, select the checkbox in the column heading row.

IMPORTANT! If you will import the sample file to another sequencer, select samples that are associated with only one application category.

3. Click  **Export** at the top of the samples table.
An XLS file is created that contains the details of the selected samples. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.
4. Open the XLS file in a software such as Excel™.

Cancer types in Oncology content packages

Administrators and managers can use a new or updated cancer type from an Oncology content package that is installed in Genexus™ Software.

Oncology content package updates can include new or updated cancer types, OncoPrint Variant Annotations rulesets, Variant Naming Services, and gene lists. For more information, see “Oncology content packages for use in assays” on page 263 and “Oncology content packages with gene lists” on page 348.

Before you use updated Oncology content updates, you need to download and install the update package. Updated Oncology content packages are available from apps.thermofisher.com.

- If a Thermo Fisher™ Connect Platform account has been configured, download and install software directly from thermofisher.com/connect. See “Download and install software updates from the App Store” on page 370
- Alternatively, if the instrument is not connected to the internet, use a USB drive to install and update the software. See “Format a USB drive for a software update” on page 372 and “Download and install software updates with a USB drive” on page 371.

After an Oncology package update is installed, you can use a new or updated cancer type sample attribute for the following procedures.

- You assign new cancer types to any samples if needed. For more information, see “Enter or import samples in Genexus™ Software” on page 43.
- You can edit a cancer type sample attribute. For more information, see “Edit Cancer Type and Cancer Stage sample attributes” on page 60.

Show or hide columns in the samples table

The samples table is organized into columns of sample attributes. You can determine which columns are shown or hidden in the samples table with column preferences. The changes you make to the columns are saved only for your user profile.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. Click **Columns**.
 - Select the checkbox next to each attribute you would like to show.
 - Deselect the checkbox next to each attribute you would like to hide in the table.
3. Scroll to the bottom of the list.
 - Click **Save** to save your new selections. Your selections persist when you reopen the table.
 - Click **Restore default** to restore the table to the default column settings.

Find samples

You can search, filter, and sort information to refine and narrow the samples that are listed in the **Samples / Manage Samples** screen in Genexus™ Software. The sort, search, and filter tools help you organize samples and find a samples of interest.

For information to search for a sample by a unique identifier, such as the sample name, see “Filter and search for samples” on page 74.

For information to filter samples by a common trait, see “Filter samples by sample type or preparation status” on page 75 and “Filter samples by creation date” on page 76.

Filter and search for samples

Role—System administrator, organization administrator and manager, operator.

Samples are listed in the **Samples / Manage Samples** screen in Genexus™ Software.

You can search for samples with keywords or text that are included in the sample attributes, such as sample name, tags, or application category.

You can also filter by sample type or preparation status, or filter by sample creation date or sample attribute value. For more information, see “Filter samples by sample type or preparation status” on page 75, “Filter samples by creation date” on page 76, and “Filter samples by sample attribute value” on page 77.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, click  (**Filter**) in the column heading for a sample attribute of interest, such as **Sample Name**, then do one or more of the following.
 - In the search field, enter 3 or more characters for the sample attribute.
 - Select a checkbox next to one or more sample attributes.
 - Click **Select All** to select all sample attributes.
 - Click **Clear** to close the filter dialog box.

In the column heading, the  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of samples is shown in the **Manage Samples** screen.

Click  **Clear Filters** at the top of the **Samples / Manage Samples** screen to remove all filters and view the full list of samples.

Filter samples by sample type or preparation status

Role—System administrator, organization administrator and manager, operator.

You can shorten the list of samples that are shown in the **Samples / Manage Samples** screen.

You can also search and filter in other columns, or filter by sample creation date or sample attribute value. For more information, see “Filter and search for samples” on page 74, “Filter samples by creation date” on page 76, and “Filter samples by sample attribute value” on page 77.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, select a category for the samples.

Option	Description
To Be Prepared	List only the samples that have not been used to prepare as a library batch. This option filters out BAM/CRAM samples.
BAM/CRAM Samples	List samples that were created from BAM and CRAM files.
All	List all samples.

The list of samples is changed to show only the selected type or preparation status of the samples.

Filter samples by creation date

Role—System administrator, organization administrator and manager, operator.

Use filter tools ▼ **(Filter)** in column headings in Genexus™ Software to narrow search results or shorten the list of samples when you view the list of samples.

You can also search and filter in other columns, filter by sample attribute value, or narrow the list by sample type or preparation status. For more information, see “Filter and search for samples” on page 74, “Filter samples by sample attribute value” on page 77, and “Filter samples by sample type or preparation status” on page 75.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, use the following options.

Option	Description
List only samples created on an exact date.	a. In the Created On column heading, click ▼ (Filter). b. In the first dropdown list, select Is equal to. c. In the calendar, select the date of interest. d. Click Filter.
List samples created within a date range.	a. In the Created On column heading, click ▼ (Filter). b. In the first dropdown list, select Is after or equal to. c. In the calendar, select the earliest date that the samples of interest were created. d. In the Boolean operator dropdown list, select And. e. In the second dropdown list, select Is before or equal to. f. In the calendar, select the latest date that the samples of interest were created. g. Click Filter.
List samples according to a date that meets either one condition or a second condition.	a. In the Created On column heading, click ▼ (Filter). b. In the first dropdown list, select Is after or equal to. c. In the calendar, select the earliest date that the samples of interest were created. d. In the Boolean operator dropdown list, select Or. e. In the second dropdown list, select Is before or equal to. f. In the calendar, select the latest date that the samples of interest were created. g. Click Filter.
List only samples that were created before a date that you indicate.	a. In the Created On column heading, click ▼ (Filter). b. In the first dropdown list, select Is before or equal to. c. In the calendar, select the date of interest. d. Click Filter.

Option	Description
List only samples that were created after a date that you indicate.	- In the Created On column heading, click  (Filter). - In the first dropdown list, select Is after or equal to. - In the calendar, select the date of interest. - Click Filter.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

Records are listed for samples created on dates that meet the filter conditions. In the column heading,  changes color to indicate that a filter is applied.

Filter samples by sample attribute value

Role—System administrator, organization administrator and manager, operator.

You can filter the list of samples by the numeric value of some sample attributes, such as **%Cellularity** or **%Necrosis**.

You can also search and filter in other columns, filter by sample creation date, or narrow the list by sample type or preparation status. For more information, see “Filter and search for samples” on page 74, “Filter samples by creation date” on page 76, and “Filter samples by sample type or preparation status” on page 75.

1. In the menu bar, click **Samples ► Manage Samples**.
2. In the **Manage Samples** screen, click **Columns** and select an attribute that uses a numeric value, such as **%Cellularity**.
The selected column is added to the table.

3. In the column that you added, use the following options.

Option	Description
List only samples with an exact sample attribute value.	- In the sample attribute column heading of interest, click  (Filter). - In the first dropdown list, select Is equal to. - Select the value of interest in the next field. - Do not change the remaining fields. - Click Filter.
List samples created within a range of sample attribute values.	- In the sample attribute column heading or interest, click  (Filter). - In the first dropdown list, select Is greater than or equal to. - In the Boolean operator dropdown list, select And. - In the second dropdown list select Is less than or equal to. - Click Filter.
List samples according to a sample attribute value that meets multiple criteria.	- In the sample attribute column heading of interest, click  (Filter). - In the first dropdown list, select Is less than or equal to. - In the Boolean operator dropdown list, select Or. - In the second dropdown list, select Is greater than or equal to. - Click Filter.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

Records are listed for sample attributes that meet the filter criteria. In the column heading,  changes color to indicate that a filter is applied.

4. Records are listed for samples created on dates that meet both conditions.

Sort the list of samples

You can sort the list of samples to make it easier to find a sample in Genexus™ Software.

By default, samples are listed in the **Samples / Manage Samples** screen by the date that the sample was created. The most recently created sample is at the top of the list.

You can change the order that the samples are listed in the table.

- In the menu bar, click **Samples ► Manage Samples**.
- In the **Manage Samples** screen, click a column heading of interest.
The list of samples is sorted by the sample attribute, in alphabetical or chronological order of the attribute, depending on the heading.
- Click the column heading again to reverse the order.

Delete samples

You can delete samples that have not been assigned to a run. Samples that are assigned to a run or library batch cannot be deleted.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to delete.
The options that you can apply to multiple samples appear.
3. Click  **Delete**.
The **Delete Sample** dialog box opens with the message *Are you sure that you want to delete selected sample(s)?*
4. Click **Yes** to delete the selected samples.

The samples are removed from the software.

Review and download audit records for a sample

The entire history of a sample definition is available to review and download in Genexus™ Software. An audit trail lists the records for each sample to which changes are made. Each audit record in the list shows the name of each sample attribute, and the original sample attribute settings and any new attribute settings.

The following actions generate an audit record.

- Create a new sample.
- Edit a sample, including changes to sample tags.
- Delete a sample.
- Add a note.
- Delete a note.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample of interest, then click **Audit**.
A list of time-stamped audit records that includes information for each action performed on the sample is shown in the **Audit Details** screen.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.

4. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the sample is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attribute mappings Manage Samples screen.

Manage library batches in Genexus™ Software

You can edit information for library batches, add and delete notes for library batches, delete library batches, and review the history of changes made to library batches in Genexus™ Software to updates of sample information, or barcode information.

Edit a library batch

Role—System administrator, organization administrator and manager, operator.

You can edit a library batch only if none of the libraries in the library batch have been assigned to a run plan. You might edit a library batch to update sample information, or barcode information, for example.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, place the pointer over the row of a library batch that you want to edit, then click **Edit**.
3. In the **Edit Library batch** screen, modify the information, then click **Submit**.

Add a note to a library batch

Role—System administrator, organization administrator and manager, operator.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, place the pointer over the row of a library batch of interest, then click **Notes**.
3. In the **Notes** dialog box, click  **Add Note**.
4. In the **Notes** dialog box, enter the note, then click **Save**.
The note appears in the **Notes** dialog box.

Delete a library batch note

Role—System administrator, organization administrator and manager, operator.

1. In the menu bar, click **Samples ► Manage Library Batches**.
2. In the **Manage Library Batches** screen, place the pointer over the row of a library batch of interest, then click **Notes**.
3. In the **Notes** dialog box, locate the note to delete, then click  **Delete**.
The note is removed from the **Notes** dialog box.

Delete a library batch

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can delete library batches that are not yet included in a run plan. You cannot delete the library batch if the library batch is used in a run plan.

1. In the menu bar, click **Samples ► Manage Library Batches**.
2. In the **Manage Library Batches** screen, select the checkbox next to one or more library batch IDs, then click  **Delete**.

Find library batches

Role—System administrator, organization administrator and manager, operator.

You can sort, search, and filter information to narrow the library batches that are listed in the **Samples / Manage Library Batches** screen in Genexus™ Software. The sort, search, and filter tools help you organize the library batches and find a specific library batch.

To sort the list of library batches to narrow your search, see “Sort the list of library batches” on page 83.

To search the list of library batches for a unique library batch identifier, such as the library batch ID, see “Search for library batches” on page 82.

To filter the list of library batches by a common trait, such as the type of nucleic acid in the library batch, see “Filter the list of library batches” on page 82.

Filter the list of library batches

You can apply filters to narrow search results, or shorten the list of library batches when you view the list of library batches in Genexus™ Software.

You can also search for library batches with keywords or text. For more information, see “Search for library batches” on page 82.

In addition to the filter tools in the column headings, you can also filter the list of library batches with a dropdown list. For more information, see “View library batches” on page 43.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, select one or more filters.

Option	Description
List all library batches that are associated with a sample.	In the Total Samples column heading, click  (Filter) then select one or more samples of interest.
Filter and search by the Library Batch ID or other column headings.	In the Library Batch ID column heading, click  (Filter) then use the options to filter the list in the filter dialog box.
Filter by the type of nucleic acid in the library.	In the Nucleic Acid Type column heading, click  (Filter) then use the options to filter the list in the filter dialog box.
Filter by the full assay name in the library batch.	In the Assay Full Name column heading, click  (Filter) then use the options to filter the list in the filter dialog box.
Filter the list of all samples that are included in library batches.	In the Total Samples column heading, click  (Filter) then use the options to filter the list in the filter dialog box.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

3. In the filter dialog box, click **Filter** to apply the filtered selections.
In the column heading,  changes color to indicate that a filter is applied.

The list of library batches is filtered according to the selections.

Click  **Clear Filters** to remove all filters and view the full list of libraries.

Search for library batches

Library batches are listed in the **Samples / Manage Library Batches** screen in Genexus™ Software. The most recently created library batch is at the top of the list.

You can search for library batches with full or partial keywords of the following items that are associated with the library batches.

- **Library Batch ID**
- **Assay Full Name**
- **Nucleic Acid Type**
- **Sample Name**

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, in the column heading of interest, click  (**Filter**).
3. In the search field, enter a full or partial name or ID of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
4. In the filter dialog box, select from the limited list.
 - Select a checkbox next to the name or ID to select one or more items.
 - Click **Select All** to select all of the items listed in the filter dialog box.
 - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
In the column heading,  changes color to indicate that a filter is applied.

The filtered list of libraries is shown in the **Manage Library Batches** screen.

Click  **Clear Filters** to remove all filters and view the full list of library batches.

Sort the list of library batches

You can sort the list of library batches to make it easier to find a library batch in Genexus™ Software.

By default, library batches are listed in the **Samples / Manage Library Batches** screen by the date the library batch was created. The most recently created library batch is at the top of the list.

You can change the order that the library batches are listed and sort the library batches alphabetically or reverse alphabetically by the **Library Batch ID** or **Assay Full Name**.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, click the column heading of interest.
The list of library batches is sorted alphabetically.
3. Click the column heading again to reverse the order.

Review and download audit records for a library batch

Role—System administrator, organization administrator and manager, operator.

The entire history of changes to a library batch is available to review and download in Genexus™ Software. An audit trail lists the records for each library batch to which changes are made. Each audit record in the list shows the name of each library batch, and the original library batch settings and any new library batch settings.

The following actions generate an audit record.

- Create a library batch.
- Edit a library batch.
- Delete a library batch.
- Add a note.
- Delete a note.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples ▶ Manage Library Batches**.
2. In the **Manage Library Batches** screen, place the pointer over the row of a library batch that you want to audit, then click **Audit**.
A list of time-stamped audit records that includes information for each action performed on the library batch is shown in the **Audit Details** screen.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.
4. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the sample is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attribute mappings Manage Library Batches screen.



Plan and manage runs

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Run plans in Genexus™ Software

Role—System administrator, organization administrator and manager, operator.

Run plans created in Genexus™ Software contain the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes. Run plans are used to track samples, consumables, and chips throughout the library preparation, templating, sequencing, and data analysis workflow.

Administrators, managers, and operators can create the following types of run plans in Genexus™ Software. The type of run plan that you use plan depends on the type of sample used for the run. For more information, see “About samples and library batches” on page 41.

Run plan type	Description
Sample to Result	An integrated run plan for sequential and automated nucleic acid purification and sequencing. For more information, see “Plan a Sample to Result run” on page 89.
Nucleic Acid to Result	A sequencing run plan that uses nucleic acid samples as input. For more information, see “Plan a Nucleic Acid to Result run” on page 97.
Library to Result	A sequencing run plan that uses libraries that you have previously prepared manually as input. For more information, see “Plan a Library to Result run” on page 103.
BAM/CRAM to Result	A run plan to analyze samples that are imported from BAM or CRAM sample files. For more information, see “Plan a BAM/CRAM to Result run” on page 106.
Purification Only	A run plan that is used only for purification of samples on the Genexus™ Purification System. Only settings for the purification run can be configured with this run type. For more information, see “Plan a Purification Only run” on page 95.

The software guides you step-by-step through the process to set up a run. The software prompts you to select required information and consumables, then provides a printed run setup guide to help you load consumables on the sequencer and the purification instrument, if applicable.

Before you plan a run

The following conditions must be met before you plan a run in Genexus™ Software.

- Sample information is accurate in the software and the name assigned to each sample is unique. For more information, see “Enter or import samples in Genexus™ Software” on page 43.
- A Genexus™ Purification System is connected to a Genexus™ Integrated Sequencer for use with a **Sample to Result** run.
- Ensure that the following conditions are met for a **Library to Result** run.
 - Library batches are prepared and each batch uses a unique library batch ID.
 - Each sample library in a library batch is prepared with and assigned a unique barcode or barcode pair.

For more information, see “Enter or import a library batch in Genexus™ Software” on page 53.

For guidelines about library quantification, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. [MAN0017910](#)).

The software returns an error message when any of the conditions are not met.

Find a run plan

You can search, filter, and sort information to narrow the run plans that are listed in the **Runs / Manage Runs** screen. The sort, search, and filter tools help you organize runs and find a specific run plan in Genexus™ Software. You can also apply the search, sort, and filter tools in the **Assays** step when you plan a run. Use the tools to limit the assays that are listed so that you can easily identify one or more assays to include in the run plan.

For information to search for a run plan by a unique identifier, such as the name of the run plan or the assay applied to the run plan, see “Search for a run plan” on page 87.

For information to filter run plans by a common trait, such as the run plans that include purification, see “Filter the list of run plans” on page 88.

Search for a run plan

Run plans are listed in the **Runs / Manage Runs** screen in Genexus™ Software. The most recently created run plan is on the top of the list.

You can search for run plans with keywords or text that for attributes such as the name of the run plan or the name of an assay that is applied to the run plan.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, in the **Run Name** column heading or **Assay Name** column heading, click ▼ (**Filter**).
When you select the **Run Name** or **Assay Name** of a **BAM/CRAM to Result** run plan that was created with Genexus™ Software 7.0, files listed can be in both BAM and CRAM format. Earlier versions of Genexus™ Software include only run plans that use the BAM file format.
3. In the search field, enter a full or partial assay or run name of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.

4. In the filter dialog box, select from the limited list.
 - Select a checkbox next to a run or assay name to select one or more items.
 - Click **Select All** to select all items listed in the filter dialog box.
 - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of run plans is shown in the **Manage Runs** screen.

Click  **Clear Filters** to remove all filters and view the full list of run plans.

Filter the list of run plans

You can apply filters to narrow search results or shorten the list of run plans when you view the list of run plans in Genexus™ Software.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, select one or more filters.

Option	Description
Filter and search by the run name or the assay selected in the run.	For more information, see “Search for a run plan” on page 87.
Filter by the number of samples in the run.	In the Total Samples column heading, click  (Filter) then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run.
List runs based on whether the run includes purification.	In the Run Status column heading, click  (Filter) then select a status. • Select Planned to list all Nucleic Acid to Result, Library to Result, and BAM/CRAM to Result planned runs. • Select Purification Planned to list planned Sample to Result runs only.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

3. In the filter dialog box, click **Filter** to apply the filtered selections.
In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

The list of run plans is filtered according to the selections.

Click  **Clear Filters** to remove all filters and view the full list of run plans.

Sort the list of run plans

You can sort the list of run plans to make it easier to find run plans in Genexus™ Software.

By default, run plans are listed in the **Runs / Manage Runs** screen by the date the run plan was created. The most recently created run plan is at the top of the list.

You can change the order that the run plans are listed and sort the run plans alphabetically or reverse alphabetically.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, click the **Run Name** heading.
The list of runs is sorted alphabetically.
3. Click the column heading again to reverse the order.

Plan a Sample to Result run

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can plan a run that begins with purification of a sample on a Genexus™ Purification System, then continues with sequencing on a Genexus™ Integrated Sequencer.

When you plan a **Sample to Result** run with multiple assays, the nucleic acid isolation is performed in a separate batch for each assay. Nucleic acid isolation is also performed in a separate batch for each sample type, except for FFPE tissue and FNA, which are grouped together in a single batch for each assay. The Genexus™ Purification System can run one nucleic acid isolation batch at a time. You can run different nucleic acid isolation batches simultaneously on multiple purification instruments or sequentially on a single instrument.

Ensure that the following prerequisites are complete before you plan a **Sample to Result** run.

- Connect a Genexus™ Purification System with the Genexus™ Integrated Sequencer.
- Enter sample information into Genexus™ Software. For more information, see “Create a single sample” on page 44, or “Create multiple samples with a sample definition file” on page 46.

Planning a **Sample to Result** run is organized into five steps: **Setup**, **Assays**, **Samples**, **Purification**, and **Review**. Progress through the steps is tracked in the **Runs / Sample to Result** screen.

1. In the menu bar, click **Runs ▶ Sample to Result**.
You can also click **+ Sample to Result** in the **Runs / Manage Runs** screen.
2. Complete the **Setup** step.
 - a. In the **Plan** section, enter a unique name.
The name is limited to 50 characters and no spaces are allowed.
 - b. In the **Reporting (Optional)** section, enable **Generate Report** to generate a variant report that uses the default report template.

- c. In the **Reporting (Optional)** section, enable **Upload BAM/CRAM files to Server** to upload BAM and CRAM files to another sequencer or to Ion Reporter™ Software.

Option	Description
Automatically upload results files in BAM or CRAM format to another sequencer that uses Genexus™ Software 7.0. Or, upload BAM files to another sequencer that uses an earlier version of the software.	In the dropdown lists, select the Genexus™ Software account and the software version. To configure a Genexus™ Software account that you can use for data uploads, click Set up Account . For more information, see “Register Genexus™ Software accounts” on page 354.
Automatically upload data for further analysis with Ion Reporter™ Software.	In the dropdown lists, select the Ion Reporter™ Software account and the software version. To configure an Ion Reporter™ Software account that you can use for data uploads, click Set up Account . To transfer data to Ion Reporter™ Software, you must add one or more Ion Reporter™ Software accounts in Genexus™ Software. To configure an Ion Reporter™ Software account that you can use for data uploads, see “Register an Ion Reporter™ Software on Connect account” on page 355 and “Register an Ion Reporter™ Server account” on page 356.

Alternatively, you can upload BAM and CRAM files to another sequencer or to Ion Reporter™ Software after a run is successfully completed. For more information, see “Upload results files to another sequencer” on page 195 and “Upload results files to Ion Reporter™ Software” on page 196.

- d. Click **Next**.

If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Assays** step, select one or more assays that you want to include in the run by selecting a research application for the assay.

Only assays that are configured for **Sample to Result** runs are listed. If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

Tip: Use the ▼ (**Filter**) tools in table column headings to find assays of interest. For more information, see “Filter the list of assays shown when you plan a run” on page 108. To create a new assay, see Chapter 8, “Create and manage assays”.

- a. In the **Research Application** column for the assay of interest, select one or more research applications, such as **DNA and Fusions**, to include the research application for the assay in the run plan.

After selecting an assay, the list is filtered to show compatible assays that can be selected and run at the same time.

- b. In the **Application Type** column for the assay of interest, select an application type, such as **DNA and Fusions**, to include the application type for the assay in the run plan.
 - c. If more than one assay is included in the run, repeat substep 3a to select a research application for each assay.
 - d. Click **Next**.
4. In the **Samples** step, select the samples that you want to run with each research application of each assay.
- a. Select the checkbox next to each sample that you want to assign to the research application of an assay, then in the **Selected Assays** pane, for the assay and research application that you want to use for the selected samples, click **Assign**.
- The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including positive controls, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.
- If the minimum reads per sample $\times$ number of samples exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you will not achieve the required reads per sample to pass QC metrics.

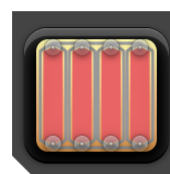
Ion Torrent™ GX5™ Chip



1-2-3	OCAv3 DNA Fus	4
Lane	w5.0.2	Samples

Green lane color denotes lane usage and sample assignment within lane capacity.

Ion Torrent™ GX5™ Chip



1-2-3-4	OCAv3 DNA Fus	7
Lane	w5.0.2	Samples

Red lane color denotes sample assignment that exceeds lane capacity.

- b. If you selected more than one research application or assay, repeat substep 4a for each research application for each assay in the run plan.
- c. If needed, change sample assignments in one of the following ways.
 - Click **View & Remove**, make the selections, then click **Update**.
 - Click **Remove All**, to remove all sample assignments for all assays.

- d. If you selected the **Upload BAM/CRAM files to Server** reporting option in the **Setup** step, make the following selections from the **Genexus Workflow** or **Ion Reporter Workflow** dropdown list for each assay that you selected.
- Select **Upload Only** to upload sample data to the selected server automatically after run completion.
 - Select the desired Genexus™ Software assay or Ion Reporter™ Software analysis workflow to upload sample data *and* perform an analysis in the target software automatically after run completion.

Note: For the Ion Reporter™ Software analysis workflow to appear in the list, you must tag the analysis workflow for use with the IonReporterUploader plugin. For more information, see the Ion Reporter™ Software help system or the user guide available at <http://www.thermofisher.com/ngssoftwaresupport>.

- e. (Optional) For each research application of each assay in the run plan, select **NTC** to include a no template control and select **Control Sample** to include a positive control. The **Chip View** updates to show the lanes used in the run for the included controls.

- f. Click **Next**.

5. In the **Purification** step, review and edit selections, if needed.

- a. Review and edit purification and control sample selections.

Option	Description
Protocol Selection	Select a protocol for each purification kit.
Elution Vol. (µL)	Modify the elution volume within the allowable range, if needed.
Review?	• Deselect the Review? checkbox in the row of a purification kit to enable the sequencer to bypass sequencing samples with concentrations below the threshold for the purified samples. This option does not require you to review samples during the run. • Select the checkbox in the row of a purification kit to review the run after purification, before sequencing, for the kit. This allows you to review samples when the quantity of a nucleic acid is below the quality threshold. You can then decide on a per sample-basis whether or not to sequence the samples. Note: This selection, whether selected or not, overrides the Review? setting of the assay.

- b. Review and edit the information for control samples, if applicable.

Option	Description
Conc. (ng/µL)	Review and edit the concentration of control samples, if needed.
Kit Barcodes	Use the connected barcode scanner to enter the extraction kit barcodes, if needed. Or you can enter the barcodes manually, if needed.

- c. Click **Next**.

6. If a configurable plugin is available, such as the **coverageAnalysis** plugin, or a user-defined plugin that allows configuration, you can configure settings for the plugin.
 - a. In the **Plugins** step, click the **Configure** link to change the settings for a plugin that includes configuration options. For more information, see Chapter 13, “Plugins for data analysis”.
You must save the run plan in the **Review** step to save any changes that are made to plugin settings.
 - b. Click **Next**.
7. In the **Review** step, review the run plan summary, then click **Save**.

A run can include multiple purification batches.

For more information, see “Plan a Sample to Result run” on page 89.

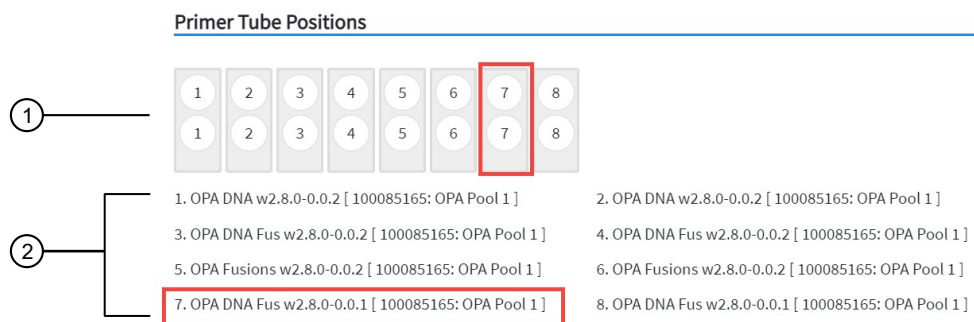
The run plan summary lists the following details.

- The consumables that are required for purification.
- The consumables that are required for sequencing.
- How much volume to load.
- Details about the assay or assays.
- Where to load samples.

Tip: Click **Sequencing** to expand the sequencing section of the run plan summary.

IMPORTANT! The consumables required for sequencing can change. After purification, review the run plan in the **Runs / Manage Runs** screen for updated sequencing consumable information.

Note: If you are using an assay with Ion AmpliSeq™ HD library chemistry, the primer pool positions show that HD primer pools occupy both rows.



① The position of the primer tubes.

② The legend for the image.

In the example outlined in red, primer tube 7, which contains OPA Pool 1 for the OPA DNA Fusion assay, is in the seventh position from the left.

The run plan appears in the **Manage Runs** screen with the name that you specified.

On the purification instrument screen, select the run and then load the samples.

The run is started on the purification instrument. You can monitor the progress of the purification run in the **Dashboard** and in the **Results / Run Results** screen.

When purification and sequencing are complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the Genexus™ Integrated Sequencer user guide.

Review purified samples for a Sample to Result run

A **Sample to Result** run integrates nucleic acid purification and sequencing. When you create a **Sample to Result** run plan, you can include an option that allows samples to be reviewed before library preparation starts. Samples that do not meet a specified concentration threshold after purification have a run status of **Purification Review Required**.

You can also apply this option when you create an assay. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.

1. In the menu bar, click **Results ▶ Run Results**.

Sample to Result runs that require review after purification have a **Run Status** of **Purification Review Required**.

2. In the **Run Results** screen, place the pointer over the row of the run plan of interest, then click **Review**.
3. In the **Purification Samples** screen, review the concentration for each sample, then select the checkbox in the row for each sample that you want to sequence. The options are as follows.
 - Sequence all samples.
 - Sequence only the samples that have a concentration within a specified threshold.
 - Review the samples that do not have a concentration within a specified threshold, then select samples to sequence on a per sample-basis.

4. Click **Submit**.

The samples are ready for sequencing.

Abort a Sample to Result run

This can be done after purification completes but before the run is continued on the sequencer.

Role—System administrator, organization administrator and manager, operator.

When administrators, managers, and operators create a **Sample to Result** run plan, an option can be set that allows samples to be reviewed before library preparation starts. When you enable the review of samples after purification, you can end a run after purification completes if the sequencing run is not started on the sequencer.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of the run that you want to abort, then click **Abort**.
3. In the confirmation dialog box, click **Yes** to confirm.

In the **Run Summary**, you can view information about the purification run. The status of the run in the **Run Summary** is **Aborted**. For more information, see “View the Run Summary” on page 200.

Plan a Purification Only run

Role—System administrator, organization administrator and manager, operator.

This procedure applies only if a Genexus™ Purification System is connected to the Genexus™ Integrated Sequencer for use with integrated purification runs. You can plan a purification run and review the purified samples on the Genexus™ Software that is installed on the sequencer.

Note: Run plans for a Genexus™ Purification System that is not connected to a Genexus™ Integrated Sequencer must be created on the purification instrument touchscreen.

Ensure that the following prerequisites are complete before you plan a **Purification Only** run.

- Connect a Genexus™ Purification System with the Genexus™ Integrated Sequencer. For more information, see the *Genexus™ Purification System User Guide* (Pub. No. [MAN0018475](#)).
- Enter sample information into Genexus™ Software. For more information, see “Create a single sample” on page 44, or “Create multiple samples with a sample definition file” on page 46.

Planning a **Purification Only** run is organized into three steps: **Setup**, **Samples**, and **Review**. Progress through the steps is tracked in the **Runs / Purification Only** screen.

1. In the menu bar, click **Runs ▶ Purification Only**.

You can also click **+ Purification Only** in the **Runs / Manage Runs** screen.

2. Complete the **Setup** step.

- a. In the **Plan** section, enter a unique name.

The name is limited to 50 characters and no spaces are allowed.

- b. In the **Purification** section, select a **Purification Kit**.

- c. In the **Purification** section, select protocol from the **Protocol Selection** menu. The **Elution Vol. (µL)** shown is based on the selected protocol. You can adjust the value if needed.

- d. Select **True** or **False** to enable or disable quantitation. When set to **True**, the concentration of the sample is shown in the sample results.

- e. Click **Next**.

3. In the **Samples** step, select the checkbox next to each sample that you want to have purified, , then click **Next**.

The list of samples that are available is based on the protocol selection. Protocols are based on the type of sample that is selected.

4. In the **Review** step, review the run plan summary, then click **Save**.

The run plan summary lists the following details.

- The name and status of the run.
- The consumables that are required for purification.
- How much volume to load.
- Where to load samples.
- The consumables that are required for purification.

Tip: Click **Print** to print the run plan summary. For more information, see “Print a run plan” on page 111.

The run plan is shown in the **Manage Runs** screen with the name that you specified.

On the purification instrument screen, select the run and then load the samples.

The run is started on the purification instrument. You can monitor the progress of the purification run in the **Dashboard** and in the **Results / Purification Only Results** screen.

Review purified samples

Administrators, managers, and operators can review samples in Genexus™ Software that were purified on a Genexus™ Purification System using a **Purification Only** run plan.

1. In the menu bar, click **Results ▶ Purification Only Results**.

Samples that have been purified in a **Purification Only** run and have a **Run Status** of **Purification Completed** can be reviewed.

The following run information is shown in the **Results/Purification Only** screen.

Column	Description
Run Name	The unique name of the run given when it was created in the software.
Purification Kit	The name of the kit that was used in the purification run.
Run Status	The status of the run. For example, Purification Planned , Purification In Progress , or Purification Completed . If a run is aborted the status of the run is Purification Run Aborted .
Archive Plate ID	A unique identification number that is included on each archive plate.
Total Samples	The total number of samples in a run.
Started On	The date and time when the run was started.
Updated On	The date and time when the last action was completed on the run.

The most recently created run is shown on the top of the list. You can sort the columns and filter the columns to find the run for a sample of interest.

Users with appropriate permissions can perform the following actions in the **Results/Purification Only** screen.

Option	Description
Actions	Available action links for a run are shown when you place the pointer over the row of a run. The actions that are available depend on the type of run. • Audit—View the audit trail for the run. • CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see “Download a Customer Support Archive” on page 586.

2. Click a run name to review the **Run Summary** for each sample. For more information, see “View the Run Summary” on page 200 .

After you review the purification results, you can add the purified samples when you plan a **Nucleic Acid to Result** run. For more information, see “Plan a Nucleic Acid to Result run” on page 97.

Plan a Nucleic Acid to Result run

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can plan a run for sequencing that starts with nucleic acid samples. Those samples might be purified using the Genexus™ Purification System or another purification instrument. A run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

Planning a **Nucleic Acid to Result** run is organized into steps: **Setup**, **Assays**, **Samples**, **Sample Plate**, and **Review**. Progress through the steps is tracked in the **Runs / Nucleic Acid to Result** screen.

Ensure that the following prerequisites are complete before you plan a **Nucleic Acid to Result** run.

- Enter sample information into Genexus™ Software. For more information, see “Create a single sample” on page 44, or “Create multiple samples with a sample definition file” on page 46.
- Ensure that the assay that you want to use in the run exists in the software. For more information, see “Assays in Genexus™ Software” on page 233.
- Quantify sample concentration, if you do not intend to dilute sample concentrations manually to the target concentration of the assay.

1. In the menu bar, click **Runs ▶ Nucleic Acid to Result**.

Alternatively, click **+ Nucleic Acid to Result** in the **Runs / Manage Runs** screen.

2. Complete the **Setup** step.

- a. In the **Plan** section, enter a unique run plan name.

The name is limited to 50 characters and no spaces are allowed.

- b. In the **Reporting (Optional)** section, enable **Generate Report** to generate a variant report using the default report template.

- c. In the **Reporting (Optional)** section, enable **Upload BAM/CRAM files to Server** to upload results files to another Genexus™ Integrated Sequencer or Ion Reporter™ Software.

Option	Description
Automatically upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer that uses Genexus™ Software 7.0. Or, upload BAM files to another Genexus™ Integrated Sequencer that uses an earlier version of the software.	In the dropdown lists, select the Genexus™ Software account and the analysis pipeline version for the software. To configure a Genexus™ Software account that you can use for data uploads, click Set up Account . For more information, see “Register Genexus™ Software accounts” on page 354.
Automatically upload data for further analysis with Ion Reporter™ Software.	In the dropdown lists, select the Ion Reporter™ Software account and analysis pipeline version for the software. To configure an Ion Reporter™ Software account that you can use for data uploads, click Set up Account . To transfer data to Ion Reporter™ Software, you must add one or more Ion Reporter™ Software accounts in Genexus™ Software. To configure an Ion Reporter™ Software account that you can use for data uploads, see “Register an Ion Reporter™ Server account” on page 356 and “Register Genexus™ Software accounts” on page 354.

Alternatively, you can manually upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software after a run is successfully completed. For more information, see “Upload results files to another sequencer” on page 195 and “Upload results files to Ion Reporter™ Software” on page 196.

3. In the **Assays** step, select one or more assays that you want to include in the run.

Tip: Use the  (**Filter**) tools in table column headings to find assays of interest. For more information, see “Filter the list of assays shown when you plan a run” on page 108.

- a. In the **Research Application** column for the assay of interest, select one or more research applications, such as **DNA and Fusions**, to include the research application for the assay in the run plan.

After you select an assay and the research application for the assay, the list is filtered to show compatible assays that can be selected and run at the same time.

- b. If more assays are included in the run, repeat substep 3a for each extra assay.

- c. Click **Next**.

4. In the **Samples** step, select the samples that you want to run with each research application of each assay.

- a. To narrow the list of samples, select an option from the dropdown list above the table to filter the samples. For example, select **Genexus Purified** to view samples that were purified using the Genexus™ Purification System.

- b. Select the checkbox next to each sample that you want to assign to the research application of an assay, then in the **Selected Assays** pane, for the assay and research application that you want to use for the selected samples, click **Assign**.

The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including positive controls, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

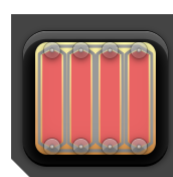
If the *minimum reads per sample × number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you will not obtain the reads needed per sample to pass QC metrics.

Ion Torrent™ GX5™ Chip



① 1-2-3 Lane OCAv3 DNA Fus 4 Samples
w5.0.2

Ion Torrent™ GX5™ Chip



② 1-2-3-4 Lane OCAv3 DNA Fus 7 Samples
w5.0.2

- ① Green lane color denotes lane usage and sample assignment within lane capacity.

- ② Red lane color denotes sample assignment that exceeds lane capacity.

- c. If you selected more than one research application or assay, repeat substep 4b for each research application for each assay in the run plan.
- d. If needed, change the sample assignment in one of the following ways.
- Click **View & Remove**, make the selections, then click **Update**.
 - Click **Remove All**, to remove all sample assignments for all assays.
- e. If you selected the **Upload BAM/CRAM files to Server** reporting option in the **Setup** step, make the following selections from the **Genexus Workflow** or **Ion Reporter Workflow** list for each assay that you selected.
- Select **Upload Only** to upload sample data to the selected server automatically after run completion.
 - Select the desired Genexus™ Software assay or Ion Reporter™ Software analysis workflow to upload sample data *and* perform an analysis in the target software automatically after run completion.

Note: For the Ion Reporter™ Software analysis workflow to appear in the list, you must tag the analysis workflow for use with the IonReporterUploader plugin. For more information, see the Ion Reporter™ Software help system or the user guide available at <http://www.thermofisher.com/ngssoftwaresupport>.

- f. If desired, for each research application of each assay in the run plan, select **NTC** to include a no-template control.

The **Chip View** updates to show the lanes used in the run for the included no-template controls.

- g. Click **Next**.

5. In the **Sample Plate** step, review the position assignments in the sample plate and change if needed. Use a Microsoft™ Excel™ spreadsheet to change well positions and sample concentrations.

- a. Click **Manage Sample Positions ▶ Export Current** to export a spreadsheet of the current plate positions to your computer.
- b. Review the spreadsheet and make changes, if needed.
- c. Click **Manage Sample Positions ▶ Import From External** then locate the spreadsheet on your computer and upload the spreadsheet to the sequencer.

6. Alternatively, modify the well position and concentration of samples the **Sample Plate** step, if needed.

- a. For a single sample, in the row of the sample of interest, in the **Conc. (ng/μl)** column, edit the concentration.

The concentration values are automatically populated for samples that are purified using the Genexus™ Purification System. The concentration values are retrieved from the purification run that is selected.

To modify the concentration of multiple samples, select the samples of interest, then click  **Edit Concentration**. In the **Bulk Edit** dialog box, enter the concentration for all selected samples, then click **Submit**. The concentration for each selected sample is updated to the new value.

The sequencer automatically dilutes the sample to the target concentration during the run if either of the following is true:

- The sample concentration is $\leq 1,024.00X$ the target concentration for an assay that performs 4 reactions per library.
- The sample concentration is $\leq 40.00X$ the target concentration for an assay that performs 6 reactions per library strip.

If a sample concentration is $> 1,024.00X$ or $40.00X$ the target concentration, depending on the assay, you must manually dilute the sample to the target concentration, or to a value within range for automated dilution before loading on the sample plate.

Note: The sample volume that is required for library preparation is not adjustable. The volume depends on the number of primer pools in the assay, sample type, and library chemistry.

- b. Drag-and-drop samples and no-template controls to change the location of samples and controls, if applicable.

7. In the **Sample Plate** step, complete the following options.

- a. If you are using purified samples, select the **Purification Run** from which each sample is used in the sequencing run.

- b. Use the connected barcode scanner to enter the kit barcode for one or more samples or controls, if applicable. For a single sample, in the row of the sample of interest, in the **Kit Barcodes** column, enter the extraction kit barcode or control kit barcode, if applicable. For multiple samples or controls, select the samples or controls, then click [✎ Assign Kit Barcodes](#). In the **Assign Kit Barcodes** dialog box, enter the kit barcode for the samples, and if applicable, enter the barcode for the no-template control.
If needed, you can enter the barcodes manually.
 - c. Ensure that sample plate information is correct, then click **Next**.
8. If a configurable plugin is available, such as the **coverageAnalysis** plugin, or a user-defined plugin that allows configuration, you can configure settings for the plugin.
 - a. In the **Plugins** step, click the **Configure** link to change the settings for a plugin that includes configuration options. For more information, see Chapter 13, “Plugins for data analysis”.
You must save the run plan in the **Review** step to save any changes that are made to plugin settings.
 - b. Click **Next**.
9. In the **Review** step, review the run plan summary, then click **Save & Print** to print the run setup guide, if desired. Click **Save** to save the run without printing.
To print the run setup guide, allow pop-ups in the browser.
The run plan summary lists the following details.
 - The consumables that are required for this run.
 - How much sample volume to load.
 - Where to load samples and primer pool tubes.
 - Details about the assay.
 - For purified samples, the **Archive Plate ID** and **Archive Well Positions**.

Genexus | Ion Torrent

Dashboard Samples Runs Results Assays

Runs / Nucleic Acid to Result

Setup Assays Samples Sample Plate Review

Review run plan summary and print the run setup guide.

Run Name: Test_Plan_1

Generate Report: true

Server: teemo.tiv - IRS14

Sequencing

Consumables Required For This Run

(1) Genexus™ Coupler (1) GX5™ Chip (1) Ion Torrent™ Genexus™ Barcodes 1-96 AS - 20 barcodes will be used from this plate

(8) Genexus™ Strip 1 (8) Genexus™ Strip 2-AS (3) 96 Well Plates

(5) Ion Torrent™ Genexus™ Pipette tips (3) Genexus™ Strip 3-GX5™ (3) Genexus™ Strip 4

(1) Genexus™ Bottle 3 (2) Genexus™ Bottle 2 (1) Genexus™ Bottle 1

(1) Genexus™ Cartridge

Consumables Installed Ready To Use

None

Primer Tube Positions

1-2-3 Lanes OCAv3 DNA Fus w5.0.2 5 Samples

1 2 3 4 5 6 7 8

1. OCAv3 DNA Fus w5.0.2 [100083283: OCAv3 DNA Pool 1] 2. OCAv3 DNA Fus w5.0.2 [100083284: OCAv3 DNA Pool 2]

3. OCAv3 DNA Fus w5.0.2 [100083285: OCAv3 RNA Pool 1] 4. OCAv3 DNA Fus w5.0.2 [100083286: OCAv3 RNA Pool 2]

5. OCAv3 DNA Fus w5.0.2 [100083283: OCAv3 DNA Pool 1] 6. OCAv3 DNA Fus w5.0.2 [100083284: OCAv3 DNA Pool 2]

7. OCAv3 DNA Fus w5.0.2 [100083285: OCAv3 RNA Pool 1] 8. OCAv3 DNA Fus w5.0.2 [100083286: OCAv3 RNA Pool 2]

Sample Locations

Well Pos.	Sample Name	Nucleic Acid Type	Vol. (µl)	Conc.(ng/µl)	Dilution Factor	Assay Name
A1	GX_6.5.25_OPA_300_thanos_10_DF_R18C7	DNA	25.0	1.11	1	OCAv3 DNA Fus w5.0.2
B1	GX_6.5.25_OPA_300_thanos_10_DF_R18C7	RNA	25.0	0.95	1	OCAv3 DNA Fus w5.0.2
C1	GX_6.5.25_OPA_300_thanos_11_DF_R18C7	DNA	25.0	1.11	1	OCAv3 DNA Fus w5.0.2

Exit Previous Save & Print Save

- ① Run information
- ② List of consumables required for the run
- ③ List of consumables that are installed on the sequencer and available for the run
- ④ Positions to load primer pool tubes
- ⑤ Chip view showing the lanes to be used in the run
- ⑥ Positions in the sample plate to load the samples
- ⑦ Table listing the sample plate position, sample type, volume to load, concentration, dilution factor, and assay for each sample
- ⑧ Description of each primer pool and its position

If you are using an assay with Ion AmpliSeq™ HD library chemistry, the primer pool positions show that HD primer pools occupy both rows.

Primer Tube Positions

1 2 3 4 5 6 7 8

1 2 3 4 5 6 7 8

1. OPA DNA w2.8.0-0.0.2 [100085165: OPA Pool 1] 2. OPA DNA w2.8.0-0.0.2 [100085165: OPA Pool 1]

3. OPA DNA Fus w2.8.0-0.0.2 [100085165: OPA Pool 1] 4. OPA DNA Fus w2.8.0-0.0.2 [100085165: OPA Pool 1]

5. OPA Fusions w2.8.0-0.0.2 [100085165: OPA Pool 1] 6. OPA Fusions w2.8.0-0.0.2 [100085165: OPA Pool 1]

7. OPA DNA Fus w2.8.0-0.0.1 [100085165: OPA Pool 1] 8. OPA DNA Fus w2.8.0-0.0.1 [100085165: OPA Pool 1]

- ① The position of the primer tubes.
- ② The legend for the image.

In the example outlined in red, primer tube 7 contains OPA Pool 1 for the OPA DNA Fusion assay, in the seventh position from the left.

After you save the run plan, the run plan appears in the **Manage Runs** screen in the run list with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer.

When the sequencing run is complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the Genexus™ Integrated Sequencer user guide.

Plan a Library to Result run

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can plan runs for sequencing that start with libraries. A run that starts with prepared libraries is called a **Library to Result** run. Before you can plan a **Library to Result** run, you must first enter sample information and prepare a library batch that associates an assay with the library batch. The library batch must be created with the assay to be used in the run. The assay specifies the barcode set that was used to prepare the sample libraries.

For more information, see “Enter or import a library batch in Genexus™ Software” on page 53.

If the sample libraries were prepared using a barcode set that is not specified in an assay that you want to use in the run, do the following.

- Create a new assay, or copy an existing assay, and specify the new barcode set in assay setup.
- Prepare a library batch that selects the new assay.

For more information, see “Create an assay” on page 234, “Copy an assay” on page 237, and “Prepare a library batch” on page 54.

Genexus™ Software guides you through the four steps of planning a **Library to Result** run: **Setup**, **Assays**, **Library Batches**, and **Review**. Progress through the steps is tracked in the **Runs / Library to Result** screen.

1. In the menu bar, click **Runs ▶ Library to Result**.

Alternatively, click **+ Library to Result** in the **Runs / Manage Runs** screen.

2. Complete the **Setup** step.

- a. In **Run Name**, enter a unique name.

The name is limited to 50 characters and no spaces are allowed.

- b. *(Optional)* In the **Reporting (Optional)** section, enable **Generate Report** to generate a variant report using the default report template.

- c. (Optional) In the **Reporting (Optional)** section, enable **Upload BAM/CRAM files to Server** to upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer or BAM files to Ion Reporter™ Software.

CRAM files can be uploaded only to a Genexus™ Integrated Sequencer that uses Genexus™ Software v7.0. Earlier versions of the software are compatible with BAM files only.

Option	Description
Automatically upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer that uses Genexus™ Software 7.0. Or, upload BAM files to another Genexus™ Integrated Sequencer that uses an earlier version of the software.	In the dropdown lists, select the Genexus™ Software account and the software version. To configure a Genexus™ Software account that you can use for data uploads, click Set up Account . For more information, see “Register Genexus™ Software accounts” on page 354.
Automatically upload data in BAM or VCF file formats for further analysis with Ion Reporter™ Software.	In the dropdown lists, select the Ion Reporter™ Software account and software version. To configure an Ion Reporter™ Software account that you can use for data uploads, click Set up Account . For more information, see “Register an Ion Reporter™ Server account” on page 356. To transfer data to Ion Reporter™ Software, you must add one or more Ion Reporter™ Software accounts in Genexus™ Software. To configure an Ion Reporter™ Software account that you can use for data uploads, see “Register an Ion Reporter™ Server account” on page 356 and “Register Genexus™ Software accounts” on page 354.

Alternatively, you can upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software after a run successfully completes. For more information, see “Upload results files to another sequencer” on page 195 and “Upload results files to Ion Reporter™ Software” on page 196.

- d. Click **Next**.

If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Assays** step, select one or more assays that you want to include in the run.

For the assay to be selectable at this step, you must have prepared a library batch that assigns the assay to the batch. The assay specifies the barcode set that was used to prepare the sample libraries.

- a. Use the ▼ (**Filter**) tools in table column headings to find assays of interest, if desired.

For more information, see “Filter the list of assays shown when you plan a run” on page 108.

- b. In the **Research Application** column for the assay of interest, select one or more research applications, such as **DNA and Fusions**, to include each selected research application for the assay in the run plan.

After you select an assay and the research application for the assay, the list is filtered to show compatible assays that can be selected and run at the same time.

To create a new assay, see Chapter 8, “Create and manage assays”.

- c. If more assays are included in the run, repeat substep 3b for each extra assay.

- d. Click **Next**.

4. In the **Library Batches** step, select the library batch that you want to use in the run, then click **Next**.

Only one library batch can be selected per assay. However, you can plan a library run that uses multiple assays if you select multiple compatible assays in the **Assays** step.

- a. If you selected the **Upload BAM/CRAM files to Server** reporting option in the **Setup** step, make the following selections from the **Genexus Workflow** or **Ion Reporter Workflow** list.
- Select **Upload Only** to upload sample data to the selected software account automatically after run completion.
 - Select the desired Genexus™ Software assay or Ion Reporter™ Software analysis workflow to upload sample data *and* perform an analysis in the target software automatically after run completion.

Note: For the Ion Reporter™ Software analysis workflow to appear in the list, you must tag the analysis workflow for use with the IonReporterUploader plugin. For more information, see the Ion Reporter™ Software help system or the user guide available at <http://www.thermofisher.com/ngssoftwaresupport>.

The **Chip View** updates to show the lanes to be used in the run as green. Lane usage is calculated based on the number of samples and minimum reads per sample entered at assay setup.



Green lane color denotes lane usage and sample assignment within lane capacity.

Red lane color denotes sample assignment that exceeds lane capacity.

If the $\text{minimum reads per sample} \times \text{number of samples}$ exceeds the chip or lane well capacity, a dialog box appears after you click **Next** asking you to confirm that you want to continue. After clicking **Yes**, the **Chip View** updates and shows the lane color as red instead of green. In the example shown at right, seven samples were included in a library batch instead of four. The run is allowed if the lane capacity is exceeded, but you will not achieve the required reads per sample to pass QC metrics.

5. If a configurable plugin is available, such as the **coverageAnalysis** plugin, or a user-defined plugin that allows configuration, you can configure settings for the plugin.
 - a. In the **Plugins** step, click the **Configure** link to change the settings for a plugin that includes configuration options. For more information, see Chapter 13, “Plugins for data analysis”.
You must save the run plan in the **Review** step to save any changes that are made to plugin settings.
 - b. Click **Next**.
6. In the **Review** step, review the run plan summary.
 - Click **Save and Print** to print the run setup guide.
 - Click **Save** to save the run without printing.

To print the run setup guide, you must allow pop-ups in the browser.

The run plan summary lists the consumables that are required for the run, where to load the library batch on the sample plate, and how much library volume to load.

The run appears in the run list on the **Manage Runs** screen with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

Plan a BAM/CRAM to Result run

Role—System administrator, organization administrator and manager, operator.

Before you can plan a **BAM/CRAM to Result** run, you must upload samples files in BAM or CRAM format into Genexus™ Software. For more information, see “Create BAM and CRAM samples from a sample definition file” on page 50.

The process to plan a run to analyze sequencing files in BAM or CRAM format is organized into steps: **Assays**, **Samples**, and **Review**. Progress through the steps is tracked in the **BAM/CRAM to Result** screen.

1. In the menu bar, click **Runs ▶ BAM/CRAM to Result**.
Alternatively, click **+ BAM/CRAM to Result** in the **Runs / Manage Runs** screen.
2. Complete the **Assays** step.
 - a. Enter a unique name.
The name is limited to 50 characters and no spaces are allowed.
 - b. Use the **▼ (Filter)** tools in table column headings to find assays of interest, if desired.
For more information, see “Filter the list of assays shown when you plan a run” on page 108.
 - c. In the **Research Application** column for the assay of interest, select the research application, such as **DNA and Fusions** or **DNA**, to include the selected research application for the assay in the run plan.
You can select only one research application of an assay per run.
 - d. Click **Next**.

3. In the **Samples** step, select the samples from the list to analyze, then click **Assign**.
Only samples that are compatible with the selected assay are listed. Compatibility is determined by the nucleic acid type, such as **DNA** or **RNA**.
 - a. If needed, click  in an assigned sample tile to remove the sample from the plan.
 - b. Click **Next**.
4. In the **Review** step, review the samples in the run plan summary, then click **Save and Launch**.
The run is listed in the **Results / Run Results** screen. The run is launched, or the run is queued if other runs are in progress.

Filter and search for an assay to use in a run plan

You can search for an assay with keywords or text that are included in the name or full name of an assay to help find the assay that you want to use in a run plan. You can also apply filters to shorten the list of options, then find an assay for use in a run plan.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. In the menu bar, click **Runs**, then click **Sample to Result**, **Nucleic Acid to Result**, **Library to Result**, or **BAM/CRAM to Result**, based on the type of run that you want to plan.
2. Enter and make the selections in the **Setup** step, if applicable, then click **Next**.
3. In the **Assays** step, filter or search for an assay.
 - a. Click a  (**Filter**) in a column heading.
 - b. In the search field, enter at least 3 characters for the name of the attribute.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - c. In the filter dialog box, select an option from the limited list.
 - Select a checkbox next to an option to select one or more option.
 - Click **Select All** to select all options in the filter dialog box.
Click **Filter**. In the column heading,  (**Filter**) changes color to indicate that a filter is applied.
 - Click **Clear** to close the filter dialog box.

4. Click the **Filter Assays By** dropdown menu, if applicable, then select **Assigned** to list only the assays that you have selected for the run plan.
 - Click **Filter Assays By ▶ All** dropdown menu, to show all assays again, if desired.

The filtered list of assays is shown in the **Assays** step. You can select the assay of interest from the limited list and proceed through the remaining steps for the run plan.

Click  **Clear Filters** to remove all filters and view the full list of assays.

Filter the list of assays shown when you plan a run

When you plan a run in Genexus™ Software, you can apply filters to shorten the list of assays, then find the assay that you want to use in the run plan.

1. In the menu bar, click **Runs**, then click **Sample to Result**, **Nucleic Acid to Result**, **Library to Result**, or **BAM/CRAM to Result**, based on the type of run that you want to plan.
2. Enter and make the selections in the **Setup** step, if applicable, then click **Next**.
3. Select from the filter and search options in the column heading of interest.

Option	Description
List assays according to the analysis version	a. Click  (Filter) in the Analysis Version column. b. Select one or more of the versions that are shown, then click Filter.
List assays according to chip type	a. Click  (Filter) in the Chip Type column. b. Select Ion Torrent™ GX5™ Chip or Ion Torrent™ GX7™ Chip then click Filter.
List assays according to library chemistry	a. Click  (Filter) in the Library Chemistry column. b. Select the library chemistry: • AmpliSeq – The standard library preparation chemistry for targeted next-generation sequencing. • AmpliSeq HD – High sensitivity library preparation chemistry for targeted next-generation sequencing with unique molecular tags to reduce the number of false positives and improve confidence in variant calling. c. Click Filter.

4. (Optional) Click  **Clear Filters** to return to the entire list of assays.
5. In the shortened list of assays, select one or more checkboxes in the **Research Application** column for each assay that you want to add to the run, then click **Next**.
6. Click the **Filter Assays By** dropdown menu, if applicable, then select **Assigned** to list only the assays that you have selected for the run plan.
 - Click **Filter Assays By ▶ All** dropdown menu, to show all assays again, if desired.

You can proceed through the remaining steps for the run plan. For more information, see “Plan a Sample to Result run” on page 89, “Plan a Nucleic Acid to Result run” on page 97, “Plan a Library to Result run” on page 103, or “Plan a BAM/CRAM to Result run” on page 106.

No-template and positive controls

You can add a no-template control (NTC) or a positive control when you assign samples in run planning, or when you create a library batch in Genexus™ Software. The configuration of each control is based on the assay that is used. For more information, see the user guide for the panel that you use.

Manage runs

Role—System administrator, organization administrator and manager, operator.

Run plans are listed in the **Runs / Manage Runs** screen. The most recently created run is on the top of the list. Only runs that have not started are listed. After a run is started, you can monitor it from the **Dashboard**, or from the **Results / Run Results** screen.

In this screen, administrators, managers, and operators can do the following run plan tasks.

- Plan a run.
- Sort and search the list of run plans.
- Review and print a run plan.
- View the audit trail of a run plan.
- Edit a run plan.
- Delete a run plan.
- View, add, or delete the notes for a run plan.
- Abort a **Sample to Result** run after purification completes but before the run is continued on the sequencer.

View a run plan

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can view detailed run plan information in Genexus™ Software.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, click the run name of interest.
Run plan details are shown in the run plan summary pane. For more information, see “Run plan details” on page 110.
3. When you are finished viewing the run plan details, click another run name to view details for another run plan, or click **✕ (Close)** to return to the **Manage Runs** screen.
4. (Optional) Click  **Print** to print the information about the run.
To print the run plan, you must allow pop-ups in the browser.

You can click **⋮ (More Options)** to perform more actions, such as edit, add notes, or audit the run plan. The actions for each run plan are also available in the **Manage Runs** screen when you place the pointer over the row of a run name.

Run plan details

Detailed run plan information is shown in the run plan summary pane.

Run plan detail	Description
Generate Report	Indicates if a report is automatically generated for the run.
Server	The Ion Reporter™ Software account or the Genexus™ Software account and the software version that was selected for uploading sample files, in BAM or CRAM format, when the run is complete.
Run Status	The run status. For example, Planned or Purification Planned .
Run Start Date	This information is available when the run is started.
Run Completion Date	This information is available when the run is completed.
Sequencing^[1]	
Consumables Required For This Run	A list of the required consumables, for example, the chip and barcodes. The quantity of each consumable is shown in parentheses before the name of the consumable.
Consumables Installed Ready To Use	A list of consumables installed on the sequencer and available for the run.
Primer Tube Positions	An image that shows the locations of the primer pool tubes.
Sample Locations	Detailed sample information and an image that shows sample locations on the 96-well plate. For more information, see “Run summary details” on page 201.
Assays	
Assay	Detailed assay information for each assay in the run. For more information, see “Run summary details” on page 201.

^[1] The **Sequencing** section is collapsible and expandable.

Additional information for Sample to Result runs

Run plan detail	Description
Purification^[1,2]	
Consumables Required For Purification Batch x	A list of the required consumables. For example, the Quantitation Plate and Quantitation Tubes . The quantity of each consumable is shown in parentheses before the name of the consumable. Information for each purification batch is listed in a separate table.
Input Sample Locations For Purification Batch x	Detailed sample information and an image that shows sample locations on the 96-well plate. Information for each purification batch is listed in a separate table.

^[1] The **Purification** section is collapsible and expandable.

^[2] For information about purification batches, see “Plan a Sample to Result run” on page 89.

Print a run plan

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can print detailed run plan information in Genexus™ Software.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, click a run name in the list.
3. Click  **Print** to print the information about the run plan.

To print the run plan, you must allow pop-ups in the browser.

Run plan details are shown in the run plan summary pane. You can review the information that was entered or generated when you created the run. For more information, see “Run plan details” on page 110.

Edit a run plan

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can edit run plans that have a run status of **Planned** or **Purification Planned**. Parameters that can be edited include reporting options, assays used in the run, samples included in the run, chip assignment, and sample concentration.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Edit**.
3. In the **Edit Sample Run** workflow, click **Next** to navigate through the steps to change settings, then click **Save**.

Delete a run plan

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can delete run plans from the software.

You must select at least one run plan in the **Manage Runs** screen before the option to delete a run plan appears.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, select the checkbox next to one or more run names to delete.
Select all of the runs in the screen by selecting the checkbox above the column.
If needed, click **Clear** to deselect all runs.
3. Click  **Delete**.
4. In the **Delete Plan** message box, click **Yes** to delete the selected runs.

Add a note to a run plan

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can add a note or notes to a run plan that is listed in the **Runs / Manage Runs** screen in Genexus™ Software. A note is any written comment or observation that you want to add to the run.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** dialog box, do the following.
 - a. Click **+ Add Note**.
 - b. Enter the note in the **Notes** dialog box.
 - c. Click **Save**.

The note is shown in the **Notes** dialog box for the run plan.

View a run note

You can view the notes that have been added to a run plan. Run plan notes are time-stamped written notes to capture comments or observations. A run plan can contain multiple notes from different users.

You cannot edit run notes. However, you can delete notes to a run plan.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. Review the note or notes.

Delete a run plan note

You can delete a run plan note in Genexus™ Software.

Notes record any written comments or observations for a run plan.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Notes** dialog box, in the row of the note you want to delete, click  **(Delete)**.
4. Click **Close** to close the dialog box.

The run plan note is no longer listed in the **Notes** dialog box for the run plan and is removed from the software.

Review and download audit records for a run plan

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can review and download the entire history of a run plan in Genexus™ Software. An audit trail lists the records of actions for the selected run plan. Each time-stamped audit record shows the original run plan values and any new values for each action.

The following user actions generate an audit record.

- Plan a new run.
- Edit a run plan.
- Delete a run plan.

Some software processes, such as a change in run plan status also generate an audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Runs ► Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to review details for each audited action.
4. (Optional) Click  **Download** to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.

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View run progress for connected instruments

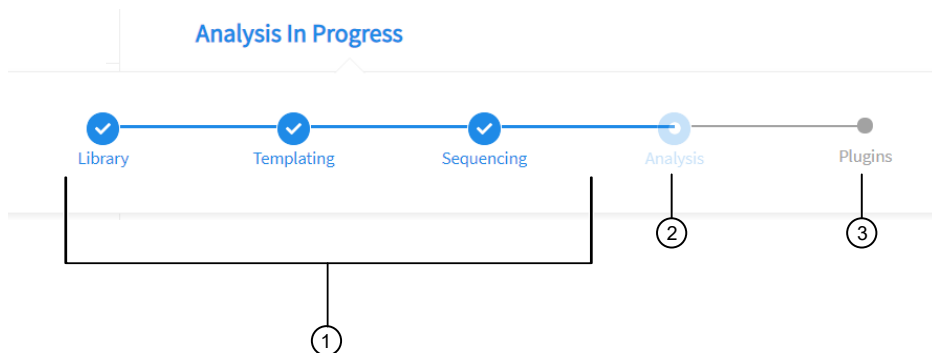
In the Genexus™ Software **Dashboard**, all users can view details for up to five recent runs for the instruments that are connected with the software.

The **Recent Runs** section shows runs and a status for each run for your organization, a link to a progress bar for the run, and a link to a summary of the run.

Runs for your organization that are reanalyzed are listed with ↻ (**Reanalysis**) after the run name. For more information, see “Reanalyze a run” on page 219.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard**, in the **Recent Runs** section, view information for recent runs.

Option	Description
View detailed run result information for completed runs.	In the left side of the screen, click a run name. The Run Summary tab opens. For more information, see “Run summary details” on page 201. In the menu bar, click Dashboard to return to the Dashboard .
View the quality control information.	The QC status for the run and for the samples is shown in the Instruments section. For more information, see “QC results” on page 124.
View assay-level results for the run.	a. In the left side of the screen, click the row for the run of interest. b. In the run details pane in the right side of the screen, click the assay name of interest. c. Review the results in the Assay Metrics tab or the Plugins tab. For more information, see “Sequencing metrics” on page 205. In the menu bar, click Dashboard to return to the Dashboard.

Option	Description
View the sample results.	a. In the left side of the screen, click the row of the run of interest. b. In the run details pane in the right side of the screen, click the sample name of interest. c. Review the Key Findings tab. For more information, see “The Key Findings tab” on page 132. In the menu bar, click Dashboard to return to the Dashboard.
View the progress of the run.	In the Results section in the right side of the screen, place the pointer over the status for the run to show the progress bar.  ① Completed steps in the run are shown with a check mark. In this example, Library preparation, Templating, and Sequencing are complete. ② Steps that are in progress are shown with a flashing circle. In this example, Analysis is in progress. ③ Steps that remain to be completed are shown in gray. In this example, the Plugins step remains to be started after Analysis is complete. If any failures occur, failed steps are also shown in gray.

Tip: If the recent run includes many samples, limit the samples that are listed. For more information, see “Filter the list of samples shown in the Dashboard” on page 116.

Filter the list of samples shown in the Dashboard

You can apply filters to shorten the list of samples shown in the **Dashboard** in Genexus™ Software.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard**, in the **Recent Runs** section, click the row of the run of interest.
Assays and samples in the run are listed in the details pane in the right side of the screen. In addition, the QC status and number of samples that passed or failed QC are shown.
3. In the details pane, filter the list of samples.

Option	Description
Shorten the list of samples.	Click  (Filter), select the samples of interest, scroll to the bottom of the filter dialog box, then click Filter .  (Filter) changes color to indicate that a filter is applied.
Refine the samples shown by the QC status of the sample result.	In the details pane, in the QC Status section, click Pass to list only samples that passed QC metrics or click Fail to list only samples that failed QC metrics.

The list of samples is filtered according to the selections.

Click  **Clear Filters** to remove all filters and view the full list of samples.

View the status of an instrument

In the **Dashboard**, when a run is in progress, you can view the status of the Genexus™ Integrated Sequencer and any Genexus™ Purification System that are connected with Genexus™ Software. You can see if a run is in progress, and view details of the run, except for the run name if the run is not within your organization.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard**, in the **Instruments** section, view the status of an instrument.

The following information is provided for all connected instruments.

- Instrument name
- Run status of the current or most recent run
- PQ status—Indicates whether the performance qualification (PQ) status of the instrument is **Verified**, **Not Verified** or **Expired**.
-  (**Alarms**)—Instrument alarms, if any
If the instrument displays one or more alarms, the number of alarms is shown in parentheses. You can place the pointer over **Alarms** to view more information about an alarm.

3. If a run is in progress, view the details of the run. In the **Instruments** section, in an instrument pane, click **Details**.

Tip: Check the status of a purification instrument for **Run completed waiting user input** to know when you must perform an action to proceed through the run.

4. Click  **Refresh** to update the information in the **Instruments** section.

Information about connected instruments

When a run is in progress, information about the instruments that are connected to Genexus™ Software is available in the **Dashboard**. You can expand the **Details** section for each instrument to view more information about a run in progress.

Item	Description
Run	The name of the run that is in progress or the last completed run on the instrument. A string of asterisks (*) for the run name indicates that the instrument is in use by another organization.
Serial	The serial number of the instrument.
Operator	The name of the operator who started the run on the instrument.
Started	The date and time that the run started on the instrument.
Completed	The date and time that the run completed on the instrument.
Elapsed Time	The total elapsed time of the run, from run start to finish.
PQ Status Not Verified	This message appears if PQ has not been performed.
Last Init	The last initialization completion date and time.
Next PQ Due	The expiration date of the current PQ verification run. Expiration of the PQ status prevents the instrument from performing a non-PQ run until a valid PQ run has been performed and signed off.

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Results navigation overview

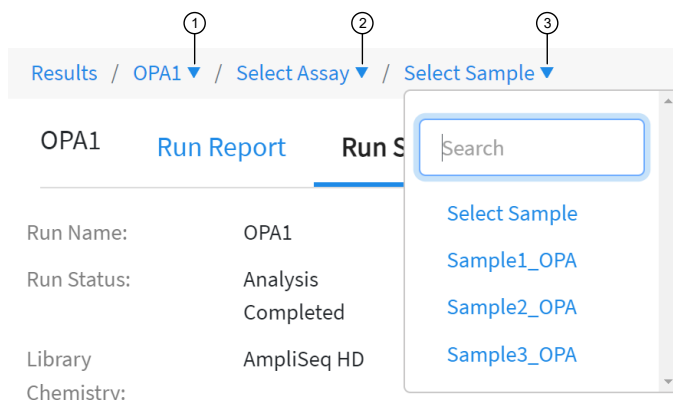
Use the **Results** menu on the menu bar to review results and data analysis, and to perform data management tasks.

Option	Description
Click Results ▶ Sample Results	Select this option to review completed sample results and reports.
Click Results ▶ Run Results	Select this option to review completed run results and reports by assay.
Click Results ▶ Purification Only Results	Select this option to review results for purification of samples on a Genexus™ Purification System that is connected to Genexus™ Software.
Click Results ▶ Verification Results	Select this option to review data from completed verification runs that were performed during sequencer installation or performance qualification.

After you select a run result or a sample result, a results navigation bar lets you toggle between different runs, and assays and sample results for each run. In addition, you can find samples or assays in the run with the search field that appears in each dropdown list.

Assay-level results include assay metrics, such as final read data.

Each sample has a run report. Samples that are analyzed with the same assay during the same sequencing run are included in the same run report. All other results are specific to the sample and are shown in the sample-level results.



Callout	Dropdown list	Description
1	Run name	The run name is listed. Multiple runs are listed only if the run has been reanalyzed. For more information, see “Reanalyze a run” on page 219.
2	Select Assay	Click an assay name to view assay metrics for the run. Search for or toggle between different assays available for the run. Only assays that are used in the run are shown in the list. Some results from a plugin that are included in the run, such as the CustomerSupportArchive plugin, apply to all samples for an assay in a run. Plugin results contain a consolidation of all samples. Results from such assay-level plugins are shown in the Plugins tab when you select an assay without a selection of samples for the run. For more information about plugins, see Chapter 13, “Plugins for data analysis”. Select Select Assay from the list to remove the assay choice. If you remove the assay and sample selections, the Run Summary tab opens with a summary of results for the run that is selected in the Run Results list. Assay metrics are also shown in the Run Report tab. For more information, see “Sequencing metrics” on page 205.
3	Select Sample	Click a sample name to view the sequencing results for the sample. You can click a different sample name to view other sample results for other samples in the run for the assay that is selected in the Select Assay list. Use the search field to find samples in the run. For Sample to Result runs that include a positive control, results for the positive control are shown when you select Control Sample. Select Select Sample from the list to remove the sample choice. If you remove the sample selection, the Assay metrics tab for the selected assay opens. If you select Select Assay with Select Sample shown, the Run Summary tab opens. Assay metrics are also shown in the Run Report tab. For more information, see “Sequencing metrics” on page 205.

Review sample results

You can review the list of sample results, and open detailed results for each sample in the **Results / Sample Results** screen. Samples that have been sequenced are listed by sample name.

See “Find sample results” on page 128 for information about how to find sample results of interest.

The following information is shown in the **Sample Results** screen. You can add other columns for sample attributes to the screen, and then view more information. For example, you can add a column for **Electronic Signatures** to view signature and lock information for variant reports. For more information, see “Change the columns in the sample results table” on page 155.

Column	Description
Sample Name	The unique identifier created when the sample was entered into the software. Click the Sample Name to open the Sample Results screen for the sample.
Assay Name	The name of the assay used in the run plan.
Run Name	The unique name of the run given when it was created in the software.
Sample Status	The status of the run or sample (for example: Completed , Running , Failed , Terminated , Pending , Stalled).
QC Status	The QC status of a completed run. ✓ (Passed) indicates the sample passed all QC metrics. ✗ (Failed) indicates the sample failed a QC metric. — (Not Calculated) indicates a sample did not undergo QC analysis.
Started On	The date and time when the run was started.
Updated On	The date and time when the last action was completed on the run.
Tags	Tags that are assigned to the sample are listed.
Updated Annotations	The information that is shown depends on the type of the assay and whether the option to use the latest annotation is configured in the assay. - The name and version of the annotation source that is applied to the sample result is shown when an assay is configured to use the latest annotations but the annotation that is applied to the results is not the latest annotation source. - No information is shown in the column (the value is blank) when the latest annotation source version is applied to the result, regardless of whether the assay is configured to use the latest annotation source. - No information is shown in the column (the value is blank) when an assay is configured to not use the latest annotation source, and instead uses a fixed version of an annotation source. N/A is shown when an assay does not use an annotation set.

You can also perform the following actions in the **Sample Results** screen.

Option	Description
Actions	Available action links for a sample are shown when you place the pointer over the row of a sample. • Report—Download the report (available only for samples with a sample status of completed). For more information, see “Variant reports” on page 225. • Assign Tags—Add or remove the tags assigned to a sample result. For more information, see “Manage sample tags” on page 69. • Audit—View the audit trail for the run. For more information, see “Review and download audit records for a run plan” on page 113. • Notes—View, add, or delete notes for sample result. • CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see “Download a Customer Support Archive” on page 586. • Variant Audit—View the history of variant classifications, notes, and changes to a variant. • Reanalyze—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step. For more information, see “Reanalyze a sample” on page 221. • Download Files—Select files to download in a ZIP folder to your computer. You can choose from sequencing files, variant files, troubleshooting file, reports, and plugin files.
Actions that you can perform when you select samples	More actions are shown above the Sample Results table when you select one or more samples. • Assign Tags—Assign a tag or tags to selected sample results in one action. The  Assign Tags command appears above the Sample Results table after you select one or more samples. For more information, see “Assign sample tags to multiple sample results” on page 123. • Compare—Compare variant results between multiple samples, or from a single sample over time. The  Compare command is not shown until you select at least two samples. For more information, see “Compare sample results” on page 184.

View sample results

Any user can view sample results, including detailed information about variants and fusion calls, and key findings for the samples. You can also view QC metrics, results from plugins that were used during the sequencing run, and reports and report templates that are associated with the sequencing run.

1. Click **Results ▶ Sample Results**.
2. In the **Sample Name** column, click a sample name.
The sample results open to the **Key Findings** tab.
3. In the sample results, you can click to view information on each of the following tabs.

Tab	Description
QC	The quality metrics for the sample or samples that were sequenced in the run. For more information, see “QC results” on page 124.
Key Findings	An overview of the results for the sample, including Sample Details , Key Metrics , Genes , and Coverage graphs. For more information, see “The Key Findings tab” on page 132.
Variants	Detailed variant results for SNVs/Indels , Fusions , and CNVs . For more information, see “The Variants tab” on page 139.
Plugins	Results generated from the plugins associated with the assay used to analyze the sequenced sample. For more information, see Chapter 13, “Plugins for data analysis”.
Reports	You can download and generate summaries of run results. There are two types of reports: • Run reports—Include detailed information about the sequencing run, including sequencing metrics, information about read data for the samples, and data about well usage for the chip used in the run. • Variant reports—Include the variant results, key findings, quality metrics, and results for plugins that were used in the sequencing run for each sample in the sequencing run. For more information, see “Reports overview” on page 223.

Sample tags in sample results

Role—System administrator, organization administrator and manager, operator.

You can assign tags to sample results in Genexus™ Software. Tags are used to label and group sample results. A tag can be assigned to a single sample or to multiple samples at once.

Assign sample tags to a sample result

Role—System administrator, organization administrator and manager, operator.

You can assign tags to sample results in Genexus™ Software to label and group sample results. For more information, see “Manage sample tags” on page 69.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of the sample result that you want to tag, then click **Assign Tags**.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample does not exist in the software, an administrator or manager can create a new sample tag. For more information, see “Create a sample tag” on page 69.
4. Select one or more tags that you want to assign to the sample result, then click **Save**.

The tag or tags are assigned to the sample result and are listed in the row for the sample result in the list of sample results.

Assign sample tags to multiple sample results

Role—System administrator, organization administrator and manager, operator.

You can assign one or more tags to multiple sample results in Genexus™ Software. For more information about sample tags, see “Manage sample tags” on page 69.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click the checkbox to the left of each sample result of interest, then click  **Assign Tags** above the list of sample results.
 **Assign Tags** is not shown until you select at least one sample result.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample result does not exist in the software, an administrator or manager can create a new sample tag. For more information, see “Create a sample tag” on page 69.
4. Select one or more tags that you want to assign to the sample results, then click **Save**.

The tag or tags are assigned to the selected sample results and are listed in the row for each sample result in the list of sample results.

View the QC results

Any user can view the quality control (QC) metrics for each sample that was sequenced in a run in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.
3. Click the **QC** tab.
The QC metrics are shown.
4. Review the QC metrics.
For more information, see “QC results” on page 124.

QC results

The quality control (QC) metrics for each sample that was sequenced in a run are displayed in Genexus™ Software.

If a sample fails a single test metric, the sample fails that QC test. A sample must meet all QC parameter thresholds of a particular QC test to pass. The QC status is divided into the following categories.

-  (Passed) indicates the sample passed all QC metrics.
-  (Failed) indicates the sample failed a QC metric.
- — (Not Calculated) indicates a sample did not undergo QC analysis.

Note: If a sample fails to meet one or more QC parameters, you can reanalyze a run after adjusting the parameter or parameters. For more information, see “Reanalyze a run” on page 219.

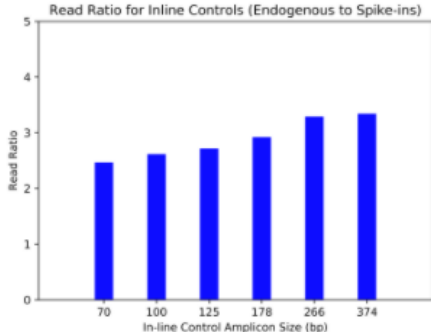
The data shown in the screen depend on the assay that was used in the run. Not all metrics are shown for every assay.

Metric	Description
Purification QC	Quality control information for nucleic acid extraction that is performed on a Genexus™ Purification System.
Sample Concentration RNA	The concentration of extracted RNA after purification.
Sample Concentration DNA	The concentration of extracted DNA after purification.
Sample Concentration TNA	The concentration of extracted TNA after purification.
Run QC	General run quality control information.
Final Reads	Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement.

(continued)

Metric	Description
Key Signal	The average signal after software processing for library ISPs that identically match the library key (TCAG).
Key Signal – Resequencing	The average signal after software processing for library ISPs that identically match the library key (TCAG) for the resequencing run. This metric is only shown for assays that include resequencing.
Percent Loading	The number of wells with ISPs divided by the number of the total addressable wells in a run.
Percent Loading – Resequencing	The number of wells with ISPs divided by the number of the total addressable wells in the resequencing run. This metric is shown only for assays that include resequencing.
Raw Read Accuracy	The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as $100 \times (1 - (\text{sum}(\text{per base error}) / \text{sum}(\text{per base depth})))$.
Templating QC—CF-1 Control	Sequencing quality metrics of the control fragment. These metrics indicate templating success.
Average Reads Per Lane	The number of CF-1 reads divided by the number of chip lanes used in the run.
Base Call Accuracy	The probability that a given base is called correctly. $1 - (\text{total number of errors for all positions in CF-1}) / (\text{total number of CF-1 base reads})$
Mean AQ20 Read Length (bp)	Average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for CF-1 reads.
Sample QC—DNA^[1]	Sequencing quality metrics of the sample DNA library.
Mapped Reads	The total valid mapped reads.
Mean AQ20 Read Length (bp)	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for all aligned reads of a library.
Mean Read Cov	The number of average reads per amplicon.
Molecular Uniformity	Uniformity of molecular coverage for all amplicons. This metric is applicable to Ion AmpliSeq™ HD library chemistry only.
Mean Read Length (bp)	The average length, in base pairs, of final library reads for the sample.
Uniformity of Amplicon Coverage	The percentage of amplicons that had at least 20 percent of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts.
Median Mol Cov	The median number of functional molecule reads per amplicon calculated over all amplicons in the assay. This metric is applicable to Ion AmpliSeq™ HD library chemistry only.

(continued)

Metric	Description
Uniformity of Base Coverage	The percentage of reads with a depth of coverage $\geq 20\%$ of the mean read coverage at each position. This metric is applicable to Ion AmpliSeq™ HD library chemistry only.
Read Length Histogram	The histogram presents all filtered and trimmed DNA library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences.
Read Ratio for Inline Controls (Endogenous to Spike-ins)	The ratio of genomic sample reads to control reads for each inline control amplicon.  Note: The Read Ratio for each inline control amplicon is expected to be approximately 3 with 10 ng DNA input for both Ion AmpliSeq™ and Ion AmpliSeq™ HD chemistries.
Sample QC – RNA [2]	Sequencing quality metrics of the sample RNA library.
Mapped Fusion Reads	The total valid mapped reads. That is, the mapped reads that pass the thresholds and filters across all of the RNA targets in the reference file. This number is a subset of the mapped reads shown in the Run Samples table in the Assay Metrics.
Mean Read Length (bp)	The average length, in base pairs, of the final library reads for the sample.
RNA Expression Ctrls Detected	The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. Fusion panels include primer pairs that cover control housekeeping genes.
Mean AQ20 Read Length	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for all aligned reads of a library.
Read Length Histogram	The histogram presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).

(continued)

Metric	Description
Read Ratio for Inline Controls (Endogenous to Spike-ins)	The ratio of genomic sample reads to control reads for each inline control amplicon. You can use this metric to measure the level of contaminating genomic DNA in RNA libraries when you select the RNA checkbox for Include Inline Controls in assay set up. Using inline controls in RNA assays will reduce the total number of panel reads. Note: The Read Ratio for each inline control amplicon is expected to be approximately 3 with 10 ng DNA input for both Ion AmpliSeq™ and Ion AmpliSeq™ HD chemistries.
NTC QC – DNA^[3]	DNA Sequencing quality metrics of the no-template control.
Average Base Coverage Depth	The average number of DNA reads of all targeted reference bases.
Mean Read Length (bp)	The average length, in base pairs, of final DNA library reads for the no template control.
Read Length Histogram	The histogram presents all filtered and trimmed reads for the no template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).
Read Ratio for Inline Controls (Endogenous to Spike-ins)	The ratio of no template control sample reads to control reads for each DNA inline control amplicon.
NTC QC – RNA^[3]	RNA Sequencing quality metrics of the no template control.
Mapped Fusion Reads	The total number of valid mapped reads for the NTC.
Mean Read Length (bp)	The average length, in base pairs, of final RNA library reads for the no template control.
Read Length Histogram	The histogram presents all filtered and trimmed reads for the no template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).
Read Ratio for Inline Controls (Endogenous to Spike-ins)	The ratio of no template control sample reads to control reads for each RNA inline control amplicon.

^[1] For BAM/CRAM to Result runs, only the **Sample QC—DNA** metrics are shown.

^[2] For BAM/CRAM to Result runs, only the **Sample QC—RNA** metrics are shown.

^[3] Metrics are shown only if NTC is included in the run plan.

MAPD copy number QC metric

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on Aneuploidy run results and other runs that detect CNVs.

MAPD is one of the metrics that we define to assess whether the panel data are useful for copy number variation (CNV) run results.

MAPD is defined as the Median of the Absolute values of all Pairwise Differences between log2 ratios per tile for a given run. Tiles roughly correspond to amplicons in an Ion AmpliSeq™ assay. Each pair is defined as adjacent in terms of genomic distance. Tiles corresponding to copy number amplicons and other amplicons are being treated equally as no differences in variability are seen between these types. Then, any two log2 ratios that are adjacent on the genome are a pair. Except at the beginning and the end of a chromosome, every log2 ratio belongs to two pairs.

Formally, if x_i is the log2 ratio at position i , with i ordered by genomic position:

```
MAPD = median ( | x(i - 1) - x(i) | )
```

MAPD is a per-sequencing run estimate of copy number variability, similar to standard deviation (SD). If one assumes the log2 ratios are distributed normally with mean 0 and a constant SD, then MAPD/0.95 is approximately equal to SD. However, unlike SD, using MAPD is robust against high biological variability in log2 ratios induced by known conditions such as cancer.

Regardless of the source of the variability, increased variability decreases the quality of CNV calls.

Find sample results

Role—System administrator, organization administrator and manager, operator.

You can search, filter, and sort information to narrow the sample results that are listed in the **Results / Sample Results** screen. The sort, search, and filter tools help you organize sample results and find a specific sample result in Genexus™ Software.

To sort the list of sample results, see “Sort the list of sample results” on page 132

To search for a sample result by a unique identifier, such as the name of a run or the name of a sample, see “Search for sample results” on page 129.

To filter sample results by a common trait, such as QC status, see “Filter the list of sample results” on page 130.

Search for sample results

Sample results are listed in the **Results / Sample Results** screen in Genexus™ Software. The most recent sample result is on the top of the list.

You can search for sample results with keywords or text that are included in the name of the sample, run name, or the name of an assay applied to the run.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. In the menu bar, click **Results ▶ Sample Results**.
 2. In the **Sample Results** screen, in the **Sample Name**, **Run Name**, or **Assay Name** column heading, click  (**Filter**).
 - a. In the search field, enter a full or partial assay, sample, or run name of at least 3 characters. When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - b. In the filter dialog box, select from the limited list.
 - Select a checkbox next to a sample, run, or assay name to select one or more items.
 - Click **Select All** to select all items listed in the filter dialog box.
-

Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

- c. In the filter dialog box, click **Filter**. In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of sample results is shown in the **Sample Results** screen.

Click  **Clear Filters** to remove all filters and view the full list of sample results.

Filter the list of sample results

You can apply filters to narrow search results or shorten the list of sample results when you view the list of sample results in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, select one or more filters.

Option	Description
Filter and search by the name of the sample, the run name or the assay selected in the run.	For more information, see “Search for sample results” on page 129.
Show or hide sample results that are archived	In the dropdown list in the upper left corner, select an archive status. • Archived • Non Archived Select All to list all sample results.
Filter by the status of the sample result.	1. In the Sample Status column heading, click ▼ (Filter) then select at least one status. • Completed • Running • Failed • Terminated • Stalled • Aborted • Pending 2. Click Filter. 3. Records are listed for sample results that meet the sample status criteria.
Filter by the QC status.	1. In the QC Status column heading, click ▼ (Filter) then select at least one QC status. • Failed • Passed • Not Calculated 2. Click Filter. Records are listed for sample results that meet the QC status criteria.
List only sample results from runs that were started on or updated on an exact date.	1. In the Started On or Updated On column heading, click ▼ (Filter). 2. In the first dropdown list, select Is equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. Records are listed for sample results from runs that were started on or updated on the date entered.

(continued)

Option	Description
List only sample results from runs that were started on or updated within a date range.	1. In the Started On or Updated On column heading, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date of the range. 4. In the Boolean operator dropdown list, select And. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date of the range. 7. Click Filter. Records are listed for sample results from runs started on or updated on dates that meet both conditions.
List sample results according to a date that meets either one condition or a second condition	1. In the Started On or Updated On column heading, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date for the sample results of interest. 4. In the Boolean operator dropdown list, select Or. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date for the sample results of interest. 7. Click Filter. Records are listed for sample results from runs that were started on or updated on dates that meet at least one of the conditions.
List only sample results for runs that were started on or before or updated on or before a date that you indicate.	1. In the Started On or Updated On column heading, click  (Filter). 2. In the first dropdown list, select Is before or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. Records are listed for sample results from runs that were started on or before the date entered.
List only sample results for runs that were started on or after or updated on or after a date that you indicate.	1. In the Started On or Updated On column heading, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. Records are listed for sample results from runs that were started on or after, or updated on or after, the date entered.

In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

Click  **Clear Filters** to remove all filters and view the full list of sample results.

Sort the list of sample results

You can sort the list of sample results to make it easier to find sample results in Genexus™ Software.

By default, sample results are listed in the **Results / Sample Results** screen by date. The most recent sample result is at the top of the list.

You can change the order that the sample results are listed in the table.

For example, you can sort the **Call** column alphabetically so that the **Absent** calls appear first. This allows you to easily view hotspots that did not pass thresholds for a variant call, such as when the allele fraction is zero.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click a column heading of interest.
The list of sample results is sorted in alphabetical or chronological order depending on the entries in the column.
3. Click the column heading again to reverse the order.

The Key Findings tab

The **Key Findings** tab shows details about the sample, a summary of key metrics for the run, and key variants for genes assayed in the run. The information that is displayed depends on the assay that was used in the run.

The **Key Findings** tab provides a color-coded visual representation of the variant results. You can change the filters that are applied to expand or restrict the list of genes that are shown. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.

Each tile in the **Key Findings** section can represent one gene and indicate whether a variant is detected. Or, if multiple variants are detected in a gene, the gene is represented with multiple tiles. When there are multiple genes that are annotated for the same variant, then only a single tile with a single gene name is shown. The gene that is shown is based on the severity of the impact for the variant.

When there are multiple genes that are annotated for the same variant, then only a single tile with a single gene name is shown. The gene that is shown is based on the severity of the impact of the variant.

View sample results in the Key Findings tab

Any user can view sample results in the **Key Findings** tab. The **Key Findings** tab is by default the first view that is shown for sample results.

You can view a color-coded visual summary of variant results, and metrics, complex biomarkers and coverage graphs for sample results in Genexus™ Software, and add filters to refine the genes that are shown.

The first time that you view variant results in the **Key Findings** tab, the filter chain and gene list that is associated with the assay, if any, are applied to the results. You can change the filter chain and gene list in the **Variants** section. The variants that are shown depend on the filter chain and the gene list that are applied. You can change the filter chain or gene list that is applied to the variants that are shown.

1. In the menu bar, click **Results ▶ Sample Results**.

2. Click a sample name.

The **Results** screen opens to the **Key Findings** tab, which shows the **Sample Details**, **Key Metrics**, **Variants** and **Coverage Graphs** for the sample results.

3. In the **Sample Details** section, review the sample details.

Sample details

Item	Description
Sample Name	A unique identifier for the sample.
Collection Date	The date that the sample was collected.
Gender	The biological sex of the sample: Female , Male , or Unknown .
Sample Type	A term that describes the sample, for example, FFPE, DNA, DNA & RNA.
Application Category	The application type of the sample.
Cancer Type	The type of cancer that is represented by the sample.
Cancer Stage	The stage of the cancer from which the sample was collected.
%Cellularity	The percentage of tumor cellularity in the sample. This is a whole number between 1 and 100. The %Cellularity attribute is entered when a sample is created. This attribute applies only to FFPE samples.
Case ID	A unique identifier for the research experiment.

4. In the **Key Metrics** section, select a view of the data, then review **General Key Metrics**.

General Key Metrics

Item	Description
Target Coverage^[1]	
Target base coverage at 100x	The percentage of reference genome bases covered by at least <i>n</i> reads.
Target base coverage at 350x	The percentage of reference genome bases covered by at least <i>n</i> reads.
Amplicon Summary	
Average Base Coverage Depth	The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage.
Uniformity Of Base Coverage	The percentage of bases in all targeted regions (or whole genome) with a depth of coverage $\geq 20\%$ of the mean read coverage. Cumulative coverage is linearly interpolated between the nearest integer base read depths.
Percent Reads On Target	The percentage of filtered reads that are mapped to any targeted region relative to all reads mapped to the reference. A read is considered on target if at least one aligned base overlaps at least one target region. If no target regions (file) was specified, this value is the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified.
Aneuploidy Score	The total number of copy number altered chromosome arms.
Cellularity	
Calculated Tumor Cellularity	The percentage of tumor content calculated from the sample.
Tumor Mutational Burden	
TMB (Mutations/Mb)	The estimated tumor mutational burden, or a measurement of the frequency of mutations in specific tumor genes, in a given sample.
TMB Status	High, Intermediate, or Low. Thresholds to determine the TMB Status are set with the TMB-Low Threshold and TMB-High Threshold in the Annotation section of the Parameters step when you create an assay. For more information, see the software help system. A classification status of Threshold not set indicates that either the TMB classification thresholds settings were not defined. Note: Threshold values are not provided in system-installed assays. The software always reports the Classification as Undefined unless a manager- or administrator-level user sets the threshold the assay.

General Key Metrics (continued)

Item	Description
Average Coverage	The following formula is used to calculate average coverage: $\frac{\text{Total base coverage across the genomic positions defined by the assay BED file}}{\text{Number of bases in the DNA BED file}}$ If the average coverage is below 150, the analysis completes successfully but Tumor Mutational Burden is reported as -1. Note: This setting cannot be changed in Genexus™ Software.
Bases Used in TMB Calculation	The number of bases used as a denominator for the tumor mutational burden calculation. Only bases with sufficient base coverage are used in the calculation, as defined in the assay. When you create an assay, you can select exonic + intronic regions covered by the panel to be used instead of exonic regions. The TMB Variant Region Type setting is in the Annotation section of the Parameters step. For more information, see the software help system. You can set the number of bases in the Sample QC - DNA section of the QC step when you create an assay. For more information, see the software help system.
Deamination Score	The estimated SNV proportion consistent with deamination (low allele frequency C:G>T:A SNVs). The deamination score can be used to determine the quality of FFPE samples. A deamination QC status of PASS/FAIL is also reported. The threshold for this QC can be adjusted when you create an assay. The Deamination QC Threshold setting is in the Annotation section of the Parameters step. For more information, see the software help system.

[1] Metrics are shown only for analyses that run the **coverageAnalysis** plugin.

5. (Optional) You can filter the selected variants that are shown in the **Key Findings** tab. Changes to these filters affect all of the views shown in the tab. We recommend that you filter data in the following order.
 - a. Change the filter chain or a gene list that is applied to the results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.
 - b. Select a subset of variants that are shown in the **Key Findings** tab. Click a checkbox to deselect the variants that you want to exclude from reports or from an exported **Selected Variants TSV** file.
 - c. Review the **Selected Variants** count at the top of the view.

IMPORTANT! If some variant rows that were selected with the checkboxes in the table rows, are hidden in the screen with column filtering, those variants are still included in the **Selected Variants** count at the top of the table. If you then download a **Selected Variants TSV** file, the downloaded file includes only the selected variants that are shown in the screen. That is, the number of variants that is shown in the **Selected Variants** count may be different from what is downloaded in the **Selected Variants TSV** file.

- Review information for the variant. Each blue tile in the **Variants** section represents a variant and summarizes information for the variant. For more information, see “Gene tiles in the Key Findings tab” on page 138.
- In the **Coverage Graphs** section, below the graphs that are shown, in **Show Coverage for Gene**, select a gene to show a coverage graph for any gene that is shown.
By default, coverage graphs are shown for the first five genes that are shown in the tiles in the **Variants** section.
A new coverage is shown for the selected gene.

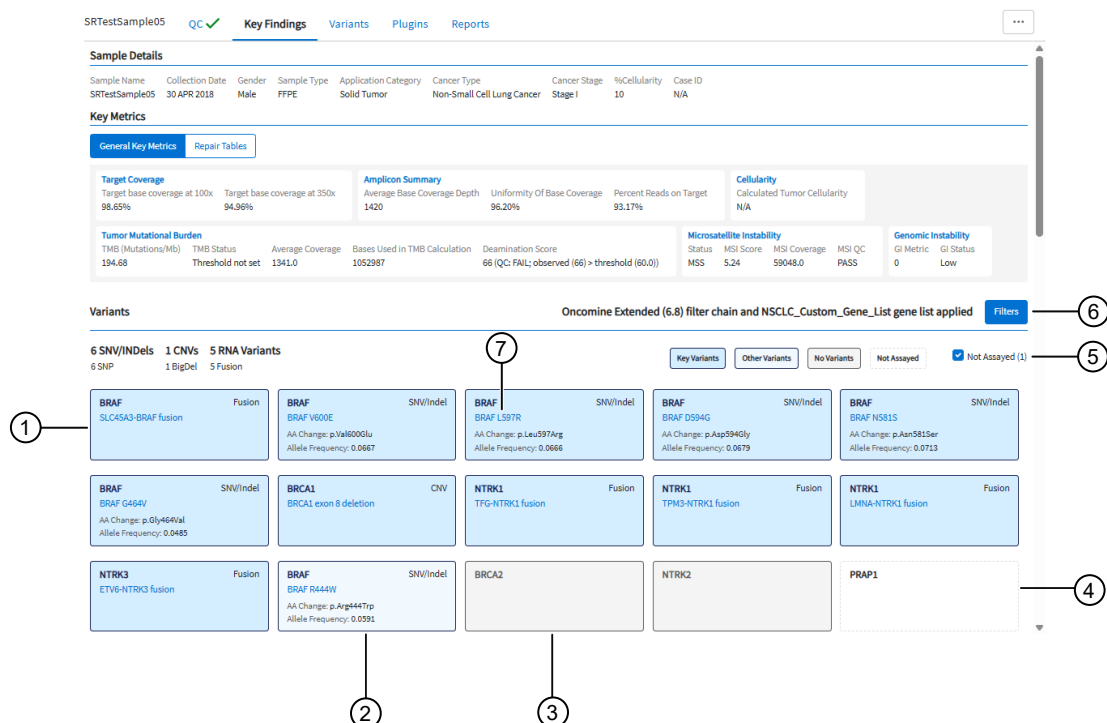


Figure 1 Results in the Key Variants tab

- Key Variants**—Key variants are annotated by the OncoPrint™ Variant Annotator. Key variants are listed in the **Variants** tab, with a value of **Yes** in the **Key Variant** column. Gene tiles are shown based on the filter chain and gene list selections.
- Other Variants**—Variants that are not key variants are listed in the **Variants** tab, with a value of **No** in the **Key Variant** column. Gene tiles are shown based on the filter chain and gene list selections.
- No Variants**—A gene is present in the gene list but there are no called variants based on the filter chain and gene list selections.
- Not assayed**—A gene is present in the gene list, but is not included in the panel used in the assay.
- Not Assayed** checkbox to hide or show genes that are not assayed.
- Filters** allow you to select and apply an available filter chain and gene list to the results. When a gene list or filter chain is changed, the genes and variants that appear in the **Variants** section reflect the selected gene list or filter chain.
- Variant Name**—The name of the variant. The **Variant Name** is a hyperlink to the pileup for the variant in the **Variants** tab.

View the amplicon coverage

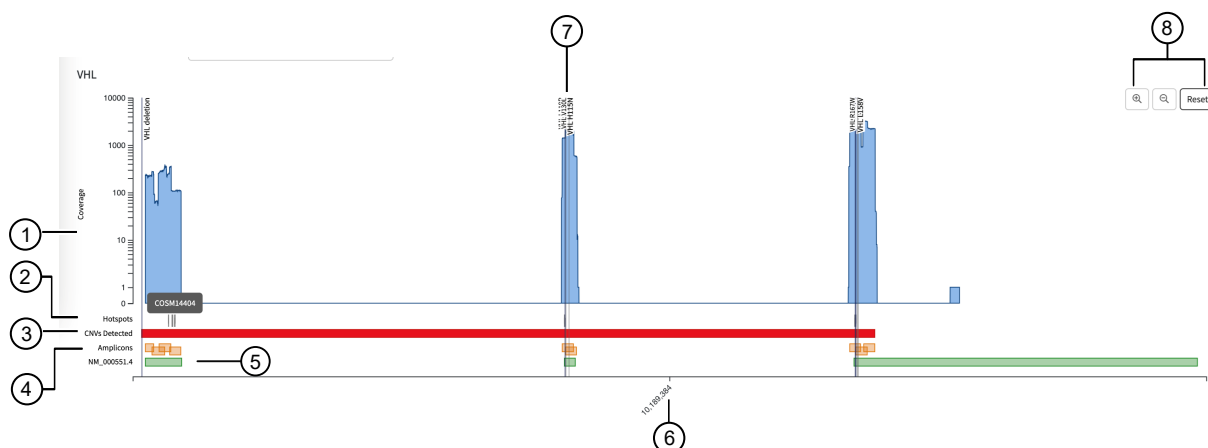
You can view DNA exon amplicon coverage in Genexus™ Software to help you determine whether the sequencing reads across a gene are uniform and sufficient. The default genes that are shown are determined by the gene list that is designated as default for the cancer type of the sample. You can also view amplicon coverage graphs for more genes. These amplicon coverage graphs provide a high-level overview of coverage. More detailed coverage information is also available in the software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Key Findings** tab in the **Results** screen.
3. Scroll to the amplicon **Coverage Graphs**.
4. Review the coverage graphs.
5. To show coverage graphs for more genes, scroll to the bottom of the screen. Click **Show Coverage for Gene**, then select the gene of interest.
6. You can adjust the amplicon coverage graph with the pointer and the buttons in the upper right corner of each graph. The coverage graphs change to show coverage for the selected gene.

Action	Procedure
Zoom in on a region of interest.	Click  one or multiple times.
Zoom out for an expanded view.	Click  one or multiple times.
Revert to the default view.	• Click  one or multiple times. • Click Reset.
Move the image left or right in the screen.	After you zoom in, click-drag at any position in the image.

Example amplicon coverage graph

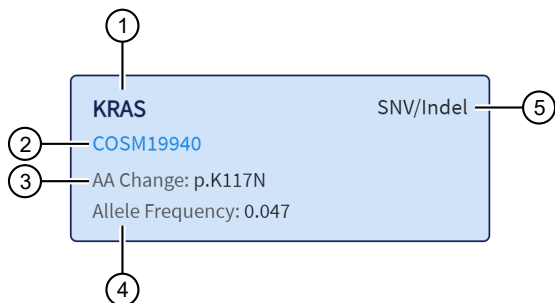
This example illustrates an amplicon coverage graph for the *VHL* gene.



- ① Base coverage is shown on the y-axis on a logarithmic scale.
- ② The location of the known hotspots is denoted with gray bars. You can place the pointer over the hotspots that are represented with gray lines to view the hotspot name.
- ③ The CNVs detected location and span are denoted with red or blue bars. GAIN is represented by blue bars and LOSS by red bars. Hover over a bar to view the details.
- ④ The location and span of the amplicons is denoted with orange bars. Place the pointer over an orange bar to view the amplicon name.
- ⑤ The location and span of the transcript track is denoted with green bars. Place the pointer over the green bar to view the exon number.
- ⑥ The genome coordinate position is shown on the x-axis. The line along the x-axis may be broken to indicate that the graph is discontinuous to show exons.
- ⑦ Called variants are indicated with a vertical line above the blue coverage plot. The variants noted in the graph depend on the filter chain applied to the results in the **Key Findings** tab. For more information, see “The Variants tab” on page 139.
- ⑧ You can adjust the amplicon coverage graph. For more information, see “View the amplicon coverage” on page 137.

Gene tiles in the Key Findings tab

Each blue tile in the **Key Findings** tab represents a variant and summarizes information for the variant.



- ① The gene name. When there are multiple genes that are annotated for the same variant, then only a single tile with a single gene name is shown. The gene that is shown is based on the severity of the impact for the variant.
- ② The **Variant ID** links to the variant details in the **Variants** tab.
- ③ The amino acid change using Human Genome Variation Society (HGVS) nomenclature.
- ④ The number of variant read counts divided by the total number of read counts for the sample.
- ⑤ The variant type.

The Variants tab

Any user can view detailed variant results in the **Variants** tab.

The variants that are shown depend on the filter chain and the gene list that are applied. The first time that you view sample results in the **Variants** tab, the filter chain and gene list that is associated with the assay, if any, are applied to the results. You can change the filter chain or gene list that is applied to the variants that are shown in the results. You can further filter data that is shown on the screen with a filter (▼ **(Filter)**) at the top of the column. You can also select or deselect the checkboxes in the list of variants to filter the variants that are included in reports and results files.

You can export the information for all variants that are included in sample results, or for selected variants, in tab-separated value (TSV) format.

You can toggle between different views of the sample results and limit the types of variants that are shown: **ALL**, **SNVs/Indels**, **Fusions**, and **CNVs**.

Variant Tables

You can toggle between different views of the sample results and also limit the types of variants that are shown.

Variant type	Description
ALL	Calls and other information for all variant types that are analyzed for each sample in a sequencing run.
SNVs/Indels	SNVs: missense and nonsense single-nucleotide variants. Multi-nucleotide variants are also included. Indels: insertion and deletion variants. For more information, see “View results for SNVs/INDELs” on page 141.
Fusions	Translocations of genetic material. For more information, see “View results for fusions” on page 146.
CNVs	Copy number variations (CNVs) are variations of the number of copies of a given gene. For more information, see “View results for CNVs” on page 152.

View all variant results

The **ALL** view in the **Variants** tab lists the calls and other information for all variants that are analyzed in each sample in a run.

Any user can view detailed variant results in the **Variants** tab.

The variants that are shown depend on the filter chain and the gene list that are applied. The first time that you view sample results in the **Variants** tab, the filter chain and gene list that is associated with the assay, if any, are applied to the results. You can change the filter chain or gene list that is applied to the variants that are shown in the results. You can further filter data that is shown on the screen with a ▼ **(Filter)** at the top of the column. You can also select or deselect the checkboxes in the list of variants to filter the variants that are included in reports and results files.

You can export the information for all variants, or for variants that are selected, in TSV format. The file that is exported reflects the rows of variants that are shown in the screen, and is based on the filters that are applied.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Ensure that **ALL** is selected to open the **ALL** view of the results.
To narrow the view to SNV/INDELs, Fusions, or CNVs, see “View results for SNVs/INDELs” on page 141, “View results for fusions” on page 146, or “View results for CNVs” on page 152.
5. (Optional) You can filter the selected variants that are shown in the **Variants** tab. Changes to these filters affect all of the views shown in the tab. We recommend that you filter data in the following order.
 - a. Change the filter chain or a gene list that is applied to the results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174
 - b. Select a subset of variants that are shown in the **Variants** tab. Click a checkbox to deselect the variants that you want to exclude from reports or from an exported **Selected Variants TSV** file.
 - c. Review the **Selected Variants** count at the top of the view.

IMPORTANT! If some variant rows that were selected with the checkboxes in the table rows, are hidden in the screen with column filtering, those variants are still included in the **Selected Variants** count at the top of the table. If you then download a **Selected Variants TSV** file, the downloaded file includes only the selected variants that are shown in the screen. That is, the **Selected Variants** count that is shown in screen always matches the number of variants in the exported **Selected Variants TSV** file.

6. (Optional) Apply a filter ▼ (**Filter**) at the top of a column to narrow the list of information that is shown in that column. For more information, see “Filter results” on page 174.

7. (Optional) Export a variants file in tab-separated value (TSV) format. Click **Export** in the upper right corner of the screen then select one of the following options.

Option	Description
Selected Variants TSV Note: The Selected Variants TSV option is not shown if no variants are selected.	The file contains the following information that is shown in the SNVs/INDELs view in the Variants tab, based on the following types of filtering. • Variants that are filtered in with the gene list that is associated with the results, or applied later to the results. • Variants that are filtered in with the filter chain that is associated with the results, or applied later to the results. • All variants that are selected with the checkboxes in the screen. • All variants from columns that are <i>not</i> shown in the screen and are hidden due to column filtering.
Currents Results TSV	The file contains the following information that is shown in the SNVs/INDELs view in the Variants tab, based on the following types of filtering. • Variants that are filtered in with the gene list that is associated with the results, or applied later to the results. • Variants that are filtered in with the filter chain that is associated with the results, or applied later to the results. • The variants that are selected with the checkboxes in the screen. The file also includes columns that contain information about annotation sources.

View results for SNVs/INDELs

The **SNVs/Indels** view in the **Variants** tab lists the calls and other information for the SNV and INDEL variants that are analyzed in each sample in a run.

Any user can view detailed variant results in the **Variants** tab.

The variants that are shown depend on the filter chain and the gene list that are applied. The first time that you view sample results in the **Variants** tab, the filter chain and gene list that is associated with the assay, if any, are applied to the results. You can change the filter chain or gene list that is applied to the variants that are shown in the results. You can further filter data that is shown on the screen with a filter (▼ **(Filter)**) at the top of the column. You can also select or deselect the checkboxes in the list of variants to filter the variants that are included in reports and results files.

You can export the information for all current variants that are shown in the software, or for variants that are selected, in tab-separated value (TSV) format.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **SNVs/Indels** to view the **SNVs/Indels** table.

For more information about the data shown in the table, see “SNVs/Indels table” on page 143.

5. (Optional) You can filter the results that are shown in the **Variants** tab. We recommend that you filter data in the following order.
 - a. Change the filter chain or a gene list that is applied to the results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.
 - b. Select a subset of variants that are shown on the **Variants** tab. Select each checkbox in the **SNVs/Indels** table for one or more variants of interest.
 - c. Apply a filter ▼ (**Filter**) at the top of a column to narrow the list of information that is shown in that column. For more information, see “Filter results” on page 174.
 - d. Review the **Selected Variants** count at the top of the view.

IMPORTANT! If some variant rows that were selected with the checkboxes in the table rows are hidden in the screen with column filtering, those variants are still included in the **Selected Variants** count at the top of the table. If you then export and download a **Selected Variants TSV** file, the exported file includes all selected variants that are shown in the screen. That is, the **Selected Variants** count that is shown in the screen always matches the number of variants in the exported **Selected Variants TSV** file.

6. (Optional) Download a variants file in tab-separated value (TSV) format. Click **Export** in the upper right corner of the screen then select one of the following options.

Option	Description
Selected Variants TSV Note: The Selected Variants TSV option is not shown if no variants are selected.	The file contains the information that is shown in the SNVs/Indels view in the Variants tab. The information that is included in the file is based on: • The columns that are shown on the screen, and not hidden due to column filtering. • The variants that are selected with the checkboxes on the screen.
Currents Results TSV	The file contains the information that is shown in the SNVs/Indels view in the Variants tab. The information that is included in the file is based on: • The gene list that is associated with the results, or applied later to the results. • The filter chain that is associated with the results, or applied later to the results. • The variants that are selected with the checkboxes on the screen. • Variants that are shown in the Results screen due to column filtering.

SNVs/Indels table

The data displayed in the **SNVs/Indels** table depends on the assay that was used in the run.

You can filter the results list in the table using filtering tools and filter chains. For more information, see “Filter results” on page 174.

Column	Description
User Classification	User-defined classification to select from the list. For more information, see “Create and assign variant classifications” on page 165.
Variant ID	The name of the hotspot as defined in the Browser Extensible Data (BED) file. The Variant ID is shown in the COSMIC column in the SNVs/Indels table. Click the link to view more annotation information. For more information, see “View annotation set details” on page 268.
Variant Name	The name of the variant.
Key Variant	Indicates whether the variant is a key variant. Possible values are Yes or No . This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
Locus	The chromosome and position of the detected variant.
OncoPrint Variant Class	The type of SNV or INDEL at the locus based on OncoPrint™ annotations. This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
OncoPrint Gene Class	The change in molecular function of the altered gene product due to the mutation, based on OncoPrint™ annotations: • Gain-of-function—The altered gene product has a new molecular function or pattern of gene expression compared to the wild-type gene. • Loss-of-function—The altered gene product lacks the molecular function of the wild-type gene. This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
Gene	The gene name.
AA Change	Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature.
Ref	The reference base or bases at that locus.
Alt	The alternate base or bases at that locus.

(continued)

Column	Description
Type	The type of variant that is detected. • snp (single nucleotide polymorphism) • mnp (multi-nucleotide polymorphism) • ins (insertion) • del (deletion) • complex • FLT3-ITD
Call	Indicates the presence or absence of a SNV/INDEL variant. When the default filter chain is applied, only the variant calls that are designated with PRESENT (HOMOZYGOUS) or PRESENT (HETEROZYGOUS) are displayed in the results table. • PRESENT (HOMOZYGOUS) or PRESENT (HETEROZYGOUS)—Indicates a high confidence call that passes all filter thresholds at a given variant position. – When the default filter chain is applied, PRESENT (HOMOZYGOUS) or PRESENT (HETEROZYGOUS) indicates the presence of the ALT (alternative) allele. • NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call. • ABSENT—Indicates that the variant is unlikely to be present in the sample.
Call Details	The reason why a variant is reported as NO CALL .
Phred QUAL Score	The Phred-scored quality. For variants found by the Long INDEL Assembler software component, this value is always set to 50. Larger values mean higher confidence in the call. Quality is calculated by posterior probability that the variant allele frequency is greater than the cutoff (min_allele_freq in the parameter file), if a variant call is made, or posterior probability that the variant allele frequency is below this cutoff (if a reference call). The posterior probability that is computed as conditional on the reads observed includes sampling variability. Quality score is typically very large for reads strongly distinguishing variants with good depth, that is, under the model assumed, evidence is overwhelming for the variant or for the reference. Marginal values can mean that either the reads do not distinguish the variant well, there is insufficient depth to resolve, or the observed allele frequency is near the cutoff.
Raw Read Depth	Total read coverage across amplicon containing the variant locus. Count of reads aligned at this locus that are eligible in variant calling after some read filters are applied.
Effective Read Depth	The number of reads that are participates in the evaluation of the variant. Read filtering and random down sampling may be applied.
Alt Allele Read Counts	The number of reads to support the alternate allele after variant calling.
Coverage	The number of total reads at a locus, wild-type + alt allele.

(continued)

Column	Description
Nuc Change	The position and identity of the nucleic acid change.
Allele Frequency	The number of variant read counts divided by the total number of read counts for the sample.
Allele Frequency (%)	The allele frequency, represented as a percentage.
Allele Ratio	The allele frequency of the REF allele and ALT allele.
Mut/WT	The ratio of mutant allele to wild-type allele.
Zygosity	Describes whether the variant is homozygous (0) or heterozygous (1).
Filtered Read Coverage	The number of reads that are participates in the evaluation of the variant. Read filtering and random down sampling may be applied.
Allele Read Count	The number of reads detected for the allele. The Allele Read Count filter sets the minimum count of the genotype alleles.
Raw Alt Allele Read Counts	Total read coverage across amplicon containing the variant locus. Count of reads aligned at this locus that are eligible in variant calling after applying certain read filters.
PPA	Possible Polyploidy Alleles (PPA). A value of Yes indicates variants that are PPA alleles. A value of No indicates variants that are not PPA alleles.
P-Value	The decision error probability associated with the Phred QUAL Score .
Mol Depth^[1]	The reports number of interrogated DNA molecules containing target. This metric defines the limit of detection at a hotspot position in a particular run and sample. For reference calls, molecular depth provides a measurable metric that serves as confirmation of variant absence among a large number of interrogated molecules. For instance, if molecular depth is $\geq 1,500$, you can have high confidence that no variant is present at $\geq 0.2\%$ variant allele frequency. If molecular depth is $\geq 2,500$, you can have high confidence that no variant is present at $\geq 0.1\%$ variant allele frequency.
WT Mol Counts^[1]	The number of detected molecules containing the wildtype allele.
Alt Allele Mol Counts^[1]	The number of detected molecules containing the alternate allele.
Mol Freq %^[1]	Molecular frequency percentage. The percentage of alternate allele reads over total reads at the locus.
% LOD^[1]	The limit of detection (LOD) of a variant allele expressed as a percentage of the WT allele. LOD is the lowest possible variant frequency in the sample that can be detected by the system with a true positive rate $>98\%$ for FFPE samples or 95% for cfTNA samples. LOD is dependent on the molecular read depth at the locus. %LOD is reported when there are no variant calls for the gene.

(continued)

Column	Description
Genomic region	If results include annotations from an imported annotation source of type Genomic region , annotations are shown in a column that contains the annotation source name.
Gene set	If results include annotations from an imported annotation source of type Gene set , annotations are shown in a column that contains the annotation source name.
Transcript	If results include annotations from an imported annotation source of type Transcript Set , or if a preconfigured or user-defined transcript set is used in the sequencing run, annotations are shown in a column with the annotation source name.
VariantDB	If results include annotations from an imported annotation source, of type VariantDB , annotations are shown in a column with the annotation source name.

^[1] Column appears only in analyses of Ion AmpliSeq™ HD sequencing data.

View results for fusions

The **Fusions** view in the **Variants** tab lists the calls and other information for the fusions that are analyzed in each sample in a run.

Any user can view detailed variant results in the **Variants** tab.

The variants that are shown depend on the filter chain and the gene list that are applied. The first time that you view sample results in the **Variants** tab, the filter chain and gene list that is associated with the assay, if any, are applied to the results. You can change the filter chain or gene list that is applied to the variants that are shown in the results. You can further filter data that is shown on the screen with a filter (▼ **(Filter)**) at the top of the column. You can also select or deselect the checkboxes in the list of variants to filter the variants that are included in reports and results files.

You can export the information for all variants, or for variants that are selected, in TSV format. The file that is exported reflects the rows of variants that are shown in the screen, and is based on the filters that are applied.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **Fusions** to open the **Fusions** view of the results.
For more information about the data shown in the table, see “Fusions table” on page 148.
5. (Optional) You can filter the selected variants that are shown in the **Variants** tab. Changes to these filters affect all of the views shown in the tab. We recommend that you filter data in the following order.
 - a. Change the filter chain or a gene list that is applied to the results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.

- b. Select a subset of variants that are shown in the **Variants** tab. Click a checkbox to deselect the variants that you want to exclude from reports or from an exported **Selected Variants TSV** file.
- c. Review the **Selected Variants** count at the top of the view.

IMPORTANT! If some variant rows that were selected with the checkboxes in the table rows, are hidden in the screen with column filtering, those variants are still included in the **Selected Variants** count at the top of the table. If you then download a **Selected Variants TSV** file, the downloaded file includes only the selected variants that are shown in the screen. That is, the number of variants that is shown in the **Selected Variants** count may be different from what is downloaded in the **Selected Variants TSV** file.

6. (Optional) Apply a filter ▼ (**Filter**) at the top of a column to narrow the list of information that is shown in that column. For more information, see “Filter results” on page 174.
7. (Optional) Export a variants file in tab-separated value (TSV) format. Click **Export** in the upper right corner of the screen then select one of the following options.

Option	Description
Selected Variants TSV Note: The Selected Variants TSV option is not shown if no variants are selected.	The file contains the following information that is shown in the Fusions view in the Variants tab, based on the following types of filtering. • Variants that are filtered in with the gene list that is associated with the results, or applied later to the results. • Variants that are filtered in with the filter chain that is associated with the results, or applied later to the results. • All variants that are selected with the checkboxes in the screen, whether those variants were selected with the gene list, the filter chain, or individually deselected.
Currents Results TSV	The file contains the following information that is shown in the Fusions view in the Variants tab, based on the following types of filtering. • Variants that are filtered in with the gene list that is associated with the results, or applied later to the results. • Variants that are filtered in with the filter chain that is associated with the results, or applied later to the results. • The variants that are selected with the checkboxes in the screen. • All variants from columns that are <i>not</i> shown in the screen, and are instead hidden due to column filtering. The file also includes columns that contain information about annotation sources.

Fusions table

The data displayed in the **Fusions** table depends on the assay that was used in the run.

You can filter the results list in the table using filtering tools and filter chains. For more information, see “Filter results” on page 174.

Column	Description
User Classification	A user-defined classification selected from the list. For more information, see “Create and assign variant classifications” on page 165.
Variant ID	The name of the fusion target as defined in the BED file. The Variant ID is shown in the COSMIC column in the Fusions table. Click the link to view more annotation information. For more information, see “View annotation set details” on page 268.
Variant Name	The name of the variant. For more information, see “Rules for variant names in Genexus™ Software” on page 525.
Key Variant	Indicates whether the variant is a key variant. Possible values are Yes or No . This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.
Locus	The chromosome positions in the reference genome that define the fusion junction.
Oncomine Variant Class	Oncomine variant class annotation that indicates fusion type based on Oncomine™ annotations. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay that is used in the run.
Oncomine Gene Class	The change in molecular function of the altered gene product due to the mutation, based on Oncomine™ annotations. • Gain-of-function—the altered gene product has a new molecular function or pattern of gene expression compared to the wild-type gene. • Loss-of-function—the altered gene product lacks the molecular function of the wild-type gene. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.
Genes (Exon)	The gene symbols and exon numbers that correspond to the donor and acceptor exons.
Read Counts	The number of mapped reads that support the presence of a fusion isoform in the sample.
Type	Assay type, (for example, Fusion, RNA exon variant (exon skipping), RNAExon Tile, Process Control or Expression Control).

(continued)

Column	Description
Call	Indicates the presence or absence of a fusion or RNA exon variant. When the default filter chain is applied, only the fusion/RNA exon variant calls that are designated with PRESENT are displayed in the results table. To view all calls, including calls that do not pass the required filter thresholds, apply the No Filter option or download the Variants (VCF) file (see “Results files” on page 178). • PRESENT—indicates a high confidence call that passes all filter thresholds at a given variant position. • ABSENT—indicates the absence of a fusion due to a variant call that falls below thresholds. • NO CALL—although some evidence for the presence of a fusion exists, the call does not pass one or more filters that are required for a high confidence fusion call.
Call Details	The reason for reporting a fusion as NO CALL or ABSENT .
Read Counts Per Million	The number of fusion read counts detected per million mapped reads.
Oncomine Driver Gene	The gene reported to be associated with increased oncogenic properties.
Gene Isoform	The name of the fusion target as defined in the BED file.
Mol Cov. Mutant ^[1]	The median molecular coverage across a fusion amplicon.

(continued)

Column	Description
Imbalance Score ^[2, 3]	Each fusion gene exhibits a characteristic Imbalance Score threshold. Scores that exceed the threshold value indicate a high likelihood of the presence of the fusion in the sample. Imbalance score calculation starts with the normalization of reads in the exon-tiling amplicons of the gene, followed by correction with a baseline that represents expression values in normal samples. Reads for observed imbalance scores come from samples. Baseline scores come from normal, fusion-negative samples and are stored in the exon tile assay baseline file for the assay. Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline) where Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples) 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample. 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample. Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline) 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene. 5' of breakpoint is the total normalized baseline values of the amplicons for the gene.
Imbalance P-Value ^[2]	The statistical significance of measure of imbalance relative to a control gene.
Predicted Break-point Range ^[2]	The exonic range for predicted fusion break point in exon tiling assays.
Ratio To Wild Type ^[2]	The molecular ratio for exon skipping assays relative to wild type control amplicons.
Norm Count Within Gene ^[2]	Exon skipping assay coverage normalized to molecular coverage of wild type (WT) MET control amplicons.

^[1] Column appears only in analyses of Ion AmpliSeq™ HD sequencing data.^[2] Column appears only in analyses that use the exon tiling fusion detection method.^[3] Applies only to FFPE assays.

Non-targeted fusion detection

In the non-targeted fusion detection method, the panel primers are used to detect fusions between novel combinations of known driver and partner genes. The sequencing reads are remapped to the fusion reference with a process that allows for the detection of multiple configurations of driver and partner genes and the detection of novel breakpoints between the known partner and driver genes.

Low confidence non-targeted fusions are identified in VCF output files with key value pairs as follows.

Key value pairs for non-targeted fusions

Type of non-targeted fusion identified	Definition	Key value pair
Out-of-frame sequence	The expected non-targeted sequence from the amplicon reference file is used to check whether the sequence is in frame. If the non-targeted sequence is out of frame (compared to the original partner or driver genes), the key-value pair <code>NT_FUSION_IN_FRAME=FALSE</code> is included in the VCF file.	<code>NT_FUSION_IN_FRAME</code> Values: • FALSE • TRUE If the key value pair is not included, then <code>NT_FUSION_IN_FRAME</code> is assumed to be <code>TRUE</code> , indicating that an out-of-frame sequence is not found.
Partner-partner fusion	Non-targeted fusions are identified if one of the genes in an isoform is not a driver gene and another is (i.e., partner-driver fusion). The input reference BED file includes annotations for driver genes, for example, in the case of <code>EML4-ALK.E6A19.COSF1296.2</code> , the <code>DRIVER_GENE=ALK</code> . In cases where the <code>DRIVER_GENE</code> field is missing in the BED file, the software checks whether at least one of the two genes involved in the non-targeted fusion is a 3p gene.	<code>NT_FUSION_DRIVER_INVOLVED</code> Values: • FALSE • TRUE If the key value pair is not included, then <code>NT_FUSION_DRIVER_INVOLVED</code> is assumed to be <code>TRUE</code> , indicating that a partner-partner fusion is not found.
Secondary isoform fusion	In some samples, a non-targeted fusion isoform might be detected in a sample while additional targeted isoforms with the same gene pair are also detected (that is, isoforms that are defined in the panel). In this case, the non-targeted fusion isoform should be identified as <code>NT_FUSION_SECONDARY=TRUE</code> , if its read count coverage is below <5% of the reads of any of the other isoforms with the same gene pair.	<code>NT_FUSION_SECONDARY</code> Values: • FALSE • TRUE If the key value pair is not included, then <code>NT_FUSION_SECONDARY</code> is assumed to be <code>FALSE</code> , indicating that a secondary isoform fusion is not found.

Additional attributes that indicate whether the non-targeted fusion is out of frame, involves partner-partner fusion, or is a secondary isoform, are added to the VCF files. This information is used by the OncoPrint™ Variant Annotator software module to annotate variants for the OncoPrint™ Comprehensive Assay Plus GX assay.

View results for CNVs

The **CNVs** view in the **Variants** tab lists the calls and other information for the copy number variants (CNVs) analyzed that are analyzed in each sample in a run.

Any user can view detailed variant results in the **Variants** tab.

The variants that are shown depend on the filter chain and the gene list that are applied. The first time that you view sample results in the **Variants** tab, the filter chain and gene list that is associated with the assay, if any, are applied to the results. You can change the filter chain or gene list that is applied to the variants that are shown in the results. You can further filter data that is shown on the screen with a filter (▼ **(Filter)**) at the top of the column. You can also select or deselect the checkboxes in the list of variants to filter the variants that are included in reports and results files.

You can export the information for all current variants that are shown in the software, or for variants that are selected, in tab-separated value (TSV) format.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **CNVs** to open the **CNVs** view of the results.

For more information about the data shown in the table, see “CNVs table” on page 153.

5. (Optional) You can filter the selected variants that are shown in the **Variants** tab. Changes to these filters affect all of the views shown in the tab. We recommend that you filter data in the following order.
 - a. Change the filter chain or a gene list that is applied to the results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.
 - b. Select a subset of variants that are shown in the **Variants** tab. Click a checkbox to deselect the variants that you want to exclude from reports or from an exported **Selected Variants TSV** file.
 - c. Apply a filter ▼ **(Filter)** at the top of a column to narrow the list of information that is shown in that column. For more information, see “Filter results” on page 174.
 - d. Review the **Selected Variants** count at the top of the view.

IMPORTANT! If some variant rows that were selected with the checkboxes in the table rows are hidden in the screen with column filtering, those variants are still included in the **Selected Variants** count at the top of the table. If you then export and download a **Selected Variants TSV** file, the exported file includes all selected variants that are shown in the screen. That is, the **Selected Variants** count that is shown in the screen always matches the number of variants in the exported **Selected Variants TSV** file.

6. (Optional) Export a variants file in tab-separated value (TSV) format. Click **Export** in the upper right corner of the screen then select one of the following options.

Option	Description
Selected Variants TSV Note: The Selected Variants TSV option is not shown if no variants are selected.	The file contains the information that is shown in the CNVs view in the Variants tab. The information that is included in the file is based on: - The columns that are shown on the screen, and not hidden due to column filtering. - The variants that are selected with the checkboxes on the screen.
Currents Results TSV	The file contains the information that is shown in the CNVs view in the Variants tab. The information that is included in the file is based on: - The gene list that is associated with the results, or applied later to the results. - The filter chain that is associated with the results, or applied later to the results. - The variants that are selected with the checkboxes on the screen. - Variants that are shown in the Results screen due to column filtering.

CNVs table

The data displayed in the table depends on the assay that was used in the run.

You can filter the results list in the table using filtering tools and filter chains. For more information, see “Filter results” on page 174.

Column	Description
User Classification	A user-defined classification selected from the list. For more information, see “Create and assign variant classifications” on page 165.
Variant ID	The identifier of the CNV variant. The Variant ID is shown in the COSMIC column in the CNVs table. The Variant ID is shown in the COSMIC column in the CNVs table. Click the link to view more annotation information. For more information, see “View annotation set details” on page 268.
Variant Name	The name of the variant.
Key Variant	Indicates whether or not the variant is a key variant. Possible values are Yes or No. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.
Locus	The starting position of the first amplicon covering the CNV gene.

(continued)

Column	Description
Oncomine Variant Class	Annotation that indicates whether CNV is an amplification or deletion. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.
Oncomine Gene Class	The change in molecular function of the altered gene product due to the mutation, based on Oncomine™ annotations: • Gain-of-function—The altered gene product has a new molecular function or pattern of gene expression, compared to the wild-type gene. • Loss-of-function—The altered gene product lacks the molecular function of the wild-type gene. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.
Gene	The gene name.
Copy Number	The copy number of a CNV gene locus per genome. This column is available when a positive call is made.
Call	Indicates the presence or absence of a CNV. When the default filter chain is applied, only the CNV-positive calls that are designated with PRESENT are displayed in the results table. • PRESENT—Indicates a high confidence call that passes all filter thresholds. • PRESENT (GAIN)—A CNV-positive call that indicates gene amplification; a high confidence variant call that passes all filter thresholds. • PRESENT (LOSS)—A CNV-positive call that indicates gene deletion; a high confidence variant call that passes all filter thresholds. • ABSENT—The absence of a variant; result is below or above the detection threshold for a CNV call. • NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.
P-Value	The statistical significance of the CNV ratio measurement.
Call Details	The reason for reporting a CNV as NO CALL.
CNV Confidence	The CNV confidence interval associated with the call. The 5% lower confidence bound value is the ploidy estimate, where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate, where it is 95% certain that the true ploidy is below that value.
CNV Ratio	The ratio of measured CNV gene locus coverage relative to coverage of non-CNV loci.
Med Read Cov Gene	The median read coverage of targeted CNV gene.
Med Read Cov Ref	The median read coverage of non-CNV reference loci.

(continued)

Column	Description
Valid CNV Amplicons	The number of amplicons spanning the CNV call.
Type	The type of variant that is detected. • CNV—Copy number variant. • LOH—Loss of heterozygosity.
Subtype	The CNV subtype. • BigDel—Deletion of at least one exon. • BigDup—Duplication of at least one exon. • GeneCNV—Whole BRCA1/BRCA2 gene deletion or duplication. • NOCALL—Read count differs from baseline by non-integer amount; evidence for a BigDel or BigDup call is weak. • REF—Read count matches reference baseline. • ARM—Aneuploidy of a chromosome arm.
Genomic region	If results include annotations from an imported annotation source of type Genomic region , annotations are shown in a column that contains the annotation source name.
Gene set	If results include annotations from an imported annotation source of type Gene set , annotations are shown in a column that contains the annotation source name.
Transcript	If results include annotations from an imported annotation source of type Transcript Set , or if a preconfigured or user-defined transcript set is used in the sequencing run, annotations are shown in a column with the annotation source name.
VariantDB	If results include annotations from an imported annotation source, of type VariantDB annotations are shown in a column with the annotation source name.

Customize the view of the Variants tab

The **Variants** tab includes detailed variant results in Genexus™ Software. You can customize the table to reflect the information that is most relevant to your research.

Change the columns in the sample results table

The sample results table in Genexus™ Software includes a default set of columns. The columns that are available depend on the assays that are installed in the software. Table columns are based on sample attributes. You can also add or remove columns for information about **Electronic Signatures**, and **Annotations Used**.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click **Columns**.

3. Select the columns that you want to add to the sample results table, or deselect the columns that you want to remove from the table.

The selected columns are added to the table or are removed from the table until you close the table.

4. Click **Save** to save the changes to the table columns.

The changes that you have made to the table appear each time that you view the sample results table. The changes are saved until you add or remove other columns, then save those changes.

To restore the table to the default view, see “Restore the default columns in the sample results table” on page 156.

Restore the default columns in the sample results table

You can restore the default view of the columns in the sample results table in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click **Columns**.
3. At the end of the list of columns that appears, click **Restore default**.

The default columns in the sample results table are restored.

Variants tab table preferences

When you view sample results in the **Variants** tab in Genexus™ Software, a default set of columns is included in a **System Default** table preference.

You can create a table preference to customize the columns that are shown for the results.

The table preference that is applied to the sample results can be associated with an assay, or applied only to the results that have the same library chemistry and research application, in the can be associated with a combination of library chemistry and research application **Variants** tab, or applied later to results that you view.

A **System Default** table preference is available to all users and can be in Genexus™ Software, or included as part of the package of assay definition files (ADFs) that are added to the software. The table preferences that you create and save are associated with the software account that you used to create the table preference and are visible only when you are signed into Genexus™ Software with that account.

Table preferences that you create include any changes that you made to each of the variant class views.

To easily create a new table preference, you can start from an existing table preference and make changes, as needed.

You can save variant table preferences in multiple ways. These save options determine how the table preference is applied—either to a set of results or to an assay.

Option	Description
Save	Saves the new table preference and adds the preference to the list of available options in the Preferences menu and is available for other results that use the same library chemistry and research application.
Save & Apply	Saves the new table preference, adds the preference to the list of available options, and applies the preference to the current variants table and any other results that use the same library chemistry and research application.
Save as Assay Default	Saves the new table preference, applies the preference to the current table, and sets the table as the default for all sample results from that assay. The new table preference replaces the preference that is applied to the open variants table. The <i>assay default</i> table preference that you save is applied to all sample results for that assay. The next time that you view results for the assay to which you save the preference as the assay default, the new assay default table preference is selected and applied to the Variants tab.

Table preferences apply only to assays that include a **Variants** tab for results shown in the software. Table preferences do not apply to results from immune repertoire assays, PGT-A assays, or microbiome assays.

Add a new table preference in the Variants tab

You can create preferences in the variants table to view the data in Genexus™ Software that is most relevant to your research. You can show and hide table columns, rearrange the order of columns, set a table preference as the default for an assay, and restore the **System Default** table preference. The table preferences that you create and save are associated with the software account that you used to create the table preference. The preferences are visible only when you are signed into the software with that account.

To easily create a new table preference, you can start from an existing table preference and make changes as needed.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to view results for the sample.
3. Click the **Variants** tab.
4. Under **Set Column Preference**, select a view of the variant class for which you want to create the table preference. The available options are based on the sample.
 - **All**
 - **SNVs/Indels**
 - **Fusions**
 - **CNVs**
5. Click **Preferences**, then click **Add New** at the bottom of the list of preferences.

6. In the **Create Table Preference** pane, name the preference, hide or show columns, and rearrange the table columns for one or more variant class view.

Option	Description
Name the table preference	Enter a new name in Table Preference Name . Use 50 or fewer characters for the table preference name.
Show or hide columns	Click  (Show) or  (Hide) to add or remove columns from the table preference.
Rearrange the column order The first two columns in the table (Selected Variants Filter Menu checkbox and  (Notes)) are fixed and cannot be moved or hidden.	Drag and drop a column name up or down to rearrange the order of the columns in the table preference.

7. Click **Save As**, then select an option to save the table preference in one of the following ways.

Option	Description
Save	Saves the new table preference and adds the preference to the list of available options in the Preferences menu and is available for other results that use the same library chemistry and research application.
Save & Apply	Saves the new table preference, adds the preference to the list of available options, and applies the preference to the current variants table and any other results that use the same library chemistry and research application.
Save as Assay Default	Saves the new table preference, applies the preference to the current table, and sets the table as the default for all sample results from that assay. The new table preference replaces the preference that is applied to the open variants table. The <i>assay default</i> table preference that you save is applied to all sample results for that assay. The next time that you view results for the assay to which you save the preference as the assay default, the new assay default table preference is selected and applied to the Variants tab.

The table preferences that you create and save are associated with your software account and are visible only to you when you are signed in to Genexus™ Software.

To apply a table preference, click **Preferences**, then select the table preference that you want to apply. If the sample results had an assay default table preference applied, the assay default preference is removed when you select any other table preference.

Edit a table preference in the Variants tab

You can edit user-defined table preferences that are shown in the **Variants** tab. You can show and hide table columns to change which columns are shown in a variant class view, rearrange the column order, and change the way a table preference is applied. You can edit only the table preferences that are associated with the Genexus™ Software account for which you are signed in.

You can also edit an existing table preference and make changes as needed to create a new table preference.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to view results for the sample.
3. Under **Edit Column Preference**, select a view of the variant class for which you want to create the table preference. The available options are based on the sample.
 - **All**
 - **SNVs/Indels**
 - **Fusions**
 - **CNVs**
4. In the sample results screen, click **Preferences**, place the pointer over the name of the table preference that you want to edit, then click  (**Edit**).
5. In the **Edit Table Preference** pane, make one or more of the following changes.

Option	Description
Change the name of the table preference	Enter a new name in Table Preference Name . Use 50 or fewer characters for the table preference name.
Show or hide columns	Click  (Show) or  (Hide) to add or remove columns from the table preference.
Rearrange the column order The first two columns of the table (Selected Variants Filter Menu checkbox and  (Notes)) are fixed and cannot be moved or hidden.	Drag and drop a column name up or down to rearrange the order of the columns in the table preference.

6. Click **Save As**, then select an option to save the table preference in the following ways.

Option	Description
Save	Saves the new table preference and adds the preference to the list of available options in the Preferences menu and is available for other results that use the same library chemistry and research application.
Save & Apply	Saves the new table preference, adds the preference to the list of available options, and applies the preference to the current variants table and any other results that use the same library chemistry and research application.
Save as Assay Default	Saves the new table preference, applies the preference to the current table, and sets the table as the default for all sample results from that assay. The new table preference replaces the preference that is applied to the open variants table. The assay <i>default</i> table preference that you save is applied to all sample results for that assay. The next time that you view results for the assay to which you save the preference as the assay default, the new assay default table preference is selected and applied to the Variants tab.

The edits are saved to the table preference.

Change the columns in the Variants tab

The sample results table in Genexus™ Software includes a default set of columns. The columns that are available depend on the assays that are installed in the software. Table columns are based on sample attributes. You can also add or remove columns from the table and save your preferences to see data of interest when you open sample results and add the results to a report. You can also add or remove a column for information about **Electronic Signatures**, and **Annotations Used**.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click **Preferences**.
3. Click a sample name to view results for the sample.
4. In the **Sample Results** screen, click the **Variants** tab.
5. Select the columns that you want to add to the **Variants** table, or deselect the columns that you want to remove from the table.

The selected columns are added to the table or are removed from the table until you close the table.

6. To save the selected columns as a table preference, click **Save As**, then select an option to save the table preference in one of the following ways.

Option	Description
Save	Saves the new table preference and adds the preference to the list of available options in the Preferences menu and is available for other results that use the same library chemistry and research application.
Save & Apply	Saves the new table preference, adds the preference to the list of available options, and applies the preference to the current variants table and any other results that use the same library chemistry and research application.
Save as Assay Default	Saves the new table preference, applies the preference to the current table, and sets the table as the default for all sample results from that assay. The new table preference replaces the preference that is applied to the open variants table. The <i>assay default</i> table preference that you save is applied to all sample results for that assay. The next time that you view results for the assay to which you save the preference as the assay default, the new assay default table preference is selected and applied to the Variants tab.

To restore the table to the default view, see “Restore default columns shown in the Variants tab” on page 161.

Restore default columns shown in the Variants tab

You can restore the default columns that are shown in the Variants table in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to view results for the sample.
3. In the sample results screen, click the **Variants** tab.
4. Click **Preferences ▶ System default**.

The system default columns shown in the table are restored.

Delete a table preference from the Variants tab

You can delete a table preference that is shown in the **Variants** tab. The table preference that you delete is removed from the list of available table preferences. The available table preferences are associated with the software account that you used to create the table preference and are visible only when you are signed into Genexus™ Software with that account.

If the table preference that you delete is applied to any **Variants** table that is saved as an assay default table preference, the default table preference that is based on the library chemistry and the research application is applied. If both table preferences are deleted, the **System Default** table preference is applied to the table.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to view results for the sample.
3. In the **Sample Results** screen, click **Preferences**, place the pointer over the name of the table preference that you want to delete, then click  (**Delete**).

If you delete the table preference that is currently applied to the **Sample Results** that are shown, the **System Default** table preference is applied. You can then optionally apply any table preference that remains available in the list of preferences.

Variants file exports

You can export files of variants in tab-separated value (TSV) format from sample results in Genexus™ Software. You can download and use the exported file to further interpret the data.

You cannot download a variants file for analyses that do not include a TSV file in the results package. For example, runs that use the immune repertoire or metagenomics assays do not include variants.

The contents of each type of variants file that is available for download are as follows.

- **Selected Variants TSV**—A TSV file that contains variants, or information for the variants, which were *filtered IN* and selected in the results. When you export a Selected Variants TSV, you will get the same number of variant records that are shown in the **Selected Variants** count that is shown at the top of the results table.

The file will contain variants based on 2 criteria.

- Variants in the sample results that are *filtered IN* and selected by the filter chain criteria.
- Variants in the sample results that are *filtered IN* and selected by the gene list criteria.

If you change the filter chain or gene list, the selected variants are reset.

Note: Column filters that are applied to the results do not affect the file. That is, any variants that are *filtered OUT* by column filters in the software, are included in the **Selected Variants TSV** file.

- **Current Results TSV**—A TSV file that contains variants, or information for the variants, as those variants are shown in the **Sample Results** screen. When you export a **Current Results TSV** file, you get the records as they are shown in the **Sample Results** screen. The file will contain variants based on 3 criteria.
 - Variants in the results that are *filtered IN* by the filter chain criteria.
 - Variants in the results that are *filtered IN* by the gene list criteria.
 - Any variants that are *filtered OUT* by column filters in the software are *not* included in the **Current Results TSV**.

Export selected variants

You can export a file in tab-separated value (TSV) format of selected variants from sample results in Genexus™ Software. The file is called a **Selected Variants TSV** file. You can download and use the exported file to further interpret the data.

The **Selected Variants TSV** contains variants, or information for the variants, which were *filtered IN* for the analysis. When you export a **Selected Variants TSV**, you get the same number of variant records that are shown in the **Selected Variants** count that is shown in the **Sample Results** screen.

If you want to download a VCF file of selected variants, see instead “Results files” on page 178.

The export file contains variants based on 2 criteria.

- Variants in the sample results that are *filtered IN* and selected by the filter chain criteria.
- Variants in the sample results that are *filtered IN* and selected by the gene list criteria.

Column filters that are applied to the results, do not affect the file. That is, any variants that are *filtered OUT* by column filters in the software are included in the **Selected Variants TSV** file.

IMPORTANT! If some variant rows that were selected with the checkboxes in the table rows are hidden in the screen with column filtering, those variants are still included in the **Selected Variants** count at the top of the table. If you then export and download a **Selected Variants TSV** file, the exported file includes all selected variants that are shown in the screen. That is, the **Selected Variants** count that is shown in the screen always matches the number of variants in the exported **Selected Variants TSV** file.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. (Optional) Select a variant type to view the **CNVs**, **SNVs/Indels**, or **Fusions** table.
The default variant type selection is **ALL**, which shows results for all variant types.

For more information about the data shown for each variant type, see “CNVs table” on page 153, “SNVs/Indels table” on page 143, and “Fusions table” on page 148.

5. Click **Export ▶ Selected Variants TSV** in the upper right corner of the screen to export a data file in tab-separated value (TSV) format.

Depending on the browser settings, the software downloads the file or prompts you to open or save the file.

Export current results

You can export a file in tab-separated value (TSV) format that contains the same variants, or information for the variants, that are shown in the **Sample Results** screen. in Genexus™ Software.

The file is called a **Curent Results TSV** file.

If you want to download a VCF file that includes specific variants that are based on filter chain and gene list selections, see instead “Results files” on page 178.

The contents of the variant files are based on the view that is selected in the **Results** screen — **ALL**, **SNVs/Indels**, **Fusions**, or **CNVs**, and the download option that you select in the software.

You can download and use the exported file to further interpret the data.

- The export file contains variants based on 3 criteria.
 - Variants in the results that are *filtered IN* by the filter chain criteria.
 - Variants in the results that are *filtered IN* by the gene list criteria.
 - Any variants that are *filtered OUT* by column filters in the software are *not* included in the **Current Results TSV**.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. (Optional) Select a variant type to view the **CNVs**, **SNVs/Indels**, or **Fusions** table.

The default variant type selection is **ALL**.

For more information about the data shown for each variant type, see “CNVs table” on page 153, “SNVs/Indels table” on page 143, and “Fusions table” on page 148.

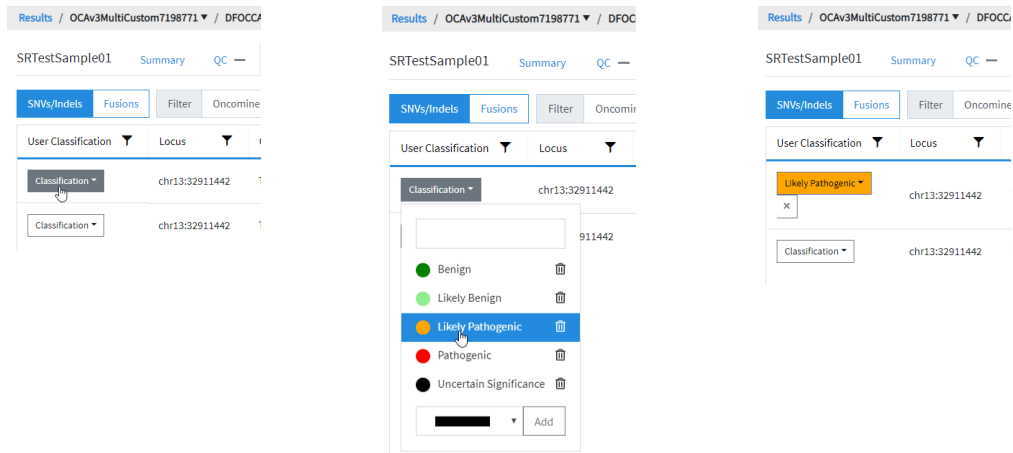
5. Click **Export ▶ Current Results TSV** in the upper right corner of the screen to download a data file in tab-separated value (TSV) format.

Depending on the browser settings, the software downloads the file or prompts you to open or save the file.

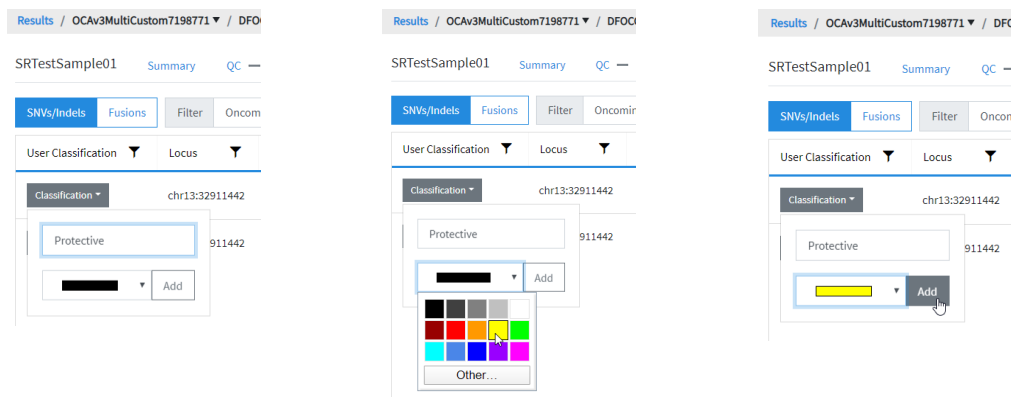
Create and assign variant classifications

You can create and assign user-defined variant classifications in the variant tables.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name in the **Sample Name** column in the row of a sample of interest to open the **Results** screen for the sample.
3. Click the **Variants** tab.
4. To refine the list of variants shown in the table, select a variant type to display: **SNVs/Indels**, **Fusions**, or **CNVs**.
5. In the **Variants** table, in the **User Classification** column, perform any of the following actions.
 - To assign an existing classification to a variant, select it from the list. The **Classification** menu changes to the name of the classification, as shown in this example.



- To create a new classification, enter a name for the classification in the text box, select a color for the new classification, then click **Add**.



- To remove a classification from a variant, click **X (Remove)**.

- To delete a classification from the list, click  **Delete** next to the classification name. The classification will be removed from all variants in all results.

Review the variant pileup view

Any user can review variant tracks with the pileup view in Genexus™ Software. The pileup view is a figure that the software generates to show each BAM track represented by a single bar, with the sequence variation or variations denoted in each track. The BAM tracks are aligned against a reference track. The variant pileup view can be used to identify potential variants.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the **Variants** tab, select the row of the variant of interest.

The pileup view is shown below the variants table.

Change the view of the variant pileup

You can zoom and pan the variant pileup view and add or remove guides and track labels to more easily understand the data. You can also adjust the view of the individual pileup tracks. For more information, see “Adjust pileup tracks” on page 167.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the **Variants** tab, select the row of the variant of interest.

Action	Procedure
Track the position as you place the pointer over the variant pileup.	Click Cursor Guide .
Mark the center of the variant pileup.	Click Center Line .
Remove the track labels.	Click Track Labels .
Zoom in to increase the magnification of the image.	Click + (Zoom in) one or more times, or double-click the image.
Zoom out to decrease the magnification of the image.	Click - (Zoom out) one or more times.
Pan across the image to move the view to another location on the screen with its current magnification.	Click and drag the pointer over the image.

Adjust pileup tracks

You can adjust the BAM read coverage track, the reference track, hotspot tracks, the target region track, and annotation tracks in Genexus™ Software to make it easier to view the data. When you adjust a track, it changes the amount of data that is shown in the plot. For example, you can see more tracks when you use the **Squish** setting.

Note: For best results, click the **Expand** option to view details about variants and base calls that are shown in each read coverage track

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the **Variants** tab, select the row of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to a track, then select an option to adjust the view of the track.

Option	Description
Expand	Select this option to view the tracks with the maximum visible height.
Squish	Select this option to view the tracks with the minimal height for each track.
Collapse	Select this option to view the tracks with overlapping transcripts shown along a single line. When Collapse is selected for BAM tracks, the BAM tracks are not shown; only the coverage density is shown.

Adjust the view of the reference track

You can adjust the view of the reference track in Genexus™ Software to view or hide the three-frame translation of the reference track sequence. You can also view the forward or reverse reference track sequence.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the **Variants** tab, select the row of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click  (Actions) next to the reference track.

Option	Description
View the reverse strand	Click Reverse . This option is available only when the forward strand is shown.
View the forward stand	Click Forward . This option is available only when the reverse strand is shown.
View the reference sequence translation	Click Three-Frame Translate . This option is available only when the amino acid sequence is not shown.
Hide the reference sequence translation	Click Close Translation . This option is available only when the amino acid sequence is shown.

Adjust the view of the BAM tracks

You can adjust the view of the BAM tracks in Genexus™ Software to dynamically customize the view of the data.

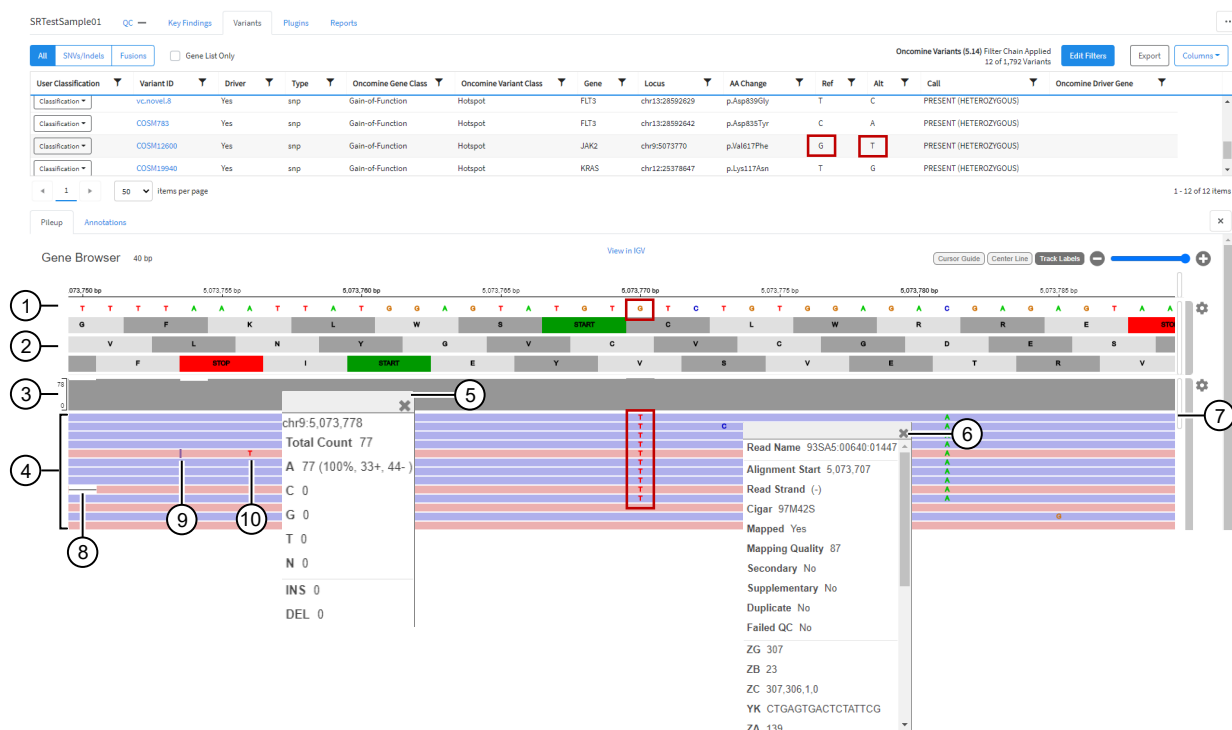
1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the **Variants** tab, select the row of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click  (Actions) next to the BAM tracks.

Option	Description
View all BAM tracks in monochrome gray	In Color By , deselect the checkmark next to Read Strand . This option is available only when the BAM tracks are colored by read strand.
Distinguish between the direction of the BAM tracks with color	In Color By , click Read Strand . This option is available only when the BAM tracks are not colored by read strand.
Expand, squish or collapse the BAM tracks	For more information, see “Adjust pileup tracks” on page 167.
Show all the bases in the BAM tracks	Click Show all bases . This option is available only when all bases in the BAM tracks are not shown.
Change the minimum threshold for the visibility of BAM tracks	- Click Set visibility window. - In the Set visibility window dialog box, enter the minimum number of bases needed to show the BAM tracks. When you zoom in and out to view the variant pileup, the scale of the tracks shown in the screen changes. For more information, see “Change the view of the variant pileup” on page 166. You can adjust the visibility window according to the scale shown in the screen.

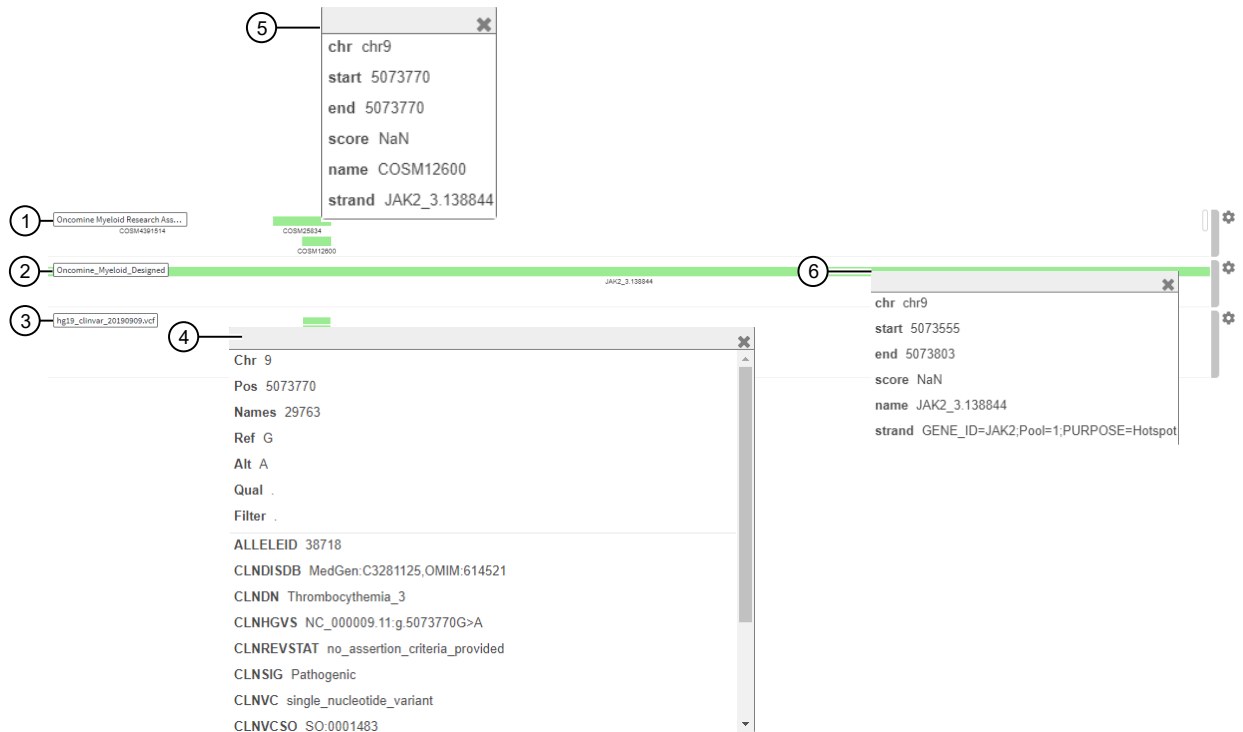
Example of a variant pileup view

The following figure is an example of a variant pileup view, shown in the **Gene Browser** in the **Variant** tab. The variant pileup view graphically represents the sequencing tracks, the reference track, and the annotation tracks, and can help you understand the variant data and analyze results. The reference allele and the alternate base for the selected variant is indicated in the image, both in the variant table and the in the pileup view. You can customize your pileup view. Reorder the tracks displayed by clicking a gray bar on the right edge of a track, then dragging the track to a different location. You can also click-drag the cursor at any position in the image to move the image left or right in the screen. For more information, see “Adjust pileup tracks” on page 167.



- ① The reference track. When zoomed out, the nucleotide sequence is no longer displayed, but the sequence is represented by colored bars.
- ② The three-frame translation is shown in this example (not by default). For more information, see “Adjust the view of the reference track” on page 167.
- ③ The coverage track.
- ④ The BAM/CRAM track.
- ⑤ Click the coverage track to see detailed information, such as the total count, the number of reads, and the number of molecules.
- ⑥ Click a BAM track to see detailed information, such as the **Mapping Quality** and **Base Quality**.
- ⑦ Use the sliders to view more tracks.
- ⑧ Deletion.
- ⑨ Insertion.
- ⑩ Alternate base in the BAM read coverage track.

The following figure is an example of the reference tracks and annotation tracks shown beneath the BAM tracks in the **Gene Browser**.



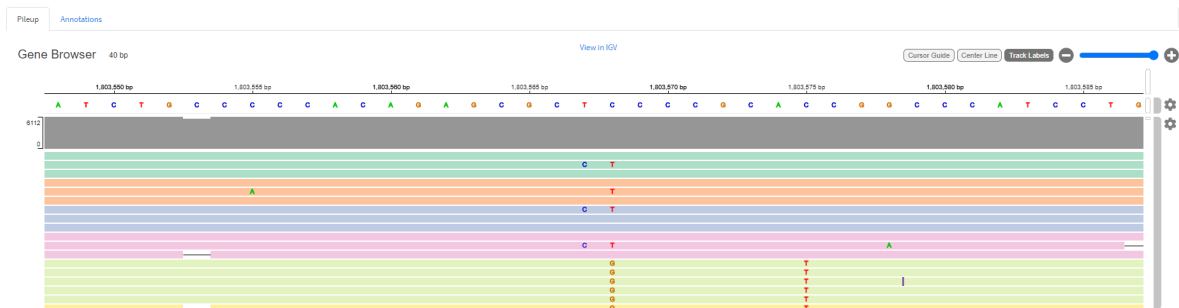
- ① The hotspot track.
- ② The target regions track.
- ③ Annotation tracks (COSMIC and ClinVar). In this example, only ClinVar is shown.
- ④ Click the annotation track for additional details; for example, the CLNHGVS (the top-level genomic HGVS expression for the variant).
- ⑤ Click a hotspot track for additional details, such as the start and end positions.
- ⑥ Click the target region track for additional details, such as the start and end positions.

View the variants from an Ion AmpliSeq™ HD sequencing run

You can view a summary of data about the identified variants, and toggle to other views that provide more details about the same variants.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the **Variants** tab, select the row of the variant of interest.

5. Review detailed variant data and read coverage tracks.

**Example visualization**

Ion AmpliSeq™ HD sequencing runs group consensus reads into families. A family is a group of reads that are associated with the same DNA molecule before library amplification. Each family is identified using the molecular tags, and consensus reads with the same molecular tags are grouped into the same family. The color of the consensus reads is used to indicate individual families.

Within each read track, each nucleotide variant is indicated by a different color. T, A, C, and G are red, green, blue, and orange, respectively. An "I" indicates insertion, and white color with a dash indicates deletion.

6. You can sort, adjust, and view details about variants and base calls that are visualized in each read coverage track.

Option	Description
View the read coverage tracks sorted by family	Click (Actions) next to the read coverage track, then select Color ▶ Molecule .
Adjust the BAM tracks	Click (Actions) next to the read coverage track, then select an option to adjust the view of the track. For more information, see “Adjust pileup tracks” on page 167.
Review detailed data about a BAM read	Single-click the read track to obtain information such as the mapping quality, the strand, and the read base.
Review distribution of base calls at a selected position	Click the density plot (the gray bar at the top of the read coverage tracks) to view information about the total count, total reads, and total number of molecules, the distribution of single nucleotides at that position, and the number of insertions and deletions.

Visualize sample results

You can visualize some sample results in Genexus™ Software in the **Variants** tab.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab, then click **SNVs/Indels**, **CNVs**, or **Fusions**.
The table opens to display SNVs/INDELs, CNVs, or Fusions results, depending on which view of the results is selected. For more information, see “View results for SNVs/INDELs” on page 141, “View results for CNVs” on page 152, or “View results for fusions” on page 146.
4. In the **Sample name** column, click the name of the sample of interest.

5. In the top right corner of the screen, click **Visualization**, then select an option to visualize the data. The options shown depend on the view of the sample results that is selected.
 - **TMB plots**
 - **BRCA**
 - **Allele Specific Copy Number**
 - **RNA Exon Variant**
 - **RNA Exon Tile Fusion Imbalance**
6. Review the plots that are shown for the sample results.

To return to the table view of **SNVs/Indels**, **CNVs**, or **Fusions** results, click **X (Remove)** next to the **Visualization** list.

View variants with Integrative Genomics Viewer (IGV)

You can view variants that you download from sample results in Genexus™ Software with the Broad Institute Integrative Genomics Viewer (IGV). The viewer is available at the Broad Institute website: <http://software.broadinstitute.org/software/igv/home>. The IGV desktop application is a standalone viewer on your computer.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Name** column, click the sample name of interest, then click the **Variants** tab.
3. In the variants table, select the row of the variant of interest.
The **Pileup** view is shown below the table.
4. At the top of the **Pileup** view, click **Export IGV Files** to download a ZIP file that contains files for upload to the Integrative Genomics Viewer (IGV).
5. Open Integrative Genomics Viewer (IGV) on your computer.
6. Unzip the files, then open the **session.xml** file in Integrative Genomics Viewer (IGV).
The files that are exported contains only information for the selected variant as follows.
 - For DNA variants
 - Sliced data is available for download that is based on the variant locus and flanking 200 base pairs.
 - Required files in the ZIP file includes all files, with the exception of the reference genome FASTA files, which are not included.
 - For DNA CNV variants
 - Sliced data is available for download that is based on the CNV start position of - 200 and CNV end position of + 200 base pairs.
 - Required files in the ZIP file includes all files, with the exception of the reference genome FASTA files, which are not included.
 - For RNA variants
 - Sliced data is available for download that is based on the fusion isoform.

Filter results

You can filter results that are shown in the **Results** screen, in the following ways.

- A filter chain is applied to the results if it is associated with the assay. You can apply a different filter chain in the **Variants** tab, or in the **Key Findings** tab. A filter chain is a set of filters that Genexus™ Software uses to narrow the list of variants that are included in results.
- A gene list is applied to the results if it is associated with the assay. You can apply a different gene list in the **Variants** tab, or in the **Key Findings** tab. A gene list is a collection of genes relevant to a cancer type, or a set of genes related to other research.

Note: If you change the filter chain or gene list, the variants that are selected with the checkboxes in the **Results** screen will change, and the number of variants in the **Selected Variants** count changes to match the selections.

- You can use the checkboxes at the left side of the table to select a subset of variants in results that are shown in the **Variants** tab.
- You can apply filters to columns of information that appear in the **Variants** tab. The filter at the top of each column narrows the list of information.

IMPORTANT! If you use a column filter to filter data that is shown in a column, variants that are selected using the checkboxes in the table rows can be filtered out in the results that are shown on the screen. However, there is no change to the **Selected Variants** count at the top of the table, as shown in Figure 2.

Results / ReportInSignoffCycle7314787 / OPA DNA Fus v3.2.0 / SRTestSample01

SRTestSample01 QC X Key Findings Variants Plugins Reports

All SNVs/Indels CNVs Fusions Selected Variants: 6

Variant Matrix Summary (5.36) Filter Chain Applied 6 of 3,159 Variants

	User Classification	Variant ID	Key Variant	Type	Oncomine Gene Class	Oncomine Variant Class	Gene	Locus
☒	Classification	COSM12473	No	snp	Loss-of-Function	Truncating	CDKN2A	chr9:219
☒	Classification	COSM10709	No	snp	Loss-of-Function	Hotspot	TP53	chr17:75

Figure 2 Effects of column filter on Selected Variants count

- ① A column filter is applied to the **Oncomine Gene Class** column that removes Gain-of-function data from the results screen.
- ② The **Selected Variants** count includes all selected variants—those that are selected using the checkboxes, and the selected variants that are filtered out with the **Oncomine Gene Class** column filter.
In this example, the **Selected Variants** count is 6, but only two *selected variants*, that is variants that are selected with a checkbox, are shown in this screen due to the column filter.

Show variant results based on a filter chain or a gene list

A filter chain is a set of filters that Genexus™ Software uses to narrow the list of variants that are included in sample results. A filter chain that is associated with the assay is applied automatically to the sample results. When you view sample results, you can change or remove the filter chain, in the **Variants** tab or in the **Key Findings** tab, and refine the list of variants that is shown.

You can also refine the variants that are shown in sample results in the **Variants** tab and **Key Findings** tabs when you change or remove a gene list that is associated with the assay and automatically to the sample results. A gene list is a collection of genes relevant to a cancer type, or a set of genes related to other research.

You cannot change or remove a filter chain or gene list in the **Variants** tab or **Key Findings** tab if a report for the sample results is signed off.

If you want to instead add or change the filter chain or gene list that is associated with an assay, you can use the settings when you create, or copy and edit, an assay. For more information, see “Panel step assay options” on page 240.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab, or the **Key Findings** tab, then click **Filters**.
The list of variants shown is based on the filter chain and gene list, if any, that are currently applied to the sample results.
4. Select one of the following options to change list of variants based on the filter chain or gene list of your choice.
The list of available filter chains is based on the reference genome that is used in the sample results.

Option	Action
Apply a filter chain to the results.	In Filter Chain, select a filter chain.
Remove a filter chain from the results.	Select No Filter Chain.
Apply a gene list to the results.	In Gene List, select a gene list.
Remove a gene list from the results.	Select No Gene List.

5. (Optional) Edit or copy a filter chain to add or remove a filter and save the changes to the filter chain before the filter chain is applied the results. The list of available filters to add to a filter chain are based on the annotation sources that are used in the sample results.
 - a. Click  (**Edit**), then make selections in the **Edit Filter Chain** dialog box to edit an existing user -defined filter chain.
 - b. Click  (**Copy**), then make selections in the **Copy Filter Chain** dialog box to make a copy of, and customize an existing filter chain.
 - c. Click **Save** to save the changes to the filter chain.
6. Click **Save** to save the filter chain and gene list selections to the sample results.
An updated list of variants is selected and shown in the **Variants** tab and the **Key Findings** tab, based on the filter chain and gene list that is selected. The filter chain or gene list selections that are saved to the results are applied when the results are reopened.

Filter the list of variants with column filters

You can use column filters to immediately narrow the list of results that are shown in the **Variants** tab. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters are applied. The changes to the list of variants are not saved. If you sign out of the software, or go to another screen in the software and then return to the screen in which the filters were previously applied, the filters that you apply to columns are cleared.

IMPORTANT! If you use a column filter to filter data that is shown in a column, variants that are selected using the checkbox in the table rows might be filtered out in the results that are shown on the screen. However, there is no change to the **Selected Variants** count at the top of the table when the column filter is used. The **Selected Variants** count includes all selected variants — those that are shown as selected with the checkboxes on the screen, and any selected variants that are filtered out with a column filter but not shown on the screen.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name in the **Sample Name** column.
3. In the **Results** screen, click the **Variants** tab.
4. Select the variant class to display the results: **SNVs/Indels**, **Fusions**, or **CNVs**.
5. In the table of variants, in the column heading of interest, click  (**Filter**).

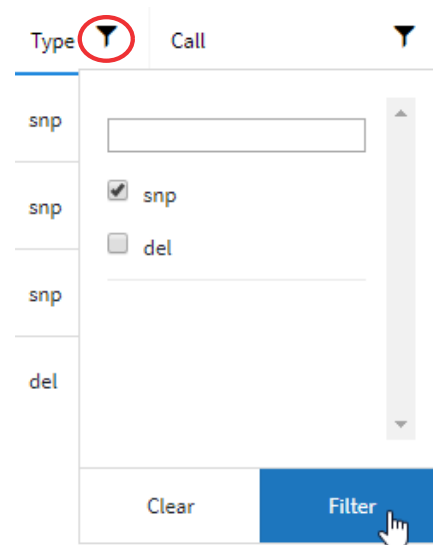
- In the search field, enter at least 3 characters, then click **Filter**.
- Select the checkbox in the row of each filter that you want to apply, then click **Filter**.

The options that are available depend on the column and variant class. For example, you can filter data in the **Type** column to show one specific variant type.

In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

6. Click  **Clear Filters** to remove all filters and view the full list of run results.

The column or columns to which you applied a filter change to reflect the filter and selected options.



Review and download audit records for a variant

Any user can review and download the entire history of a variant in Genexus™ Software. An audit trail lists the records of actions made to the selected variant. Each time-stamped audit record shows the original variant values and any new values for each action.

The following user actions generate an audit record.

- Changes to variant classifications
- Changes to a variant
- Addition of a note

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, place the pointer over a completed run, then click **Variant Audit**.
If there are no variants in the results, a message indicates that no variant audit is available.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.
4. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp. Depending on the browser settings, the software downloads the file or prompts you to open or save the file.

Results files

The following files can be downloaded for each sample. Results files include the sequencing data, results from the analyses, such as variant files, and audit and log files.

The files that are available for download depend on the assay.

For a list and descriptions of plugin output files, see “Output files generated by the coverageAnalysis plugin” on page 414, “Output files generated by the molecularCoverageAnalysis plugin” on page 430, and “Output files generated by the sampleID plugin” on page 434.

Results can be contained within a ZIP file that contains multiple files, or a single file. For instructions to download results files, see “Download results files for multiple sample results” on page 183 and “Download results files for a sample result” on page 183.

Option	File name	Description
Variants		
Genome VCF (.vcf)	<code><Sample_name>_<LibPrepID>_variant.filtered.genome.vcf</code>	A genome VCF (gVCF) file contains all variants reported in the sample results plus coverage at non-variant loci. Coverage information uses the samtools-depth calculation and the coverage profile is segmented using a Hidden Markov Model and Viterbi algorithm. Coverage information for all sites is included whether or not a variant call exists for the site. For more information on the gVCF file type, see https://gatk.broadinstitute.org/hc/en-us/articles/360035531812-GVCF-Genomic-Variant-Call-Format. You can view the file individually, or upload a VCF file to a software application that requires VCF files.
Filtered Variants (.vcf)	<code><Filter_chain_name>_filtered.vcf</code> <code><gene-list-name>_filtered.vcf</code> <code><filter-chain-name>_<gene-list-name>_filtered.vcf</code>	File or files that list filtered variant results in variant call format (VCF). The variants that are included in the VCF file depend on whether a filter chain or gene list, or both, are applied to the sample results. A file is not generated if no filter chain or gene list is selected. For more information, see “Show variant results based on a filter chain or a gene list” on page 174. You can view the files individually, or upload a VCF file to a software application that requires VCF files.
All Variants (.vcf)	<code><Sample_name>_<LibPrepID>_<analysisID>.vcf</code>	File that lists variant results in variant call format (VCF). You can view the file individually, or upload a VCF file to a software application that requires VCF files.

(continued)

Option	File name	Description
All Variants (.tsv)	<Sample_name>_<LibPrepID>_<analysisID>.tsv	The file in TSV format contains the information that is shown in the variants table in tab-separated value format (TSV). The file contains all variants in the sample result. In addition to the information shown in the variants table, the file includes information for the columns that are available but are not selected. The file also includes information about annotation sources.
Selected Variants (.vcf)	<Sample_name>_selected_variants.vcf	File that lists variants that are selected in the software in the Variants tab, in tab-separated value format (TSV) for the selected view (All, SNVs/Indels, CNVs, or Fusions), and is based on the following types of filtering. • Variants that are filtered in with the gene list that is associated with the results, or applied later to the results. • Variants that are filtered in with the filter chain that is associated with the results, or applied later to the results. Only variants that are selected with the checkboxes in the screen are included in the file, whether those variants were selected with the gene list, or the filter chain. Variants with checkboxes that are deselected in the screen are not included in the file. You can view the file individually, or upload a VCF file to a software application that requires VCF files.
Copy Number results (.png)	cn_results.png	A file that represents the CNV amplicons in the X scale and log2 ratio in Y scale. You can open the file with a text editor.
Amplicon Data (.txt)	amplicon_data.txt	A tab delimited file that contains information about the copy number estimates calculated per gene and the adjustments applied from the baseline, including pool correction, amplicon GC correction, amplicon length correction, and PC correction.
Sequencing Results		
DNA Unmapped CRAM File (.cram)	<barcode>_rawlib.basecaller.cram	Unmapped DNA barcode CRAM file; output after mapping reads to reference.

(continued)

Option	File name	Description
DNA Mapped CRAM File (.cram)	merged.cram	Mapped CRAM file of combined barcode reads.
DNA Mapped CRAM File (.crai)	merged.cram.crai	Mapped CRAM Index file.
DNA Processed CRAM File (.cram)	merged.bam.ptrim.cram	Mapped CRAM file of combined barcode reads.
DNA Processed CRAM Index File (.crai)	merged.bam.ptrim.cram.crai	Mapped CRAM index file.
RNA Unmapped CRAM File (.cram)	<barcode>_rawlib.basecaller.cram	Unmapped RNA CRAM file; output of base calling, contains unmapped reads.
RNA Mapped CRAM File (.cram)	<barcode>_rawlib. basecaller_alignments.cram	Mapped CRAM file of combined barcode reads.
RNA Mapped CRAM Index File (.crai)	<barcode>_rawlib. basecaller_alignments.cram.crai	Mapped CRAM index file.
DNA Unmapped Bam File (.bam)	<barcode>_rawlib.basecaller.bam	Unmapped DNA barcode BAM file; output after mapping reads to reference.
DNA Mapped Bam File (.bam)	merged.bam	Mapped BAM file of combined barcode reads.
DNA Mapped Bam Index File (.bai)	merged.bam.bai	Mapped BAM Index file.
DNA Processed Bam File (.bam)	merged.bam.ptrim.bam	Mapped BAM file of combined barcode reads.
DNA Processed Bam Index File (.bai)	merged.bam.ptrim.bam.bai	Mapped BAM index file.
DNA Basecaller FASTQ File (.fastq)	<barcode>_rawlib. basecaller.fastq	FASTQ file of the DNA barcodes used.

(continued)

Option	File name	Description
RNA Unmapped Bam File (.bam)	<barcode>_rawlib.basecaller.bam	Unmapped RNA BAM file; output of base calling, contains unmapped reads.
RNA Mapped Bam File (.bam)	<barcode>_rawlib.basecaller_alignments.bam	Mapped BAM file of combined barcode reads.
RNA Mapped Bam Index File (.bai)	<barcode>_rawlib.basecaller_alignments.bam.bai	Mapped BAM index file.
RNA Basecaller FASTAQ File (.fastq)	<barcode>_rawlib.basecaller.fastq	FASTQ file generated from the unmapped BAM file of the RNA barcodes used.
Test Fragment Basecaller FASTAQ File (.fastq)	rawtf.basecaller.fastq	FASTQ file for the test fragment.
Reports		
Report	<language>_<sampleName>_<mode>_<templateName>_<assayName>_<date>.pdf	A PDF report that contains sample-specific results. For more information, see “Variant reports” on page 225.
Sample Summary	Info.csv	Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary. Files are available for both user-defined and system installed assays in Genexus™ Software.
Audit and Log		
Analysis Log File	analysis.log	Analysis log file.
Sample Summary	Info.csv	Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary. Files are available only for assays from Genexus™ Software version 6.2 and earlier.
Run Audit	PlannedRun-AuditTrail.pdf	Contains all audit records pertaining to the run.

(continued)

Option	File name	Description
Troubleshooting Files		
Log Files^[1]	analysis.log	Analysis log file.
	summary.<timestamp>.log	Start and end time for each time an assay module is executed for the analysis.
	various.err various.out	Analysis pipeline logs used by Field Service Engineers (FSEs) for troubleshooting.
	Info.csv	Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary.
	PlannedRun-AuditTrail.pdf	Contains the audit trail of the run plan in PDF format.
Other^[1]	analysis.ini analysisSamples.json	Analysis configuration files, including secondary and tertiary INI files.
VCF Files	analysis.vcf SmallVariants.vcf SmallVariants.filtered.vcf	Summary of variant results in variant call format (VCF). Summary of all results for small variant in variant call format (VCF). Summary of filtered results for small variant in variant call format (VCF). You can view the files individually, or upload a VCF file to a software application that requires VCF files.

^[1] Separate folders are generated for each sample. If included in the run, separate folders are also generated for an NTC and positive control. Runs that use an assay that is been enabled for resequencing include a separate folder called Reseq that contains the log files for the resequencing portion of the run.

Download results files for multiple sample results

You can download results files for multiple sample results in Genexus™ Software. If the run included more than one assay or more than one research application for the assay, you can download the results for each assay or research application at once.

If you want to export a TSV file that includes specific variants based on options to apply filter chains, gene lists, column filters and variants that you select in the screen, see instead “The Variants tab” on page 139.

1. In the menu bar, click **Results ▶ Run Results**.
2. Select the assay if the run included more than one assay or more than one research application for the assay.
3. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Download Files**.
4. In the **Download Files** dialog box, select the files to download, then click **Download**.
For information about the files, see “Results files” on page 178.

The selected results files are downloaded in one ZIP folder named `Runname_assayname`. The folder contains subfolders for each sample result except for any NTC and positive control samples. Control sample information is in the `SmallVariants.vcf`. For more information, see “Results files” on page 178.

You can view the extracted files individually, or upload a VCF file to a software application that requires VCF files.

Download results files for a sample result

You can download results files in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Name** column, click the sample name of interest, then click the **Variants** tab.
3. In the **Variants** tab, click **⋮ (More Options) ▶ ▶ Download Files**.
4. In the **Download Files** dialog box, select the files to download, then click **Download**.
For information about the files, see “Results files” on page 178.

The selected results files are downloaded in one ZIP folder.

You can view the extracted files individually, or upload a VCF file to a software application that requires VCF files.

Run plugins after a sequencing run is complete

You can run plugins for sample results after a sequencing run is complete using the **Run Plugin** function in the **Results** screen for the sample. Use this command to run one or more plugins on sample results for the first time, or to rerun any plugin that has been run before (for example, if a plugin has been updated or failed during analysis).

1. In the menu bar, click **Results** ▶ **Sample Results**.

2. Click a sample name in the **Sample Results** screen.

3. Click the **Plugins** tab, then click **Select Plugins**.

If you run the CustomerSupportArchive plugin, ensure that the latest plugin version is selected. For more information about the latest version, see the Genexus™ Software release notes.

4. Select the assay to which the plugin run will apply, then select the plugin or plugins to rerun. Optionally, click **Configure** to change settings for a plugin, if needed.

5. Click **Run Plugins**.

While plugins are running, the options to run the plugins is unavailable for that run.

6. When the plugin run is complete, you can review the plugin results. Scroll to the section for the plugin in the **Plugins** tab of the **Sample Results** screen. For more information, see Chapter 13, “Plugins for data analysis”.

Compare sample results

Any user can compare variant results across samples with Genexus™ Software. Comparisons between various results can be made.

- To compare results from immune repertoire assays, see “Compare immune repertoire results” on page 191.
- To variant results across all other assays, see “Compare variant results” on page 184.

Results generated using the system-installed Oncomine™ TCR Beta-LR Assay GX or any other immune repertoire assay can only be compared to sample results from runs that use the same assay.

Compare variant results

You can compare variant results and create comparison plots from multiple sample results with Genexus™ Software. A description of the comparison of results from immune repertoire is not included here. For more information, see “Compare immune repertoire results” on page 191. Comparisons across samples or runs can be used to analyze results for a hotspot of interest. You can also view the trends of a reported variant over time.

The following comparisons can be made in Genexus™ Software.

- Comparisons of multiple samples, sequenced and analyzed with the same or different assays.
- Comparisons of multiple samples over time, sequenced and analyzed with the same or different assays.
- Comparisons of the results from one sample, sequenced and analyzed with different assays.

If samples from different assays are compared, the research application (such as DNA or DNA and Fusions) must be comparable. That is, results generated from DNA-only assays cannot be compared to results generated from RNA-only or fusion-only assays. However, if the results from a research application include the type of results from another research application, the comparison can be made. That is, you can compare variant results generated from a DNA and Fusion assay to the variant results generated from a DNA-only assay.

View variant result comparisons

You can view variant comparisons in tabular format in **Fusions**, **SNVs/Indels**, and **CNV** comparison tables in the **Compare** screen.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click the checkboxes next to the sample names of interest.

Tip: In the **Sample Status** column heading, click  (**Filter**), then select **Completed** to limit the entries that are displayed on the **Sample Results** screen.

3. Above the list of sample results, click  **Compare**.

Note:  **Compare** is not enabled until the **Variant ID** checkboxes for at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. The **OncoPrint™ Extended** filter is the default filter for the data displayed in the tables.

4. (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different filter chain to the variants displayed in the tables. For more information, see “Filter results” on page 174.
5. (Optional) Click **Export** to download an XLSX spreadsheet file of the variant comparison table.

Results / Compare

Compare Samples

⑤ SNVs/Indels

① Export

② ✓ SRTestSample01 | 2021-03-26

③

④ ✓ SRTestSample03 | 2021-03-26

⑥ Variant ID	⑦ Gene	⑧ AA Change	⑨ Call	Call
☐ COSM5673	CTNNB1	p.S33Y	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
☐ COSM6128	CTNNB1	p.S45del	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
☐ COSM763	PIK3CA	p.E545K	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
☐ COSM775	PIK3CA	p.H1047R	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
☐ COSM6252	EGFR	p.G719S	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)

⑩

- ① **Export:** Download a Excel™ file of the sample comparison **Fusions** table.
- ② The QC status of the sample.
- ③ The sample name.
- ④ The date the sample was analyzed.
- ⑤ The variant type. In this example: **SNVs/Indels**.
- ⑥ **Variant ID:** The name of the hotspot as defined in the Browser Extensible Data (BED) file.
- ⑦ **Gene:** The gene name.
- ⑧ **AA Change:** Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature.
- ⑨ **Call:** Indicates the presence or absence of a SNV/INDEL variant.
- ⑩ Table slider. Use to extend table to view more samples.

The following table lists and describes the default information shown in the comparison tables.

Column	Description
Fusions comparison tables	
Variant ID	The name of the hotspot as defined in the BED file.
Oncomine Driver Gene	For fusion class variants, the 5' gene that drives proliferation of expression.
Read Counts	The frequency that the fusion was detected in the sample.
Call	The presence or absence of a fusion for the specific hotspot.
Read counts per million	The number of fusion read counts detected per number of total reads (in millions).
SNVs/Indels comparison tables	
Variant ID	The name of the hotspot as defined in the BED file.
Gene	The gene name.
AA Change	Identification of the amino acid change using HGVS nomenclature.
Call	Indicates the presence or absence of an SNV/Indel variant. See SNVs/Indels section for more information.
CNVs comparison tables	
Variant ID	The name of the hotspot as defined in the BED file.
Gene	The gene name.

(continued)

Column	Description
Copy Number	The frequency that the fusion was detected in the sample. Note: Copy Number is not displayed for TNA CNV result comparisons.
Call	Indicates the presence or absence of a CNV.
CNV Ratio	Ratio of CNV amplicons after outlier removal.

Create time series graphs

You can create line graphs to view changes and trends in variant results over time.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click the checkboxes next to the sample names of interest.

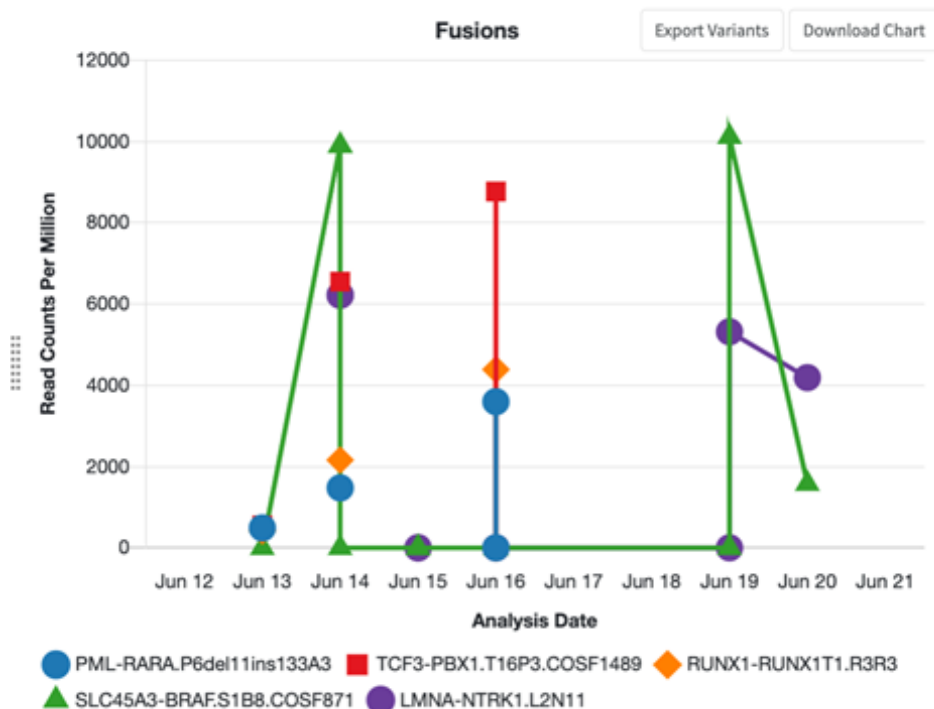
Note: Select **Filter Runs by** ▶ **Sample Status** ▶ **Completed** to limit the entries that appear in the **Sample Results** screen.

3. Above the list of sample results, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. **OncoPrint™ Extended** is the default filter for the data that appear in the tables.

4. (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different filter chain to the variants that are shown in the tables. For more information, see “Filter results” on page 174.
5. In the **Compare Samples** table, select the checkboxes next to the **Variant IDs** that you want to view in a plot.
The datapoints are added to the line graph with a live preview that is shown to the right of the **Compare Samples** table.
6. Download a file of the line graph and the information in the variant table that was used to create the graph, if needed.
 - Click **Export Variants** to download the information in the **Compare Samples** variant table for the selected variants in XLSX format.
 - Click **Download Chart** to download a PNG file of the time series line graph.



Create and customize comparison plots

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click the checkboxes next to the sample names of interest.

Note: Select **Filter Runs by** ▶ **Sample Status** ▶ **Completed** to limit the entries that appear in the **Sample Results** screen.

3. In the upper right corner of the screen, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. **OncoPrint™ Extended** is the default filter for the data that appear in the tables.

4. (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different filter chain to the variants that are shown in the tables. For more information, see “Filter results” on page 174.
5. In the **Compare Samples** table, select the checkboxes next to the **Variant IDs** you want to view in a plot.
The datapoints are added to the line graph with a live preview that is shown to the right of the Compare Samples table.
6. (Optional) To customize the chart, click **Chart Options** in the upper right corner of the screen.

7. In the **Preferences** dialog box, change the parameters to customize the plot.

X-Axis Options

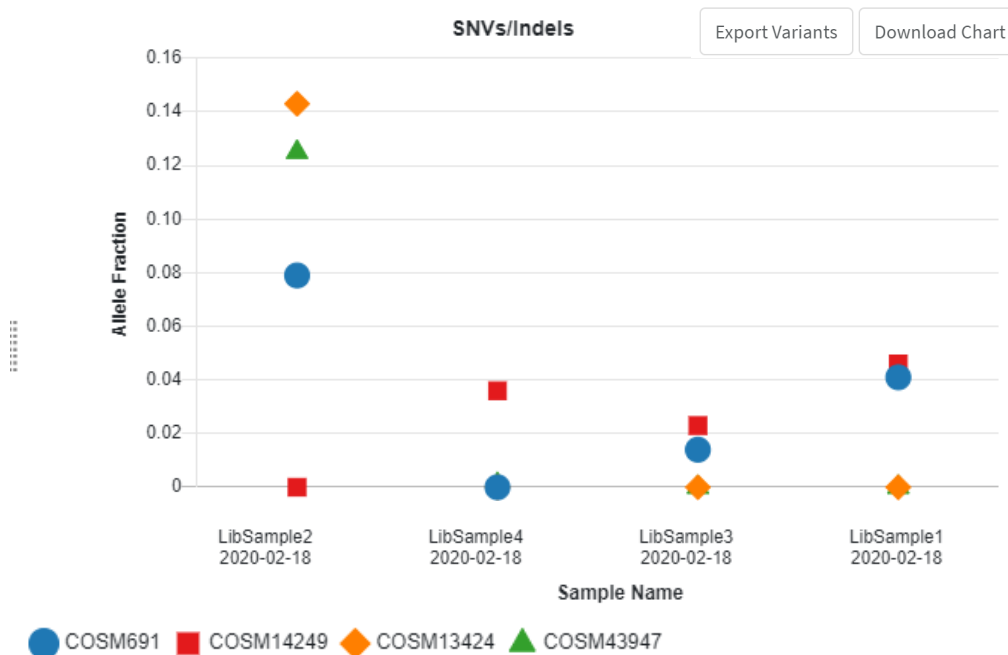
Option	Description
Sample Name	The unique identifier for a sample.
Analysis Date	The date of the sequencing run. The analysis date or run completion date is typically included in the Sequencing run details of the variant report.
Collection Date	The date that the sample was collected.

Y-Axis Options

Option	Description
SNVs/Indels	See “View results for SNVs/INDELs” on page 141.
Fusions	See “View results for fusions” on page 146.
CNVs	See “View results for CNVs” on page 152.

Note: Y-Axis options depend on the assay or assays used to analyze the samples.

8. Click **Apply** to update the chart with the new selections.
9. If needed, download a file of the image and the information in the variant table that was used to create the graph.
- Click **Export Variants** to download in XLSX format the information in the **Compare Samples** variant table for the selected variants.
The columns that are selected in **Table Columns** determine the headings that appear in the spreadsheet. If you change the column selections, you can download a new spreadsheet with updated headings.
 - Click **Download Chart** to download a PNG file of the comparison plot or time series line graph.



View and customize comparison tables

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click the checkboxes next to the sample names of interest.

Note: Select **Filter Runs by** ▶ **Sample Status** ▶ **Completed** to limit the entries that appear in the **Sample Results** table.

3. In the upper right corner of the screen, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. **OncoPrint™ Extended** is the default filter for the data that appear in the tables.

4. (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different chain to the variants that are shown in the tables. For more information, see “Filter results” on page 174.

5. (Optional) To customize the table or tables, click **Table Columns** in the upper right corner of the screen. Then, select or deselect which columns to add or remove.

The available options depend on the assay or assays used to analyze the samples. For descriptions of options, see the following sections.

Option	Description
SNVs/Indels	See “View results for SNVs/INDELs” on page 141.
Fusions	See “View results for fusions” on page 146.
CNVs	See “View results for CNVs” on page 152.

Selected options are added as column headings to the **Fusions**, **SNVs/Indels**, and **CNV** comparison table with live preview.

6. Click **Table Columns** again to close the dialog window.
7. Click **Export Variants** to download an XLSX spreadsheet file of the Fusions, SNVs/Indels, or CNVs variant comparison table if needed.

Compare immune repertoire results

You can compare immune repertoire variant results with Genexus™ Software. A description of the comparison of results generated from all other assays is not included here. For more information, see “Compare sample results” on page 184.

Compare the immune repertoire between samples

Genexus™ Software can perform multi-sample (or cross-sample) analyses to compare the immune repertoire between samples.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click the checkboxes next to the sample names of interest.
Select **Filter Runs by ▶ Sample Status ▶ Completed** to limit the entries that are displayed on the **Sample Results** screen.
The immune repertoire comparison applies only to samples that are sequenced with the system-installed OncoPrint™ TCR Beta LR - GX5 - RNA assay (or any other user-defined or system-installed immuno-oncology assay).
3. In the upper right corner of the screen, click  **Compare**.
 **Compare** is unavailable until at least two samples are selected.
The **Overview of Repertoire Sequencing Results for Selected Samples** table is displayed.

Results / Compare

Search Go Join Type: CDR3 NT [Compare Samples](#)

Overview of Repertoire Sequencing Results for Selected Samples

Metrics indicate the total number of reads reported after data processing, the number of clones detected, and the Shannon Diversity and Evenness of detected clones. Click on sample name to view detailed report. Data visualizations, clone summary and repertoire metrics are available via Download button. To track clones across samples, use the checkbox to select two or more samples, then click Compare Samples.

☐	Analysis	Sample	Barcode	Project	Reads	Clones	Shannon Diversity	Evenness	Sequenced On	Actions
☐	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	JP_Sample3	IonDual_0122	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	253601	13682	9.4818	0.6901	May 29 2019 06:26 AM	Download
☐	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	JP_Sample1	IonDual_0121	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	212790	11612	9.2409	0.6843	May 29 2019 06:26 AM	Download
☐	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	JP_Sample2	IonDual_0124	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	266390	14113	9.3596	0.679	May 29 2019 06:26 AM	Download
☐	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	JP_Sample4	IonDual_0123	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_8min_extension_283day20...	241379	13160	9.4192	0.6883	May 29 2019 06:26 AM	Download

1 - 4 of 4 items

- (Optional) Click the hyperlinked sample name to open the Immune Repertoire Results for that sample.
- Click the checkboxes next to the analyses that you want to compare.
- In the dropdown list at the top of the screen, select how you want to compare clones across the sample. The options that are available depend on the immune repertoire assay.

Option	Description
CDR3 NT	Compare the clones based on the CDR3 nucleotide sequence. If the nucleotide sequence is shared across the samples, those are considered identical clones. This is the least stringent comparison option.
Variable Gene + CDR3 NT	Compare the clones based on the rearrangement of the variable gene plus the CDR3 nucleotide sequence.
Variable Gene + CDR3 NT + Variable Gene SHM Locations	Compare the clones based on the rearrangement of the variable gene plus the CDR3 nucleotide sequence plus somatic hypermutation (SHM) locations.
Variable Gene + CDR3 NT + Variable Gene SHM Locations + Isotype	Compare the clones based on the rearrangement of the variable gene plus the CDR3 nucleotide sequence plus somatic hypermutation (SHM) locations plus isotype. This is the most stringent comparison option.

- Click **Compare Samples** to generate a comparison table and plots.
Compare Samples is unavailable until at least two samples are selected.

8. View variable comparison results in the **Comparison Table** tab.

Results / Compare

Comparison Table **Comparison Plots**

Search Go

②

Variable	Joining	CDR3 AA	CDR3 NT	R_2019_05_28_16_19_10_sl...	R_2019_05_28_16_19_10_sl...	R_2019_05_28_16_19_10_sl...	R_2019_05_28_16_19_10_sl...
① TRBV3-1	TRBJ2-1	ASSQQLGEQF	GCCAGCAGCCAACAATTA...	0.1652714	0.1639175	0.1765607	0.1731592
TRBV3-1	TRBJ2-3	ASSQDGGQNTDTQY	GCCAGCAGCCAAGATGG...	0.0976337	0.111556	0.1141034	0.1087709
TRBV28	TRBJ1-2	ASSLHHKSNYGYT	GCCAGCAGTTTACATCAC...	0.0263682	0.0240425	0.0236533	0.0267215
TRBV29-1	TRBJ2-2	SIHQNTGELF	AGCATCATAATTGAGAAC...	0.0230953	0.0178862	0.0158377	0.0145415
TRBV7-9	TRBJ2-7	ASSRGPAYEQY	GCCAGCAGTCGAGGGCC...	0.0135291	0.0139386	0.0064417	0.0074447
TRBV28	TRBJ2-7	ASSFLGAGGLYEQY	GCCAGCAGTTTCTTGGT...	0.0135291	0.0133606	0.0124404	0.0117367
TRBV29-1	TRBJ2-1	SVEDSLASGSGEQF	AGCGTTGAAGATTCTTTG...	0.0103825	0.0085906	0.0070686	0.0068109
TRBV2	TRBJ1-2	ASTEWSGNVGYT	GCCAGCACTGAATGGTCC...	0.0089116	0.0142535	0.0111303	0.0106844
TRBV11-2	TRBJ2-3	ASSLTALGRSPDTQY	GCCAGCAGCTTAACCGCC...	0.0084069	0.0069364	0.0080709	0.0085592
TRBV5-1	TRBJ2-2	ASSLGFNTGELF	GCCAGCAGCTTGGGATT...	0.0073344	0.009789	0.0081009	0.0089362

1 2 3 4 5 6 7 8 9 10 ... 10 items per page 1 - 10 of 43498 items

- ① Each row represents a clone.
- ② The frequency of the clone for the listed sample.

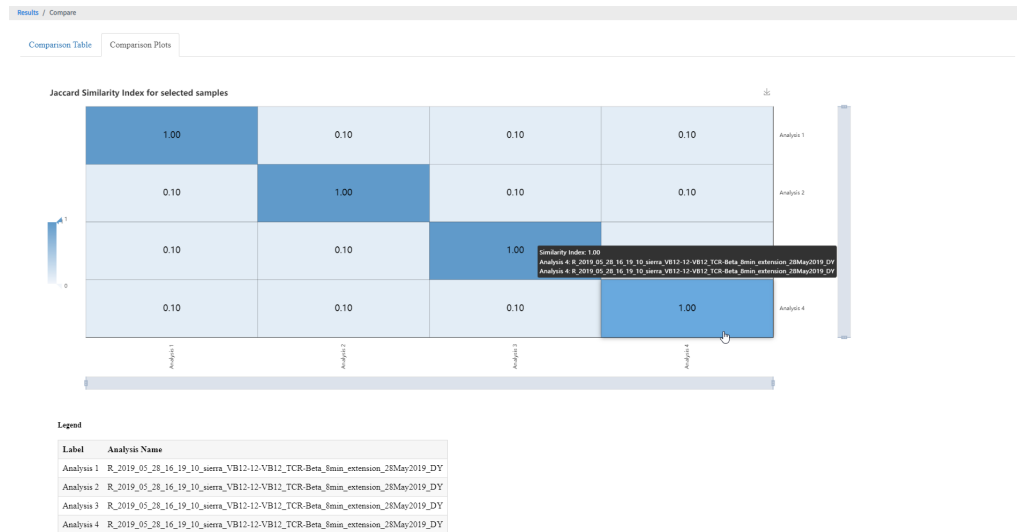
9. View comparison plots in the **Comparison Plots** tab.

Option	Description
Comparisons between 2 analyses.	An interactive scatter plot for the selected analyses is shown in the Comparison Plots tab.
Comparisons between more than 2 analyses.	A Jaccard Similarity Index for the selected analyses is shown in the Comparison Plots tab.

- The interactive scatter plot indicates the frequency of clones across two analyses. For more information, see “Example correlation and proportion of shared clones” on page 194.
- The Jaccard similarity index is determined for each pairwise comparison and is displayed in heat map format. For more information, see “Example Jaccard Similarity Index for selected samples” on page 194.

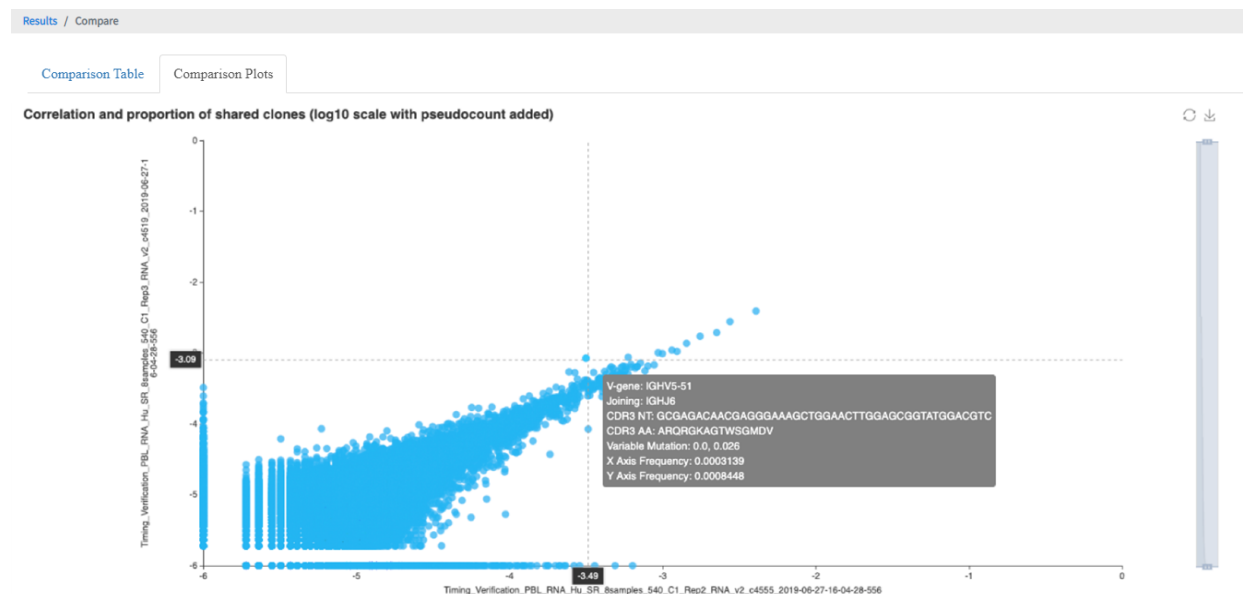
Example Jaccard Similarity Index for selected samples

This is an example of a **Comparison Plots** tab showing the Jaccard Similarity Index for selected samples. The Jaccard similarity index is determined for each pairwise comparison and displayed in heatmap form. The Jaccard Similarity Index ranges from 0 to 1 and is calculated as the total number of shared clones divided by the total number of distinct clones across two samples.



Example correlation and proportion of shared clones

This is an example of a **Comparison Plots** tab scatterplot. The illustration indicates the frequency of clones across two samples. Frequency values are log10 transformed with a pseudocount frequency of 1E-6 added to each value. Place the pointer over a point to see the CDR3NT and AA sequence of a clone, the variable mutation, and the frequency in either sample.



Upload results to another software account

Role—System administrator, organization administrator and manager, operator.

You can upload sample results files, in BAM or CRAM format, to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software.

Before you can upload results files to another sequencer, you must register the account that you can use for data uploads. For more information, see “Register Genexus™ Software accounts” on page 354.

Before you can upload results files to Ion Reporter™ Software, you must configure an Ion Reporter™ Software account.

- To configure an Ion Reporter™ Server account, see “Register an Ion Reporter™ Server account” on page 356.
- To configure an Ion Reporter™ Software on Connect account, see “Register an Ion Reporter™ Software on Connect account” on page 355.

Upload results files to another sequencer

Role—System administrator, organization administrator and manager, operator.

When a run completes successfully in Genexus™ Software, you can manually upload the sample results (BAM or CRAM files) to another Genexus™ Integrated Sequencer.

Tip: When you create a run plan, select the **Upload BAM/CRAM files to Server** checkbox in the **Setup** step to automatically upload sample results files to another sequencer when a run is complete.

Before you can upload results files to another sequencer, you must register the account that you can use for data uploads. For more information, see “Register Genexus™ Software accounts” on page 354.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **BAM Uploader**.
3. In the **Upload Samples to Server** dialog box, make the following selections.
 - a. From the **Configured Account** list, select the Genexus™ Software account and the software version.
 - b. In the **Select** column, select one or more assays from the list to upload the results files from the selected assays.
 - c. In the **Genexus Workflow** column, select whether you want analysis to start on the selected sequencer immediately.
 - Select **Upload Only** to upload the sample results (BAM and CRAM files) to the software.
 - Select an assay of interest to upload the sample results (BAM and CRAM files) and automatically start analysis with the selected assay on the sequencer.
 - d. Click **Upload**.

The results are uploaded to another sequencer.

Upload results files to Ion Reporter™ Software

Role—System administrator, organization administrator and manager, operator.

When a run completes successfully in Genexus™ Software, and the sample results were not automatically uploaded to Ion Reporter™ Software, you can manually upload the results (BAM or CRAM files) to Ion Reporter™ Software for further analysis.

You can also upload the samples to Ion Reporter™ Software, then use a user-defined Ion Reporter™ Software analysis workflow to view analysis results. Or you can upload sample results files to run assays that are not yet available on the Genexus™ Integrated Sequencer.

Note: To automatically upload results files when a run is complete, select the **Upload BAM files to Ion Reporter™ Software** checkbox in the **Setup** step of planning a run.

Before you can upload results files to Ion Reporter™ Software, you must configure an Ion Reporter™ Software account.

- To configure an Ion Reporter™ Server account, see “Register an Ion Reporter™ Server account” on page 356.
- To configure an Ion Reporter™ Software on Connect account, see “Register an Ion Reporter™ Software on Connect account” on page 355.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **BAM Uploader**.
3. In the **Upload Samples to Server** dialog box, make the following selections.
 - a. From the **Ion Reporter™ Software Account** list, select the Ion Reporter™ Server account.
 - b. In the **Select** column, select one or more assays from the list to upload the results files from the selected assays.
 - c. In the **Ion Reporter Workflow** column, do one of the following:
 - Select **Upload Only** to upload the sample results (BAM files) Ion Reporter™ Software. You must sign in to Ion Reporter™ Software, and then launch an analysis before analysis results are available in the software.
 - Select an appropriate Ion Reporter™ Software analysis workflow to upload the sample results (BAM files) and automatically launch the selected analysis workflow in Ion Reporter™ Software. Analysis results are available when you sign into Ion Reporter™ Software.
 - d. Click **Upload**.

The sample results are uploaded to Ion Reporter™ Software.

Review and download audit records for sample results

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can review and download the entire history of sample results in Genexus™ Software. An audit trail lists the records of actions for the selected sample results. Each time-stamped audit record shows the original values and any new values to show changes made or actions completed for the sample results.

The following actions generate audit records for a sample result.

- Sign-off on sample results.
- Run a plugin for sample results after a sequencing run.
- Generate a report for sample results.
- Change the filter chain or gene list that is applied to sample results.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample** screen, place the pointer over the row of the sample results of interest, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to review details for each audited action.
4. (Optional) Click  **Download** to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.

Run results

Use the **Results / Run Results** screen to view a list of runs that were performed on the Genexus™ Integrated Sequencer.

To view runs that are performed on the Genexus™ Purification System with a **Purification Only** run plan, see instead “Review purified samples” on page 96. To view verification runs that are performed on the sequencer, see “Verification results” on page 216.

Tip: You can filter the list of run results by clicking  (**Filter**) in a column of interest, then entering a full or partial run or assay name, or other applicable filter term. See “Find a run result” on page 211 for information about how to find run results of interest.

The following run information is shown in the **Results / Run Results** screen.

Column	Description
Run Name	The unique name of the run given when it was created in the software. Click a run name to open the Run Summary . For more information, see “Run summary details” on page 201. Runs that are reanalyzed are listed with ↺ (Reanalysis) after the run name. For more information, see “Reanalyze a run” on page 219.
Assay Name	The name of the assay selected for the run.
Run Status	The status of the run. For example, Analysis In Progress , Executing Plugins , Analysis Completed , Terminated , Archival: In Progress , Purification In Progress , or Purification Completed . For integrated purification runs that use a Sample to Result run plan and have a status of Purification In Progress , Purification Complete , or for failed or aborted purification runs, place the pointer over the shaded number, in the Status column to view the status of each purification batch for the run. The shaded number, such as 1 or 2 , represents a purification batch for the run. For example, a status can be 1 Completed , 2 Started , and 3 Planned , which indicates that the first batch is complete, the second batch has started, and the third purification batch has not yet started, but is planned.
Total Samples	The total number of samples in a run.
PCR Plate Number	A unique identifier for the 96-well plate used for library preparation and templating. For more information, see the Genexus™ Software help system or the user guide available at http://www.thermofisher.com/ngssoftwaresupport .
Started On	The date and time when the run was started.
Updated On	The date and time when the last action was completed on the run.

Users with appropriate permissions can perform the following actions in the **Results / Run Results** screen.

Option	Description
Actions	Available action links for a run are shown when you place the pointer over the row of a run. The actions that are available depend on the type of run. • Delete—Delete the run. • BAM Uploader—Upload run information to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software for further analysis. BAM Uploader is not available for BAM or CRAM run results, or for runs in which BAM or CRAM files have been removed. For more information, see “Upload results files to another sequencer” on page 195 and “Upload results files to Ion Reporter™ Software” on page 196. • Audit—View the audit trail for the run. For more information, see “Review and download audit records for a run plan” on page 113. • CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see “Download a Customer Support Archive” on page 586. • Assign PCR Plate—Enter a unique identifier for the 96-well plate used for library preparation and templating. For more information, see “Assign PCR Plate” on page 210.
Actions	These actions are available only for Sample to Result runs. • View Plan—View detailed run plan information. For more information, see “Run plan details” on page 110. • Review—Review samples that do not have a concentration within a specified threshold after purification, but before library preparation. For more information, see “Review purified samples for a Sample to Result run” on page 94. • Abort—Abort a run after purification, but before sequencing. This action is available when the run status is Purification Review Required, or when the run status is Purification Completed and some purification samples have been excluded from sequencing.

View the Run Summary

Role—System administrator, organization administrator and manager, operator.

The **Run Summary** provides an overview of the run. The information that is displayed includes the name of the assay used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information. Metrics from sample purification are also provided, if applicable. The information that is available depends on the run.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Name** column, click the run name of interest.
3. In the **Run Summary** tab, review the run information.

Action	Procedure
View the assay metrics.	In the Assays section, click the assay name of interest.
View the sample locations in an image of a 96-well sample plate.	In the Sample Locations section, click PCR Plate View .
Reanalyze a run with a new assay.	In the upper right corner of the screen, click ... (More Options) ▶ Reanalyze . For more information, see “Reanalyze a run” on page 219.
Download customer support archive (CSA) log files for the run to help with troubleshooting.	In the upper right corner of the screen, click ... (More Options) ▶ CSA . For more information, see “Customer Support Archive” on page 417.
Upload results to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software for further analysis.	In the upper right corner of the screen, click ... (More Options) ▶ BAM Uploader . For more information, see “Upload results files to another sequencer” on page 195 and “Upload results files to Ion Reporter™ Software” on page 196.
Download the run summary files.	In the upper right corner of the screen, click ... (More Options) ▶ Download Files , then in the Download Files dialog box, select files to download in a ZIP folder to your computer. You can choose from variant files, reports, sequencing results, plugin files, audit and log files, and troubleshooting files.
View the audit record trail for selection of variants, variant classifications, and notes for variants.	In the upper right corner of the screen, click ... (More Options) ▶ Variant Audit . The information is available only for results that include variant classifications.
View the run report.	Click the Run Report tab.

Run summary details

The run summary shows run information, assays used in the run, sample locations, metrics from sample purification, information about the reagents used in the run, primer tube positions, and instrument information.

Depending on the type of the run and the selections made in the run plan, the run information can include the following items.

Run information

Item	Description
Run Name	The name of the run.
Started On	The date and time when the run was started.
Completed On	The date and time when the run was completed.
Run Status	The status of the run.
Report Template	If the option to generate a report was enabled in the run plan, the report template used is listed.
Chip Type	The semiconductor sequencing chip used in the run, such as the Ion Torrent™ GX5™ Chip.
Library Chemistry	The type of library chemistry used in the run, such as Ion AmpliSeq™ or Ion AmpliSeq™ HD.
Server	The Ion Reporter™ Software account or the Genexus™ Software account and the software version that was selected for uploading sample files, in BAM or CRAM format, when the run is complete.
Run Type	The type of run. For example, Nucleic Acid to Result.

Sequencing instrument information

Item	Description
Instrument Name	The name of the sequencer.
Instrument Serial Number	The serial number of the sequencer.
Status	The status of the sequencing portion of the run.
Operator	The name of the person who was signed in the Genexus™ Integrated Sequencer when the sequencing run was performed.
Start Date	The date and time when the run was started on the sequencer.
Completion Date	The date and time when the run was completed on the sequencer.
Sequencing Flows	The number of flows performed by the sequencer.

Purification instrument information

This information applies only to sequencing runs that use a **Sample to result** run plan.

Item	Description
Purif. Instrument	The name of the Genexus™ Purification System.
Instrument Serial Number	The serial number of the purification instrument.
Status	The status of the purification portion of the run for the batch.
Operator	The name of the person who was signed in the purification instrument when purification was performed.
Start Date	The date and time when the batch was started on the purification instrument.
Completion Date	The date and time when the batch was completed on the purification instrument.
Purif. Batch	The nucleic acid isolation batch.

Assays table

Item	Description
Assay Full Name	The name of the assay used in the run. Click the assay name to display the details of the assay.
Assay Name	An abbreviated name of the assay or assays used in the run.
Analysis Version	The version of the assay used for analysis.
Research Application	The research application for the assay, such as DNA or DNA and Fusions .
Lane	The chip lane or lanes used in the sequencing run for the assay.
Total Samples	The total number of samples sequenced for the assay. A single sample can correlate with multiple wells on the plate for some assays when multiple nucleic acid types are contained within the sample.
Updated Annotations	The information that is shown depends on the settings and the type of the assay. • The name and version of the annotation source that is applied to the sample result is shown when an assay is configured to use the latest annotations but the annotation that is applied to the results is not the latest annotation source. • No information is shown in the column (the value is blank) when the latest annotation set version is applied to the result, regardless of whether the assay is configured to use the latest annotation source. • No information is shown in the column (the value is blank) when an assay is configured to not use the latest annotation source, and instead uses a fixed version of an annotation source. • N/A is shown when an assay does not use an annotation set.

Purification Samples table

This information applies only to sequencing runs that use a **Sample to result** run plan.

Item	Description
Batch ID	When the run includes more than one purification batch, the Batch ID is listed. The Batch ID , such as 1 or 2 , indicates the purification group for each sample.
Sample Name	The unique identifier created when the sample was entered into the software.
Sample Type	A term that describes the sample, for example, FFPE or Blood (Plasma) .
Nucleic Acid Type	DNA , RNA , or DNA+RNA .
Conc. (ng/μl)	The concentration of the sample measured by the purification instrument. In some instances, such as when purification is in progress or when the sample is a no-template control, the values is listed as N/A . If the concentration is not within the QC concentration range specified by the assay, a Quantity Not Sufficient (QNS) alert is displayed.
QC Conc. Range (ng/μl)	The QC concentration range for the assay.
Batch Status	The run status of a purification batch. The status can be N/A , Planned , Review , Started , Aborted , or Completed .
Archive Position	The sample positions in the archive plate. Sample positions can be in rows A to D and in columns 1 to 8 .
Library Prep	An indicator of whether the sample is selected for sequencing. ✓ (Checkmark)—Indicates that the sample is selected for sequencing. ⌀ (No icon)—Indicates that the sample is not selected for sequencing. N/A—Indicates that purification has not started or is in progress.

Sample Locations table

Item	Description
PCR Plate View	Click to view the sample locations in an image of a 96-well sample plate.
Well Pos.	The well position indicates the location of the sample on the plate.
Library Batch ID	The unique identifier created for the library batch. This information is available only for Library to Result runs.
Sample Name	The unique identifier created when the sample was entered into the software.
Nucleic Acid Type	The sample nucleic acid type, such as DNA , RNA , or TNA .
Vol. (μl)	The volume of the sample.
Conc. (ng/μl)	The concentration of the sample.

Sample Locations table (continued)

Item	Description
Dilution Factor	The dilution factor of the sample.
Kit Barcodes	The barcode for the kit used for nucleic acid extraction, if applicable.
Barcode	The name of the barcode adapter or adapters that are associated with the sample, for example, IonHDdual_0101 .
Assay Name	The assay that was used to sequence the sample for the indicated well position.
Library Position	The plate location of libraries. For completed runs that were planned using samples, the Library Position indicates the location of unused libraries that can be sequenced by planning and initiating a library run.

Reagents table

Item	Description
Consumable	Consumables, such as the PanelKit and LibraryKit , that are used in the run are listed.
Barcode	The consumable barcode, if applicable.
Part #	The consumable part number, if applicable.
Expiration Date	The consumable expiration date, if applicable.
Lot #	The consumable lot number, if applicable.

The **Primer Tube Positions** table shows an image that indicates the locations of the primer pool tubes.

View the Run Report

Role—System administrator, organization administrator and manager, operator.

The **Run Report** provides detailed sequencing metrics about a run, such as the loading of the wells on the chip, and the number of reads that were filtered out. For runs that use multiple assays, sequencing metrics are provided for each assay in the run.

1. Click **Results ▶ Run Results** to view sequencing results for a sample.
2. In the **Run Name** column, click the run name of interest.

Action	Procedure
View the Run Report	Click the Run Report tab.
View the sequencing metrics for a single assay	In the Select Assay dropdown list at the top of the screen, select the assay of interest.

For more information, see “Sequencing metrics” on page 205.

Sequencing metrics

The run report provides detailed information, such as chip metrics for the run, well, and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics in the **Run Samples** table. Read data for individual samples for the assay are listed in the **Run Samples** table.

Barcode-specific metrics for barcodes that are included in the run are listed in the **Barcodes With Reads Reported** table, which follows the **Run Samples** table. The CSA file for the run contains information for barcodes that are not assigned to samples in the run. Information in the CSA file can help you troubleshoot results, if needed. For more information, see “Download a Customer Support Archive” on page 586.

Run Report

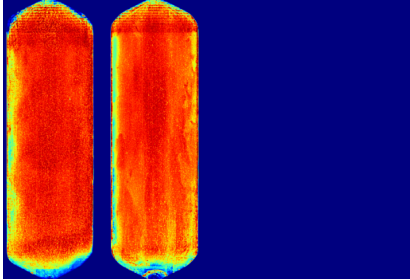
The Run Report table lists the following information.

Metric	Description
Loading	The number and percentage of total addressable wells on the chip that contain an ISP.
Enrichment	The number and percentage of wells ISPs that contain live ISPs.
Library	The number and percentage of wells with live ISPs that contain Library ISPs.
Combined Final Reads	For runs that include resequencing, the combined final reads is the sum of library reads from both sequencing and resequencing.
Final Reads	Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement.
Total Bases	The number of filtered and trimmed base pairs that are reported in the output BAM file.
Combined Total Bases	For runs that include resequencing, the combined total bases is the sum from sequencing and resequencing.
Raw Read Accuracy	The raw read accuracy across each individual base position in a read calculated as, $(1 - [\text{total errors in the sequenced reads}] / [\text{total bases sequenced}]) \times 100$. Raw read accuracy is measured at each base across the length of the read and is based on 1x sequencing coverage; raw read accuracy is <i>not</i> based on consensus accuracy across multiple reads for the same base position.
Wells with ISPs	The number of wells that contain an ISP.
Unenriched	The number and percentage of wells with ISPs that do not contain live ISPs.
Total Addressable Wells	Wells on the chip that can be physically reached by a library.
Empty Wells	The percentage of total addressable wells on the chip that do not contain an ISP.

Run Report (continued)

Metric	Description
Wells with Live ISPs	Loaded wells with ISPs with a signal of sufficient strength and composition to be associated with the library or control fragment key.
Wells with Library ISPs	Loaded wells with live ISPs with a key signal that is identical to the library key signal.
Control ISPs	Loaded wells with live ISPs with a key signal that is identical to the control fragment key signal.
Filtered out	The total percentage of filtered reads, or the sum of the percentages of polyclonal, low quality, and adapter dimer reads.
Polyclonal	Wells with a live ISP that carries clones from two or more templates. To view polyclonal metrics, mouse over the first low quality portion (gray) of the Final Reads bar plot. Final Reads 58.39% ? Filtered out 41.61% 30,735,120 Final Reads(30,735,120) / Wells with Library ISPs(52,635,124) Polyclonal: 15935106 (30.27%)
Low Quality	Loaded wells with a low or unrecognizable signal. To view polyclonal metrics, mouse over the second low quality portion (gray) of the Final Reads bar plot. Final Reads 58.39% ? Filtered out 41.61% 30,735,120 Final Reads(30,735,120) / Wells with Library ISPs(52,635,124) Low Quality: 3208258 (6.1%)
Adapter Dimer	Loaded wells with a library template of an insert size less than 8 bases. To view adapter dimer metrics, place the pointer over the lightest gray portion of the Final Reads bar plot. Final Reads 43.57% ? Filtered out 56.44% 52,494,934 Final Reads(52,494,934) / Wells with Library ISPs(120,492,758) Adapter Dimer: 10862794 (9.02%) Note: In assays using Ion AmpliSeq™ HD library chemistry, adapter dimer reads represent a small proportion of total reads. Place the pointer over the right end of the Final Reads bar.

Run Report *(continued)*

Metric	Description
Loading Density	A visual representation of chip loading. Red color indicates areas of higher density of loading. Blue color indicates areas of lower density of loading. The following example shows a sequencing experiment where two lanes on the chip are uniformly loaded with ISPs. 

Run Samples

The **Run Samples** table lists read data for each individual sample in the assay.

Column	Description
Sample Name	The unique identifier created when the sample was entered in the software.
Nucleic Acid Type	The sample nucleic acid type, such as DNA , RNA , DNA+RNA , or TNA .
Barcode	The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample.
Total Reads	The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample.
Mean Read Length	The average length, in base pairs, of usable library reads for each sample.
≥Q20 Bases	The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample.
Mapped Reads	The number of reads that are mapped to the reference file.
On Target Reads	The percentage of sequencing reads mapped to any target region of the reference.
Mean Depth	The average number of reads of all targeted reference bases.
Uniformity	The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage.
Read Length Histogram	A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). 

Barcodes with Reads Reported

The **Barcodes with Reads Reported** table lists barcode-specific metrics

Column	Description
Barcode	The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample.
Total Reads	The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample. The reads are independent of length reported in the output BAM file.
Mean Read Length	The average length, in base pairs, of usable library reads for each sample.
≥Q20 Bases	The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample.
Read Length Histogram	A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). 

Download information about run samples

Information about each sample in a run, such as the barcode for the sample and the total reads, are shown in the run samples table. You can download the information shown in the run samples table.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. Click the **Run Report** tab, then scroll to the **Run Samples** table.
4. In the upper right corner above the **Run Samples** table, click  **Export to Excel**.

An XLSX file that contains the information shown in the run samples table is downloaded.

Read length histograms are not included in the file.

Download barcodes with reads reported

Metrics for the barcodes that are included in a run are listed in the barcodes with reads reported table. You can download the information shown in the barcodes with reads reported table.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. Click the **Run Report** tab, then scroll to the **Barcodes with Reads Reported** table.
4. In the upper right corner above the **Barcodes with Reads Reported** table, click  **Export to Excel**.

An XLSX file that contains the information shown in the barcodes with reads reported table is downloaded.

Read length histograms are not included in the file.

View resequencing metrics

For assays that include resequencing, you can view resequencing metrics when a run is complete.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In **Select Assay**, select the assay name of interest.
The assay metrics for the sequencing run are shown.
4. Click **Resequencing Metrics**.
The resequencing metrics for the assay are shown.

Retain run results

Role—System administrator, organization administrator.

Administrators can retain run results in Genexus™ Software. When you retain a run result, run data, including the run result, .wells files, and intermediate files, are retained and cannot be deleted. When a run result is retained, it overrides the automatic deletion settings in the software. For more information, see “Set up a schedule to delete run result files” on page 380.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to retain.
3. Click ☐ **Retain Data**.

Run results that have been retained are denoted with ☐ after the run name in the **Run Results** screen. The run result is retained in the system and cannot be deleted. To revert the run to a run that is not retained, see “Release run results” on page 210.

Release run results

Role—System administrator, organization administrator.

Run results that have been retained in Genexus™ Software can be released. For more information, see “Retain run results” on page 209.

When you release run results, you can then delete the run results. The released run results, the associated .wells files, and intermediate files are available to be auto-deleted according to system settings. For more information, see “Set up a schedule to delete run result files” on page 380.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, click **Release Data** in the row of the run that is retained.
Run results that have been retained are denoted with  **(Retained)** after the run name.
3. Click **Yes** to confirm that you want to proceed with the data release.

The run result is released and is no longer denoted with  **(Retained)** after the run name. The run result can be deleted manually or with automatic deletion settings in the software.

Assign PCR Plate

Role—System administrator, organization administrator and manager, operator.

Genexus™ Software lets you track and associate a run with the PCR plate used in the run. The PCR plate is the 96-well plate that is used for library preparation and templating. You can assign a unique identifier, a **PCR Plate Number**, to runs that have a status of **Library Preparation Completed**, **Sequencing Completed**, or **Run Completed**. The PCR plate number that you enter is shown in the **Run Results** screen and if needed, can help you track libraries and troubleshoot. Later, if you sequence the remaining libraries in a different sequencing run and assign the PCR plate number to the run, you can easily search for and find all run results associated with the libraries in the PCR plate.

Note: You should assign a PCR plate to a run if there are leftover libraries that you want to use at a later time to perform a Library to Result run. You can look up the PCR plate number on completed runs to get the library and barcode mapping, then enter the same Barcode information into the Library to Result run plan.

For more information to find a run with a search of the PCR Plate Number, see “Search for run results” on page 211.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Assign PCR Plate number**.
3. In the **Assign PCR Plate Number** dialog box, confirm, edit, or enter the **PCR Plate Number**.
The PCR plate number must be between 1 and 10 characters. Only alphanumeric characters (numbers 0 to 9 and letters A to Z), period (.), underscore (_), or hyphen (-) are allowed. Spaces are not allowed.
4. Click **Submit** to associate the PCR plate with the run.

Find a run result

Role—System administrator, organization administrator and manager, operator.

You can search, filter, and sort information to narrow the run results that are listed in the **Results / Run Results** screen. The sort, search, and filter tools help you organize run results and find a specific run result in Genexus™ Software.

To sort the run results list, see “Sort the list of run results” on page 215.

To search for a run result by a unique identifier, such as the name of a run, see “Search for run results” on page 211.

To filter run results by a common trait, such as run results that have a status of analysis completed, see “Filter the list of run results” on page 213.

Search for run results

Run results are listed in the **Results / Run Results** screen in Genexus™ Software. The most recent run result is on the top of the list.

You can search for run results with keywords or text that are included in the name of the run name, the name of an assay applied to the run, the number of samples in the run, the date on which the run was started or updated, or by run status or the PCR Plate number. For more information about how to use the PCR Plate number to track libraries, see “Assign PCR Plate” on page 210.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name**, **Assay Name**, **Run Status**, **Total Samples**, **PCR Plate Number**, **Started On**, or **Updated On** column heading, click ▼ (**Filter**).
 - a. In the search field, enter a full or partial assay or run name or PCR plate number of at least 3 characters.

When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - b. In the filter dialog box, select from the limited list.
 - Select a checkbox next to a run or assay name to select one or more items.
 - Click **Select All** to select all items listed in the filter dialog box.

Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

- c. In the filter dialog box, click **Filter**.

In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of run results is shown in the **Run Results** screen.

3. Click  **Clear Filters** to remove all filters in the **Run Results** screen and view the full list of run results. Or, click  (**Filter**) in a column heading, then click **Clear** to remove the filter or filters that are applied to a column.

Filter the list of run results

You can apply filters to narrow search results, or shorten the list of run results when you view the list of run results in Genexus™ Software.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, select one or more filters.

Option	Description
Filter and search by a column heading shown in the run results	For more information, see “Search for run results” on page 211.
Show or hide run results that are archived	- In the dropdown list in the upper left corner, select an archive status. • Archived • Non Archived - Select All to list all run results.
Filter by the status of the run result.	- In the Run Status column heading, click ▼ (Filter) then select at least one status. • Analysis Completed • Analysis In Progress • Analysis Failed • Analysis Aborted • Analysis Stalled • User Aborted • Backup In Progress • Run Aborted • Purification Completed • Purification Run Aborted • Purification Failed • Purification Review Required • Purification In Progress • Selected on Purification Instrument • Executing Plugins • Plugins Queued • Library Preparation Aborted • Library Preparation Completed • Library Preparation Started • Selected on Instrument • Sequencing Completed • Sequencing Started • Templating Aborted • Templating Completed • Templating Started • Kit Barcodes Required • Sequencing Aborted • Kit Barcodes Review Required • Restore: In Progress • CRAM to BAM Conversion: In Progress - Click Filter. Records are listed for run results that meet the run status criteria.
Filter by the number of samples in the run.	In the Total Samples column heading, click ▼ (Filter) then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run.

Option	Description
List only run results from runs that were started on or updated on an exact date.	- In the Started On or Updated On column heading, click ▼ (Filter). - In the first dropdown list, select Is equal to. - In the calendar that appears, select the date of interest. - Click Filter. Records are listed for run results that were started on or updated on the date entered.
List only run results from runs that were started on or updated within a date range.	- In the Started On or Updated On column heading, click ▼ (Filter). - In the first dropdown list, select Is after or equal to. - In the calendar that appears, select the earliest date of the range. - In the Boolean operator dropdown list, select And. - In the second dropdown list, select Is before or equal to. - In the calendar that appears, select the latest date of the range. - Click Filter. Records are listed for sample results started on or updated on dates that meet both conditions.
List run results according to a date that meets either one condition or a second condition.	- In the Started On or Updated On column heading, click ▼ (Filter). - In the first dropdown list, select Is after or equal to. - In the calendar that appears, select the earliest date for the sample results of interest. - In the Boolean operator dropdown list, select Or. - In the second dropdown list, select Is before or equal to. - In the calendar that appears, select the latest date for the sample results of interest. - Click Filter. Records are listed for sample results for runs that were started on or updated on dates that meet at least one of the conditions.
List only run results for runs that were started on or before or updated on or before a date that you indicate.	- In the Started On or Updated On column heading, click ▼ (Filter). - In the first dropdown list, select Is before or equal to. - In the calendar that appears, select the date of interest. - Click Filter. Records are listed for run results that were started on or before the date entered.
List only run results for runs that were started on or after, or updated on or after, a date that you indicate.	- In the Started On or Updated On column heading, click ▼ (Filter). - In the first dropdown list, select Is after or equal to. - In the calendar that appears, select the date of interest. - Click Filter. Records are listed for runs that were started on or after, or updated on or after, the date entered.

In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

Click  **Clear Filters** to remove all filters and view the full list of run results.

Sort the list of run results

You can sort the list of run results to make it easier to find a run result of interest in Genexus™ Software.

By default, run results are listed in the **Runs / Run Results** screen by date. The most recent run result is at the top of the list.

You can change the order that the run results are listed to sort the run results, and reverse the order of the sorted list.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, click the **Run Name**, **Assay Name**, **Started On**, or **Updated On** heading.
The list of results is sorted based on the selected heading.
3. Click the column heading again to reverse the order.

Review and download audit records for run results

Role—System administrator, organization administrator and manager, operator.

Administrators, managers, and operators can review and download the entire history of a run results in Genexus™ Software. An audit trail lists the records of actions for the selected run results. Each time-stamped audit record shows the original run results values and any new values for each action.

The following user actions generate an audit record.

- Run plan creation
- Run status changes
- Report generation
- Report sign off
- Plugin runs

Some software processes, such as a change in run results status also generate an audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of the run results of interest, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to review details for each audited action.
4. (Optional) Click  **Download** to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.

Verification results

The following information is available in the **Results / Verification Results** screen.

Column	Description
Run Name	The name of the run that is assigned to the run when run plan is created. Click the name to open the Verification Results Details screen.
Assay Full Name	The name of the assay used in the run.
Field Engineer Name	The name of the support specialist who performed the run.
Instrument Name	The name of the sequencer that was verified.
Run Status	The current status of the full sequencing run, including analysis.
QC Status	Indicates whether a sequencing run passed or failed, based on the sequencing QC metrics selected for the assay.
Started On	The date and time that the run was started.
Updated On	The date and time that the run was last updated.

View verification run results

Role—System administrator, organization administrator and manager.

Administrators and managers can view verification run details, including the run summary, QC summary, and the reagents used in the run. Verification runs are performed during installation to validate the performance of the instrument or instruments. Sequencing verification runs are performed to validate the Genexus™ Integrated Sequencer.

If a Genexus™ Purification System is also connected, a verification run that validates both the purification instrument and sequencer is performed.

Note: Field Service Engineers (FSEs) use system-installed verification templates to validate the installation of the Genexus™ Integrated Sequencer. FSEs can view the template information but cannot modify it.

1. In the menu bar, click **Results ▶ Verification Results**.
2. In the **Verification Results** screen, view or download information for the verification run.

Option	Description
View verification results details	In the Run Name column, click the verification run name of interest to open the Verification Results Details screen. In the QC section of the Verification Results Details screen, click the > symbol to the left of the Lane column in the lane of interest to expand the results details pane.
Download a verification report that has been signed	Place the pointer over the row of the verification run of interest, then click Report .

3. You can perform the following actions for **Verification Results**. Action links are available when you place the pointer over the row of a verification result.

Option	Description
View Plan	If the run has not started on an instrument, you can view the run plan for the verification run.
Audit	Displays the list of users who created and edited the run. You can export and print information from the list from the Audit Trail dialog box.
CSA	CSA —Customer Support Archive. Click this link to download the sequencer log files. Log files contained within the CSA can be useful when troubleshooting problems with the sequencer. For more information, see “Customer Support Archive” on page 417.
Report	Download a verification report that has been signed.

Sign verification run reports

Role—System administrator.

The system administrator can sign results reports for verification runs. However, we recommend that only qualified support specialists sign performance qualification (PQ) reports.

Verification run reports can be electronically signed only when all samples pass QC analyses.

1. Click **Results ▶ Verification Results**.
2. In the **Verification Results** screen, click the name of the verification run of interest.
3. In the **Verification Results Details** screen, click  **Sign Off**.
The  **Sign Off** function is unavailable if any samples did not pass QC analysis.
4. In the **Sign Off Report** dialog box, enter or make the following selections.
 - a. Enter the password.
 - b. In **Electronic Signature**, select **Approval**.
 - c. In **Sign off Comments**, enter a comment.
 - d. In **Laboratory Comments** and **Footer Field**, enter optional information.
5. Click **Sign Off**.

The verification run report is electronically signed.

You can view, print, or download the report in a PDF file.

Verification templates

Field Service Engineers (FSEs) use the verification templates during Genexus™ Integrated Sequencer installation.

IMPORTANT! The **Assays / Verification Templates** screen lists the verification templates that are used only by FSEs during Genexus™ Integrated Sequencer installation and performance qualification testing. These templates should not be used or modified in any way.

Details about each verification template is available when you click the name of the template in the software.

- **Performance Qualification Assay**
- **System Install Qualification Assay**
- **Integrated Purification Performance Qualification Assay**

Reanalysis

If a run fails to meet one or more QC parameters defined by the assay, you can adjust the assay parameters and reanalyze a run or a sample from the run. For more information to adjust assay parameters, see “Edit the QC parameters of a locked assay” on page 262 and Chapter 8, “Create and manage assays”.

Runs can be reanalyzed with the same assay or a different assay. Reanalysis of runs can start from the alignment, basecalling, or signal processing steps. A sample can be reanalyzed starting only from the alignment step.

For more information, see “Reanalyze a sample” on page 221 and “Reanalyze a run” on page 219.

Reanalyze a run

Role—System administrator, organization administrator.

Administrators can reanalyze a run with the same assay or with a different assay in Genexus™ Software. Reanalysis can start from the alignment, basecalling, or signal processing steps. When you reanalyze a run, the reanalysis is applied to all samples in the assay.

IMPORTANT! Not all sequencing runs can be reanalyzed. Only sequencing runs that use assays that meet a set of criteria used in the software can be reanalyzed. The criterion is based on several factors, including application type, primer pool size, extraction type, flow order, barcode set, chip, application category and whether resequencing is enabled. For more information about which runs can be reanalyzed, contact a support representative, or Field Bioinformatics Specialist (FBS).

Reanalysis from the	Description
Signal processing step	The option to reanalyze a run from the signal processing step is only available when the following conditions are met. • When a run fails during the basecalling step or earlier. • When the run has not already been successfully reanalyzed. Reanalysis at the signal processing step uses DAT files.
Basecalling step	The option to reanalyze a run from the basecalling step is only available when a run or reanalysis successfully completes the signal processing step. Reanalysis at the basecalling step uses .wells files.
Alignment step	The option to reanalyze a run from the alignment step is only available when a run or reanalysis successfully completes the basecalling step. Reanalysis at the alignment step uses BAM or CRAM files.

Note:

- Administrators can reanalyze a sequencing run only if the run completed without any critical alarms or errors. If the run is aborted or produced major alarms or errors, the run cannot be reanalyzed.
- QC parameters at the limits of stringency cannot be adjusted further. If samples fail QC and you cannot adjust the QC parameters further, you can sequence the sample library again. For information to find, recover, and purify the leftover library preparations, see the Genexus™ Integrated Sequencer user guide. Alternatively, you can prepare a new library for sequencing.

The files needed for the stage of reanalysis that you select must be present in the software. For example, if the .wells files for the run have been removed from the software, you cannot reanalyze from basecalling. Instead, reanalyze the BAM and CRAM files at alignment. Administrators can manage the settings and schedule to backup and delete files and data. For more information, see “Manage backups” on page 373..

You can reanalyze a run with any compatible assay that exists in the software. Alternatively, you can create a new assay or copy the original assay that was used in a run and modify assay parameters if needed. For more information, see Chapter 8, “Create and manage assays”.

1. In the menu bar, click **Results** ▶ **Run Results**.
2. In the **Results / Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In the upper right corner of the screen, click ⋮ **(More Options)** ▶ **Reanalyze**.
4. In the **Reanalyze** dialog box, enter or select the following information.
 - a. In **Reanalysis Run Name** field, enter a reanalysis run name.
 - b. In **Start Reanalysis from** dropdown list, select **Alignment**, **Basecalling** or **Signal Processing**.
 - c. In the **Current Assay in Run** column, select the checkbox in the row of each assay that you want to reanalyze. Then, in the **Choose Assay to Reanalyze** column, select an assay for each reanalysis from the dropdown list.

Reanalyze

×

Reanalysis Run Name

Reanalysis Run 1

Start Reanalysis from

Alignment

Current Assay in Run	Choose Assay to Reanalyze	Updated Annotations
☒ Oncomine Myeloid v2 - GX5 - DNA - w5.0.0 - v0.0.2	Oncomine Comprehensive Plus - C	clinvar(20220328)

Cancel

Reanalyze

5. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Run Results** screen in the **Run Status** column and in the **Results / Sample Results** screen in the **Sample Status** screen. When reanalysis is complete, view the new results by clicking the run name corresponding to the reanalysis assay in the **Results / Run Results** screen. Runs that have been reanalyzed are appended with ↺ after the run name.

Reanalyze a sample

Role—System administrator, organization administrator.

Administrators can reanalyze a sample starting only from the alignment step. Reanalysis at the alignment step uses BAM or CRAM files. You can reanalyze all samples in a run from the basecalling or signal processing steps. For more information, see “Reanalyze a run” on page 219.

Administrators can reanalyze a sample only if the run completed without any critical alarms or errors. If the run is aborted or produced major alarms or errors, the sample cannot be reanalyzed.

The option to reanalyze a sample from the alignment step is only available when a sample or reanalysis successfully completes the basecalling step.

You can reanalyze a sample with any compatible assay that exists in the software. Alternatively, you can create a new assay or copy the original assay that was used in a run and modify assay parameters if needed. For more information, see Chapter 8, “Create and manage assays”.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of a sample of interest, then click **Reanalyze**.
Alternatively, you can reanalyze a sample when you click a sample name, then click **⋮ (More Options) ▶ Reanalyze**.
3. In the **Reanalyze Sample** dialog box, enter or select the following information.
 - a. In **Reanalysis Run Name** field, enter a reanalysis run name.
 - b. In **Start Reanalysis from** dropdown list, select **Alignment**, **Basecalling** or **Signal Processing**.
 - c. In the **Current Assay in Run** column, select the checkbox in the row of each assay that you want to reanalyze. Then, in the **Choose Assay to Reanalyze** column, select an assay for each reanalysis from the dropdown list.
4. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Sample Results** screen in the **Sample Status** screen. When reanalysis is complete, view the new results by clicking the sample name corresponding to the reanalysis assay in the **Results / Sample Results** screen. Samples that have been reanalyzed are listed in the table of sample results with the run name appended with ↻.

Data files and flow

During a Genexus™ Integrated Sequencer run, raw data (DAT) files from the first flows are available. Genexus™ Software starts processing the data, producing a 1.wells file. Basecalling is performed on the 1.wells file data, producing an unmapped BAM (uBAM) file. Subsequent analysis produces mapped reads (BAM) and variant calls (VCF) files.

The following table shows the flow of data and typical file size from runs that use a GX5™ Chip or GX7™ Chip as Genexus™ Software processes data.

IMPORTANT! Data from approximately 200 sequencing runs can be accommodated on the sequencer before disk space becomes a limiting factor and data archiving is required for runs if you ensure that backup settings are set to delete 1.wells files and intermediate files for after a cleanup threshold for disk space is reached. You can perform approximately 45 to 50 whole chip runs with a GX5™ Chip, and 20 to 25 whole chip runs with a GX7™ Chip without deleting any wells files on the system. For more information about data archival of the analysis files, see “Manage backups” on page 373.

Process	File type	File size for GX5™ Chip runs	File size for GX7™ Chip runs
Sequencing ^[1]	DAT	2 TB	2 TB
▼			
Signal Processing ^[2]	Wells	250 GB	300 GB
▼			
Base Calls (reads)	uBAM	15 GB	40 GB
▼			
Mapped Reads	BAM	15 to 20 GB	45 GB
▼			
Variant Calls	VCF	120 Kb	25 MB

^[1] The sequencing raw data (DAT) files are deleted from the sequencer after run completion.

^[2] A configurable disk space cleanup process is executed nightly to free up space for future runs and help ensure sufficient hard disk space is maintained. If the system memory available for storing run analysis files reaches the threshold that is configured, the system auto-deletes 1.wells files for the oldest completed runs. A minimum of 1TB of space is required to start new runs. If the system approaches the 1TB threshold, an alert notifies you when you sign in. New run cannot be started if the 1TB threshold is reached. If desired, you can back up 1.wells files to an external drive.



Create, view, and sign reports

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Reports overview

Genexus™ Software provides two types of reports.

- “Run reports” on page 223
A PDF report of the information that is shown in the **Run Report** tab. The run report includes assay metrics and the record of reagents that were used in a run.
- “Variant reports” on page 225
A PDF report of the results for a sample in a sequencing run. You can use a system-installed report template to generate a variant report, or you can customize the layout and contents in a user-defined variant report. A variant report can be electronically signed off by more than one user, including users who have different roles. One user can be designated to lock reports after reports are signed off.

To save or distribute a variant report or a run report, you can download the report.

Administrators and managers can create user-defined report templates for their organization in Genexus™ Software. For more information, see “Report templates” on page 316.

Run reports

The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “Sequencing metrics” on page 205. If you entered extraction kit barcodes for samples or control samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

View and download a run report

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can view and download a run report in Genexus™ Software. The run report includes assay metrics and the record of reagents that were used in a sequencing run.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of a completed run.
3. Click the **Run Report** tab.
The run report shows assay metrics and the record of reagents that were used in a run. For more information, see “Sequencing metrics” on page 205.
4. To download the run report, click ⋮ **(More Options) ▶ Download Files**, click **Reports**, then click **Download**.

The selected files are downloaded to your computer.

View a run report for sample results

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can view a run report for sample results in Genexus™ Software. The run report for sample results include assay metrics and the record of reagents that were used for a specific sample in a sequencing run.

A run report can be for a single sample or multiple samples. A run report might not be available for every sample that was included in a sequencing run.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name for a completed run.
3. Click the **Reports** tab.
4. In the **Run Report** pane, click ⋮ **(More) ▶ View Report**.

The run report opens in a new browser tab, where you can download the report.

Download a run report for sample results

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can download a run report for a sample result in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name for a completed run.
3. Click the **Reports** tab.
4. In the **Run Report** pane, click **(More) ▶ Download Report**.

The run report is downloaded to your computer.

Variant reports

Role—System administrator, organization administrator and manager, operator, reporter.

The variant report contains the results for each sample in a sequencing run. You can use a system-installed report template to generate a variant report, or administrators and managers can create variant report templates that are customized for your needs. For more information see “Report templates” on page 316.

Variant reports that are automatically generated are based on one or more default report templates. When a report template is set as the default report template for an assay, a report is generated automatically after a sequencing run that completes successfully.

Tip: To generate a variant report automatically that uses a default variant report template that is associated with an assay or with an application category, enable the **Generate Report** option in the **Setup** step when you plan a run. For more information, see Chapter 4, “Plan and manage runs”.

You can also generate a variant report manually when you view sample results after a sequencing run is complete. For more information, see “Generate a variant report” on page 229.

When a variant report has been generated for a sample result, it is available for download in PDF file format.

Locked variant reports can be electronically or manually signed by users. Electronic signatures are shown in the **Electronically Signed By** section of the report, if the section is included in the report template.

For information about the sections that are included in a variant report, see “Understand the sections of a variant report” on page 227.

View a variant report

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can view a variant report in Genexus™ Software. A variant report includes details of sample results. The results that are shown in the report depend on the filter chain and gene list that is applied to the sample results. The report shows the variants that are selected in the **Sample Results** screen.

Before a variant report is available to view, it must be generated automatically during a sequencing run, or manually, after a sequencing run

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name for a completed run.
3. (Optional) You can apply or change a filter chain or gene list that is applied to the sample results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.
4. Click the **Reports** tab.

A pane for each variant report that has been generated is shown. If no report is shown, see “Generate a variant report” on page 229.

The pane for a variant report includes the following information.

- **Draft** label if the report is not locked.
- **Signed Off** label if the report is locked.
- Report name.
- Report template name.
- Date that the report was generated.
- The name of the user who generated the report.
- If a variant report has been electronically signed, the signature information is listed in the pane for the variant report. If multiple users have signed the report, place the pointer over the electronic signature information to view more details. If the electronic signature that is used is associated with a lock-level signature type, the report is locked.
- Language used for the report.
- If a variant report is locked,  **(Lock)** is shown in the upper right corner of the pane.

5. To view the variant report, click  **(More) ▶ View Report**.
6. To download the variant report, click  **(More) ▶ Download Report**.

The variant report is downloaded to your computer.

Understand the sections of a variant report

Users can view variant reports in Genexus™ Software. A variant report can contain the following sections and information.

A variant report may or may not have information about variants. For example, results from immune repertoire assays, PGT-A assays, or microbiome assays do not include information about variants.

The sections shown in a variant report depend on the template design and on the application category and assay that is used in a run. The sections that appear in a variant report are listed and described here in alphabetical order.

Section	Description
Header	Header Information is repeated on the top of each page of the variant report. The header includes the title of the report, the organization address, and an optional logo.
Footer	Footer information is repeated on the bottom of each page of the variant report. The footer includes the page numbering, regulatory information, and version of the Genexus™ Software.
Complex Biomarkers	This section provides key metrics that are available for only some assays. The report may include the following biomarkers: Tumor Mutational Burden, Microsatellite Instability, Genomic Instability, Homologous Recombination Repair, Aneuploidy Score and DNA Mismatch Repair.
Custom Text	This section allows you to add custom text to sections in the variant report during report generation.
Electronic Signatures	This section allows you to add one or more electronic signatures to the variant report. You can include the name of the user who signs the report, the signature type, the date, and comments.
Image	This section allows you to add an image to sections in the variant report during report generation. You can add an image to the variant report from your computer or from an analysis Images can be in PNG, GIF, JPG, TIFF, and JPEG formats. Multiple image sections may be present.
Key Metrics	This section includes amplicon information for the results. The variant report may include the following key metrics: Targeted Coverage, Amplicon Coverage, and Calculated Cellularity.
Kit Barcodes	This section includes details about each kit barcode that is used in the run.
Manual Signatures	This section allows physical signatures on the printed report.

(continued)

Section	Description
QC Metrics	This section includes quality control information. The following information may be included in the variant report PDF output. • Run QC—A summary of the run quality control metrics • NTC QC – DNA—A summary of the DNA no-template control quality control metrics • NTC QC – RNA—A summary of the RNA no-template control quality control metrics • Purification QC—A summary of QC metrics for the purification run. • Sample QC – RNA—A summary of RNA sample quality control metrics • Sample QC – DNA—A summary of DNA sample quality control metrics • Templating Control QC – CF-1—A summary of the CF-1 quality control metrics
Reagents	This section includes details about run consumable kits, including part numbers, expiration dates, and lot numbers.
Sequencing Run Details	This section includes details about the sequencing run. The following information may be included on the variant report PDF output. • Assay Name—The assay used in the run • Assay Revision • Completed On—The date and time when the run was completed • Instrument—The name of the instrument • Filter Chain—The filter chain applied to results • Panel—The panel used in the run • Started On—The date and time that the run was started • Operator—The user who sent the run to the instrument • Serial Number—The serial number of the instrument • Gene List—The gene list applied to the results • Latest Annotations—The name and version of the latest annotation set that is applied to the sample result, if applicable.
Sample Details	This section includes sample information that is entered into the software.
Variant details	This section provides details about any variants that are detected or are not detected in the run. Subsections may be included for SNVs/INDELs, Fusions, and CNVs.
Variant Notes	This section allows you to add notes to any variants that were detected in the run. The notes cannot be removed from a report. The variant name and notes are shown in columns. Only the latest note for each variant is shown.

(continued)

Section	Description
Variants Summary	This section summarizes information about any variants that are detected or not detected in the run. The following details may be included on the variant report PDF output. • SNVs/Indels—The number of variants for each variant type. • Fusions—The number of intergenic variants for each variant type. • CNVs—The number of variants for each variant type, including Focal CNVs and ARM level CNVs. • RNA Variants—The number of exon skip and exon deletion variants.

Generate a variant report

Role—System administrator, organization administrator and manager, operator, reporter.

Any user with permissions to electronically sign off variant reports can generate a variant report after a sequencing run is complete. You can generate multiple variant reports for the same sample result, if each report is named uniquely, that uses a different report template or is generated in a different language. Multilanguage support for report generation is provided.

A  (**Lock**) in the variant report indicates that the electronic signature option for the report is locked. After a variant report is locked, the report cannot be electronically signed by any other user.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Results / Sample Results** screen, in the **Sample Name** column, click the sample for a completed run.
3. (Optional) You can apply a filter chain or gene list to the sample results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.
4. Select the **Reports** tab, then click **+ Generate Variant Report**.
5. In the **Generate Report** dialog box, set the following options.
 - a. Change the name of the report to be generated, if needed.
 - b. Select the report template and language of the report
 Report templates that are associated with only the application category, or both the application category and assay, that was used in the sequencing run are available in the **Report Template** list. For more information, see “Create a report template” on page 316.
 If you select the same report template that was used to generate a variant report, and have not locked that report, the new selections you make override the previous variant report.
 - c. Enter a title in the **Custom Text** section and a description, if applicable.
 Text is editable if the option **Editable on Report Generation** is selected when the report template is created.

- d. If the selected report template includes an **Image** section with the option to include images from your computer after a sequencing run is completed, find and upload the image to the report. Then, enter a title for the uploaded image, and if needed enter a description and footnote for the image.
6. Click **Generate**.
A draft version of the report is added to the **Reports** tab.
7. To view the variant report, click **(More) ▶ View Report**.
8. (Optional) To download the variant report, click **(More) ▶ Download Report**

Sign off a variant report

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can electronically sign a variant report in Genexus™ Software if they have signing permissions. A sample result can have multiple variant reports only if each report uses a different report template, and a different language. However, only one of the reports can include electronic signatures.

IMPORTANT! If you regenerate an existing report before signoff and use the same template and language, the existing report is overwritten.

Multiple signatures can be added to the variant report that is used for sign-offs. Account permissions for electronic signatures are associated with a signature type. A lock-level signature type locks the report. If your signature permissions are associated with a lock-level signature type and you sign the report, you are the last person to sign the report before it is locked.

A filter chain or gene list that is applied to the sample results must be locked before you can electronically sign a variant report. After users begin electronic sign-offs, changes cannot be made to the filter chain, gene list, or classification, and plugins cannot be run for the sample results.

The signature information appears in the variant report PDF file or a user-created report, if electronic signatures are included in the template that is used to generate the report.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

You must have electronic permissions enabled to sign reports. Reports that can be electronically signed are labeled as **Draft** and are not locked.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click the sample result of interest in the **Sample Name** column.
3. Click the **Reports** tab, then in a variant report pane that is labeled as **Draft**, click **Sign Off**.
The **Sign Off Report** dialog box opens and shows the name of the report, the template used for the report, and the following information about the user who signs the report.
 - **Electronic Signature**
 - **User Name**
 - **Language**

4. In **Password**, enter the password.
5. In **Sign Off Comments**, enter a comment.
6. In **Footer Field**, enter any text that you want to appear in the footer of the PDF report pages.
If you entered footer information in the **Footer Field** when you created a report template, or if text was entered during a previous sign off, the same footer information is shown in the **Sign Off Report** dialog box. You can edit the text in the **Footer Field** to change the information that appears in the footer of the PDF report.
7. If desired, select an image to include with the electronic signature, and then enter a description and footnote for the image.
Only one image can be added when a report is signed off. If an image was previously added, the option to add an image is not available.
8. In the **Confirm Sign Off** dialog box, click **Yes** to confirm the electronic signature.
The pane for the signed variant report is updated to include the signature information. If the electronic signature is locked, a  (**Lock**) is shown in the variant report pane.

The report is signed. The **Electronic Signatures** section of the report pane is updated to include your username and the signature type for your account. Place the pointer over the signature type to show the list of signature types that are completed for the report. If multiple electronic signatures are added to the report, an electronic signature type for each signer is shown in the **Electronic Signatures** column of the sample results table.

If a signature type used is designated to lock the report, the variant report is signed and locked, and no other variant reports for the sample result can be signed or generated. If you must change the report, reanalyze the sample and generate a report for the reanalyzed sample.

You can send notifications for report sign-offs to other users. For more information, see “Send a notification to another user” on page 231. For information to download the signed report, see “Download a variant report” on page 232.

Send a notification to another user

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can send a notification to another user or users in Genexus™ Software. For example, you can notify another user when you have signed a variant report. When the recipient signs in to the software, unread notifications,  (Notifications), are shown in the menu bar. For more information, see “View notifications” on page 38.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample for a completed run.
3. In the upper right corner of the screen, click **⋮ (More Options) ▶ Send Notification**.
4. In the **Send Notification** dialog box, in **Send Notification to**, select one or more users of interest.

5. In **Comments** enter a message.
6. Click **Send**.

Download a variant report

Role—System administrator, organization administrator and manager, operator, reporter.

Any user can download a variant report in Genexus™ Software.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name for a completed run.
3. Click the **Reports** tab. then, in the variant report pane, click ⋮ **(More)** ▶ **Download Report**.
You can also download the variant report in the **Results** screen as part of the results files for a specific sample. In this case, place the pointer over the row of a completed run for the sample of interest, click ⋮ **(More Options)** ▶ **Download Files**, then in the **Download Files** dialog box, select the files that you want to download.

Delete a variant report

Role—System administrator, organization administrator and manager, operator, reporter.

Any user who has permissions to electronically sign reports can delete a variant report that is not locked for electronic signatures.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name for a completed run.
3. Click the **Reports** tab.
4. In the variant report pane, click ⋮ **(More)** ▶ **Delete**.
5. Click **Yes** to confirm.
The variant report pane is removed from the **Reports** tab.



Create and manage assays

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Role—System administrator, organization administrator and manager.

Assays in Genexus™ Software

Assays contain the settings and parameters for library preparation, templating, controlling the sequencing run, analyzing, and reporting the results. Assays also define the panels, kits, and chips that are used in a run, and specify the reference files and threshold values for quality control and variant detection. The software files that contain the assay settings and parameters are packaged in a ZIP file called an assay definition file (ADF).

An assay is a reusable experimental design that contains predefined settings appropriate for use with common types of research applications. An assay can be used to plan many runs and plays an important role in enabling rapid throughput across the sequencing instrument. Assays help reduce the chance of errors and saves time since information is stored, and then applied to runs, instead of entered manually for each run.

You can create new assays from system-installed assay templates, and you can copy a locked assay to create an assay, or copy and edit user-defined assays.

Before you can create a user-defined assay, you must add a panel file, and hotspot and copy number baseline files (if needed for the assay), to the software. The customization of parameters in user-defined assays is for advanced users. For help, contact Technical Support at thermofisher.com/support.

Assay creation overview

Role—System administrator, organization administrator and manager.

Administrators and managers can create assays in Genexus™ Software if system-installed assays do not meet needs.

View the assay parameters and other components of the assays in Genexus™ Software. For more information, see “View assay details” on page 258 and “Assay details” on page 259.

Create a user-defined assay in one of these ways.

- Use a template to create a user-defined assay that contains preconfigured settings. The template includes settings for various genetic research analysis applications. For more information, see “Create an assay” on page 234.
- Copy an existing system-installed assay or a locked user-defined assay, then edit the settings of the assay to create a new assay. For more information, see “Copy an assay” on page 237.

After you create an assay, you can optimize the assay configuration, if needed. You can plan a run with a draft assay, view the results, then change the configuration and plan new runs before you lock the draft assay. Lock an assay to prevent copy or edits of the assay.

Create an assay

Role—System administrator, organization administrator and manager.

Administrators and managers can create a new assay from a system-installed template in Genexus™ Software. Each template allows you to select kits, chips, primer panels, and adjustable quality control (QC) thresholds and other parameters that are appropriate for the assay type. The assay is then available to select when you plan a run.

IMPORTANT! Use the default settings for system-installed assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

1. In the menu bar, click **Assays ▶ Create Assay**.
2. In the **Create Assay** screen, select the assay type to open the template that provides the appropriate kits, chips, primer panels, adjustable QC thresholds, plugins, and parameters.
The type of assay templates that are shown are based on the organization and the application category selections.

Assay template type	Description
Generic Sequencing	Use this template to perform only sequencing. The assay performs alignment if a panel is selected in the Panel step. Variants are not included in the results for runs that use this assay.
Generic Sequencing + Variant Calling	Use this template to create an assay that detects germline variants. Annotations are not included in the results. Variant annotations are not included in the results for runs that use this assay.
DNA Germline	Use this template to create an assay that detects and annotates germline variants in DNA samples. For use only with Ion AmpliSeq™ library chemistry. This template is designed for use with a human genome reference.
DNA Somatic	Use this template to create an assay that detects and annotates somatic variants in DNA samples. This template is designed for use with a human genome reference.
DNA and Fusions	Use this template to create an assay that detects and annotates somatic DNA variants and RNA fusion targets in a sample. This template creates a combination of assays that include a DNA-only , a Fusion-only , and a DNA and Fusions research application. Then, when you plan a run, you can select one or more research applications to use in the run. This template is designed for use with a human genome reference.
Fusions	Use this template to create an assay that detects and analyzes variants in fusion samples. This template is designed for use with a human genome reference.
Liquid Biopsy DNA	Use this template to create an assay that detects and annotates somatic variants at low LOD from targeted DNA libraries from cell-free liquid biopsy samples. This template is designed for use with a human genome reference and for use only with Ion AmpliSeq™ HD library chemistry.
TNA	Use this template to create an assay that detects and annotates DNA and Fusion variants in total nucleic acid (TNA) samples. This template is designed for use with a human genome reference and for use only with Ion AmpliSeq™ HD library chemistry.

3. In the **Panel** step, select a panel and other options for the assay.
For descriptions of the panel information appropriate for the assay, see “Panel step assay options” on page 240.
4. Click **Next**.
5. In the **Reagent** step, select the kits and chips that you are using. Ensure that the reagents are appropriate for the primer panel that you are using.
For descriptions of the reagent information that is available for the assay, see “Reagent step assay options” on page 245.
6. Click **Next**.

7. In the **Parameters** step, accept the recommended default values, or adjust the values within the allowed ranges, then click **Next**.

You can set the parameters by uploading an advanced parameters configuration file that overrides the default values in the template.

- a. At the bottom of the **Parameters** step, click **Download** to download a ZIP file of multiple JSON files or a single JSON file depending on which options you select.
- b. Edit the file parameters, then save the file to the computer.
- c. At the bottom of the **Parameters** step, click **Upload**.
- d. In the dialog box, click **Select files**, navigate to the file on the computer, then click **Open**. Click **Upload** to upload the updated JSON file and override the default values in the parameters file and in the software.

For more information about parameters, see “Parameters step assay options” on page 246.

8. In the **QC** step, enter QC metric values, then click **Next**.

For descriptions of the QC information for the assay, see “QC step assay options” on page 248.

9. In the **Plugins** step, select one or more plugins to run with this assay, then click **Next**.

You can configure any plugin that includes configuration options, such as the **coverageAnalysis** plugin, or a user-defined plugin that allows configuration.

For more information, see Chapter 13, “Plugins for data analysis”.

10. In the **Save** step, enter an assay name, an assay full name, and an optional description, then click **Save**.

The types of **Supported Run Types** listed depends on the selections in the **Reagent** step. For example, if a purification kit is selected, **Sample to Result** run type is shown. If library kits are selected, then **Nucleic Acid to Result** run type is shown.

Library to Result run type is shown for every assay type. **Bam to Result** run type is supported for the assays created with **Analysis Pipeline Version 5.16** or later. The analysis pipeline version used to create the assay is shown in the assay details pane.

The new assay is listed in the **Manage Assays** screen and is ready for use in the software. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay. Lock the draft assay to copy the assay or to prevent the assay from being edited. You can also download parameters from assays in **Draft** or **Locked** statuses. Lock the draft assay to copy the assay or to prevent the assay from being edited.

Copy an assay

Role—System administrator, organization administrator and manager.

Administrators and managers can create a new assay by copying an existing system-installed assay or other user-defined assay, then modifying it. Only locked assays can be copied.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of the assay that you want to copy, then click **Copy**.

The **Copy Assay** screen opens to the **Panel** step. The assay settings can be modified for the new assay.

3. Proceed through the workflow steps to modify certain assay settings in the **Parameters** or **QC** steps if needed. To modify some values in the Panel or Reagent step, you need to first save the copy of the assay, then go back to those steps. For more information, see “Assay options” on page 240.

If you copy an assay from a previous software version, ensure that the latest CustomerSupportArchive plugin version is selected. For more information about the latest version, see the Genexus™ Software release notes.

4. In the **Save** step, enter a new assay name, an assay full name, and an optional description, then click **Save**.

The new assay is listed in the **Manage Assays** screen and is ready for use in the software. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay. Lock the draft assay to copy the assay or to prevent the assay from being edited. You can also download parameters from assays in **Draft** or **Locked** statuses. Lock the draft assay to copy the assay or to prevent the assay from being edited.

Create an assay for an integrated purification and sequencing run

Role—System administrator, organization administrator and manager.

Administrators and managers can create a new assay or copy and edit an existing assay that supports a **Sample to Result** run type.

This is an example for how to create an assay for an integrated run that uses a **Sample to Result** run type and allows you plan runs for nucleic acid purification on a Genexus™ Purification System and sequencing on a Genexus™ Integrated Sequencer. **Sample to Result** run plans support integrated sequential nucleic acid purification and sequencing. The concentration of purified nucleic acid for each sample is calculated after purification, before sequencing. You can choose to review samples after purification or proceed directly to sequencing.

1. In the menu bar, click **Assays ▶ Create Assay**.
2. In the **Create Assay** screen, click the assay type for the template that provides the appropriate kits, chips, primer panels, and adjustable QC thresholds and read mapping parameters.

For more information about selections, see “Create an assay” on page 234.

3. In the **Panel** step, select a panel and other options for the assay. .
For descriptions of the panel information appropriate for the assay, see “Panel step assay options” on page 240
4. Click **Next**.
5. In the **Reagent** step, in **Purification Kit**, select the appropriate kit.
If you select **Other**, the assay cannot be used in an integrated purification and sequencing run that uses a **Sample to Result** run type.
6. Select the kits and chips that you are using. Ensure that the reagents are appropriate for the primer panel that you are using, then click **Next**.
For descriptions of the reagent information appropriate for the assay, see “Reagent step assay options” on page 245.
7. In the **Parameters** step, accept the recommended default values, or adjust the values within the allowed ranges, then click **Next**.
You can set the parameter values with an advanced configuration file. For more information, see “Edit assay parameters in a file” on page 253.

Purification parameters

Option	Description
Sample Type	The types of samples that are compatible with the selected purification kit are listed.
Review Required	Use this option to review the concentration of all purified samples prior to sequencing to determine which samples to include in the sequencing run. • Select True to review the concentration of all samples after purification and select which samples to sequence. • Select False to enable automated sequencing of all samples that are within the specified concentration after purification. You can override this assay setting when you plan a purification run. For more information, see “Plan a Sample to Result run” on page 89.
QC Concentration Minimum	QC concentration minimum thresholds in ng/μl for DNA or RNA.
QC Concentration Maximum	QC concentration maximum thresholds in ng/μl for DNA or RNA.

For more information about parameters, see “Parameters step assay options” on page 246.

8. In the **QC** step, enter QC metric values, then click **Next**.
For descriptions of the QC information appropriate for the assay, see “QC step assay options” on page 248.
9. In the **Plugins** step, select one or more plugins to run with this assay, then click **Next**.
Click the **Configure** link to change the settings for a plugin that includes configuration options, such as the **coverageAnalysis** plugin, or a user-defined plugin that allows configuration.

For more information, see Chapter 13, “Plugins for data analysis”.

10. In the **Save** step, enter an assay name, an assay full name, and an optional description, then click **Save**.

The new assay is listed in the **Manage Assays** screen. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay and the assay is available to lock, edit, or delete, and audit. You can also download parameters from assays in **Draft** or **Locked** statuses.

Enable tumor mutational burden calculation in an assay

Tumor mutational burden (TMB) is a calculation of somatic mutations per megabase (Mb). Tumor mutational burden calculation is available in Genexus™ Software.

You can enable the calculation in a custom Ion AmpliSeq™ or Ion AmpliSeq™ HD assay. When enabled, tumor mutational burden and other data values and graphics are included in sample results from that assay. You can enable the tumor mutational burden calculation when you create a new assay, copy and edit any system-installed assay, or edit a user-defined assay that is not locked.

Use this procedure to create an assay, and then set the **Tumor Mutational Burden Filter Chain** parameter to enable the tumor mutational burden calculation.

1. In the menu bar, click **Assays ▶ Create Assay**.
2. In the **Create Assay** screen, select one of the following assay types to open the template that provides the appropriate kits, chips, primer panels, adjustable QC thresholds, plugins, and parameters.
 - **DNA Germline**
 - **DNA Somatic**
 - **DNA and Fusions**
 - **Liquid Biopsy DNA**
 - **TNA**
3. Click **Next**
4. In the **Parameters** step, under **Annotation**, find the **Tumor Mutational Burden Filter Chain** parameter, then select an option that is based on the reference genome for the panel.
 - **TMB (Non-germline Mutations (7.0))**
 - **TMB GRCh38 (Non-germline Mutations (7.0))**

The analysis pipeline version that is used to calculate tumor mutational burden is included in the name for the **Tumor Mutational Burden Filter Chain** option.

5. Adjust other values for tumor mutational burden calculation if needed.

Note: You can upload an advanced parameters configuration file that overrides the default values in the assay template to set parameters for **TMB Standardization**.

For more information about parameters, see “Parameters step assay options” on page 246.

6. Click **Next**.
7. Proceed through the remaining workflow steps and change other assay settings if needed. For more information, see “Assay options” on page 240.
8. In the **Save** step, enter an assay name, an assay full name, and an optional description, then click **Save**.

The new assay is listed in the **Manage Assays** screen and is ready for use in the software. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay. Lock the draft assay to copy the assay or to prevent the assay from being edited. You can also download parameters from assays in **Draft** or **Locked** statuses. Lock the draft assay to copy the assay or to prevent the assay from being edited.

Assay options

Panel step assay options

Role—System administrator, organization administrator and manager.

Administrators and managers can use the **Panel** step to select the appropriate reference files and other settings for use with an assay that is created or copied and edited in Genexus™ Software.

IMPORTANT! Use the default settings for system-installed assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

The options that are available in the **Panel** step depend on the type of assay template that is selected.

Option	Description
Panel	The panel to use in the assay. The remaining options are populated in the screen based on the selected panel. The panel is a set of files that defines both the DNA target regions, and a fusion panel, or only DNA target regions, or a fusion panel.
Hotspots	The hotspots file to use in the assay. The hotspots file is a tab-delimited BED file that indicates the positions where a DNA variant is expected to occur. It also annotates these positions with the reference allele, variant allele, and anchor base. Select None if the run plan for which the assay is used will not include a hotspots file.
Target Regions	The target regions BED file that is appropriate for the sample. Depending on the application, you may need to select a separate DNA and Fusions target regions file. The panel is a tab-delimited BED file that defines the coordinates of the regions of interest that are amplified by the matching primer set. If you use the Generic Sequencing assay template, the option None, if selected, applies no target regions file.
Genome Reference	The genome reference file for the panel that is selected is displayed. When you create or upload a panel, you select the genome reference. For more information, see “Import or add a custom panel” on page 328.

(continued)

Option	Description
Library Chemistry	The type of library chemistry to use in the assay. This option is selected, based on the panel.
Extraction Method Type	The method used to extract the DNA, RNA, or DNA and RNA from the sample, or as a single library TNA from the sample. This option is selected, based on the panel. For the Generic Sequencing assay template, if no panel is selected, you must select an Extraction Method Type.
Minimum Read Count Per Sample DNA	The minimum number of reads that are needed per sample to meet the coverage depth required in a DNA only and a DNA & RNA assay. This value, usually associated with the panel, determines the number of samples that can be run on the lanes that are available on a chip. The system uses this value to calculate how many lanes a run uses and assigns samples according to the calculation. For example, if the lane capacity is 2,000,000 reads and the minimum read count per sample is 1,000,000 reads, then the software allows you to run two samples in each lane.
Minimum Read Count Per Sample RNA	The minimum number of reads per sample that are needed to meet the coverage depth required in an RNA assay. This value, usually associated with the panel, determines the number of samples that can be run on the lanes that are available on a chip. The system uses this value to calculate how many lanes a run uses and assigns samples according to the calculation. For example, if the lane capacity is 2,000,000 reads and the minimum read count per sample is 1,000,000 reads, then the software allows you to run two samples in each lane.
Run CNV	When Run CNV is enabled, the software analyzes CNV results based on the selected CNV baseline file. When you enable this setting, you must also select a Copy Number Baseline for CNV Calling to detect copy number variants (CNVs). If the default setting, Do Not Run CNV Algorithm , is selected, the software does not run the CNV algorithm even if Run CNV is enabled.
Copy Number Baseline for CNV Calling	You must select a baseline file to detect copy number variants (CNVs). The copy number baseline is a set of control samples that are used to create a baseline for the detection of copy number variants (CNVs). You can use system-installed baselines, import or create a baseline, or copy and edit an existing baseline. You can view the settings that are used for the baseline in the Assays/Manage Presets/Copy Number Baselines tab. For more information, see “Copy number baselines” on page 288.
CNV Calling method	Select either Mosaic, Full Ploidy, or both. When mosaicism detection is enabled, the software can make non-integer ploidy calls. CNV events are reported as a decimal ploidy value instead of an integer value. CNV calling with Full Ploidy includes read coverage correction for GC bias. Corrected coverage is compared to a baseline coverage from control samples of regions with known expected normal ploidy. That is, 2 on autosomes and X in females, and 1 on sex chromosomes in males. This option is available only for aneuploidy assays.

(continued)

Option	Description
Adjust Copy Number by Sample Cellularity	The percent cellularity that is the minimum value that the assay requires of a sample. Only samples above the threshold value can be assigned to a run plan with the assay when the choice is enabled. Click Adjust Copy Number by Sample Cellularity to enable the setting in the software. You can adjust the percentage in the dropdown list that appears when the setting is enabled. For example, if you select 50, you cannot add a sample with a %Cellularity of 49 to a run plan that uses the assay. This option is available only for assays that use a DNA and Fusions panel or a DNA panel.
Fusion Reference	The tab-delimited BED file to use in the assay that indicates the positions where an RNA fusion target is expected to occur. It also annotates these positions.
Fusion Panel	The tab-delimited BED file to use in the assay that defines the coordinates of the regions of interest to be amplified with the selected primer set.
Fusion Baseline	The fusion baseline used in the assay.
Primer Pools	The number of primer pools used in the assay.
Annotation Sets	The annotation set to use in the assay.
Apply OncoPrint Variant Annotations	When you select the Apply OncoPrint™ Variant Annotations option in the Panel step to create or edit an assay, analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types is used as a reference to annotate variants that are relevant to cancer. The annotations are shown in results in the OncoPrint™ Gene class and OncoPrint™ Variant class columns. For more information, see “Annotation criteria for the OncoPrint™ Variant Annotator” on page 527 or contact a support representative, or Field Bioinformatics Specialist (FBS). This option is available only for assays that use custom Ion AmpliSeq™, Ion AmpliSeq™ HD and OncoPrint™ panels.

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Option	Description
Oncomine™ Variant Annotations Ruleset	Select an option in the Oncomine™ Variant Annotations Ruleset menu in the Panel step to determine whether a set of annotation rules is used with this assay. The date in the ruleset name is provided as part of the version number to distinguish annotations ruleset versions. Rulesets that are shown match the version of the analysis pipeline that is used in the assay. When you apply an Oncomine™ variant annotations ruleset to an assay, the Oncomine™ Variant Annotator in Genexus™ Software uses analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types as a reference to annotate variants that are relevant to cancer. The annotations are shown in results in the Oncomine™ Gene Class and Oncomine™ Variant Class columns. For more information, see “Annotation criteria for the Oncomine™ Variant Annotator” on page 527 or contact a support representative, or Field Bioinformatics Specialist (FBS). If you select None for this option, an Oncomine™ Variant Annotations Ruleset is not applied to sequencing runs for which the assay is used. This option is available only for assays that use custom Ion AmpliSeq™, Ion AmpliSeq™ HD and Oncomine™ panels designed for use with human genome references. You can use an Oncomine™ Variant Annotations Ruleset that is included in an installed Oncology content package update. For more information, see “Oncology content packages for use in assays” on page 263.
Use Latest Variant Name	When enabled, this setting instructs the assay to use the most recent version of the rules and conventions used in the software to name genomic variants. Standard variant names are derived from properties that are available in the software per variant, such as variant, gene name, and amino acid change. This option is applicable in Genexus™ Software 6.8 and later. You can use an updated set of naming rules and conventions from an Oncology content package update that is installed in the software. For more information, see “Oncology content packages for use in assays” on page 263.
Use Latest Annotations	- When you enable this setting, the latest version of the ClinVar annotation source that is available in the software is used by the assay. - When you disable this setting, the ClinVar annotation source version that is associated with the assay is used regardless of whether newer versions of the annotation source are installed in the software. That is, if a newer version of the ClinVar annotation source is installed in the software and the setting is inactive, that version is not associated with the assay. This option is available only for assays that use the ClinVar annotation source from the software to annotate data. For more information, see “Use the latest ClinVar annotation source to annotate results” on page 263 and “Revert to the ClinVar annotation source included with the annotation set” on page 264.

(continued)

Option	Description
Gene List	You can select a gene list—a collection of genes of interest—to apply to the sample results of the assay that is used for the sequencing run. If you do not select a Gene List, the default selection No Gene List is applied to the assay. The gene list that you select can be in draft or locked status, if the assay is in draft status. Before you can lock an assay, you must lock the gene list that is associated with the assay. A gene list that is compatible with Genexus™ Software 6.6 and later which includes an earlier analysis pipeline, can be associated with an assay, if an assay from the earlier version of Genexus™ Software is copied and edited. You can use an updated gene list from an Oncology content package update that is installed in the software. For more information, see “Oncology content packages with gene lists” on page 348.
Filter Chain	You can select a filter chain—a set of filters that Genexus™ Software uses to narrow the list of variants that appear in a sample result—to apply to the sample results of the assay that is used for the sequencing run. If you do not select a Filter Chain, the default choice No Filter Chain is applied to the assay. The filter chains that are available depend on the genome reference that is selected for the assay. The filter chain that you select can be in draft or locked status, if the assay is in draft status. Before you can lock an assay, you must lock the filter chain that is associated with the assay. A filter chain that is compatible with Genexus™ Software 6.6 and later that includes an earlier analysis pipeline, can be associated with an assay. To apply a filter chain for an earlier version of the assay, copy and edit the assay from the earlier version of Genexus™ Software.

Reagent step assay options

Role—System administrator, organization administrator and manager.

Administrators and managers can use the **Reagent** step when an assay is created or copied and edited in Genexus™ Software.

In the **Reagent** step, you select the kits and chips to use in an assay, and ensure that the kits and chips that you select are appropriate for the primer panel that you are using.

IMPORTANT! Use the default settings for system-installed assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

The options that are available in the **Reagent** step depend on the type of assay.

Purification

Option	Description
Purification Kit	The purification kit that is used for nucleic acid purification. This option is not available for Generic Sequencing + Variant Calling and Liquid Biopsy DNA assay templates.

Library Preparation

Option	Description
Library Kit	The library kit that is used to prepare the library.
Barcode Kit	The barcode kit to use in the assay.
Include Inline controls	You can select DNA or RNA, or both DNA and RNA, depending on the assay template that is used. For example, you might want to ensure that both DNA and RNA are selected if you use the DNA/RNA template. By default, only DNA is enabled and RNA is disabled. RNA is available for selection only if the assay template includes it. TNA is an option for TNA assays.

Templating

Option	Description
Sequencing Chip	The sequencing chip to use in the assay. When you change the Sequencing Chip option, other assay parameters are changed to the default settings that are associated with the chip. Review the settings in the Parameters step after you make this change.
Templating Kit	The templating kit to use in the assay. If you change the Templating Kit option, other assay parameters are changed to the default settings that are associated with the templating kit. For more information, see “Edit assay parameters in a file” on page 253.

Templating *(continued)*

Option	Description
Template Size (bp)	The template size, measured in base pairs, to use in the assay. • 200 • 400 Based on the Template Size (bp) selection, the sequencing flows get updated. When the sequencing flows get updated, the enabling of read sequencing is modified. Read sequencing is disabled based on the following threshold values for flows. – For assays with Ion AmpliSeq™ library chemistry, read sequencing is disabled if the value exceeds 500 flows. – For assays with Ion AmpliSeq™ HD library chemistry, read sequencing is disabled if the value exceeds 600 flows.

Sequencing

Option	Description
Sequencing Kit	The sequencing kit to use in the assay.
Sequencing Flows	The number of flows performed by the sequencer during the sequencing stage of the run.

Parameters step assay options

Role—System administrator, organization administrator and manager.

Administrators and managers can use the **Parameters** step to select the appropriate parameter values for use with an assay that is created or copied and edited in Genexus™ Software.

IMPORTANT! Use the default parameter settings for assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

When you click a parameter field, the software shows the default value and the range that is allowed for the parameter. You can see a description of most parameters when you place the pointer over [?](#) **(Learn More)** next to a parameter.

The options that are available in the **Parameters** step depend on the type of assay and the selections that are made in the **Reagent** step.

- **Purification**
- **Library Prep & Templating**
- **Primary Analysis**
- **Annotation**
- **MSI** (Microsatellite Instability)

Advanced Command Line Arguments in the **Primary Analysis** section can be adjusted when you click **Enable customization**.

IMPORTANT! Important: This option is for use only with assay kits, panels and ADFs that support MSI analysis. Do not enable this option for use with any other assays.

- **CNV Finding**

IMPORTANT! If a JSON parameter file is uploaded under **Parameters** ▶ **CNV**

Finding ▶ **Parameter file**, although the following parameters are specified in the JSON file, the parameters are not considered and are overridden by command line arguments.

• <i>cnv.brca.gene.threshold</i>	• <i>cnv.brca.gene.sid.loss.threshold</i>
• <i>cnv.brca.gene.min.factor</i>	• <i>cnv.brca.gene.sid.gain.factor</i>
• <i>cnv.brca.gene.max.mapd</i>	• <i>cnv.brca.gene.sid.loss.factor</i>
• <i>cnv.brca.gene.max.cv</i>	• <i>cnv.brca.gene.sid.max.cv</i>
• <i>cnv.brca.gene.sid.gain.threshold</i>	
-

- **Fusions**
- **Read Mapping**
- **Variant Finding**

In the **Parameters** step, you can accept the default settings for the assay parameters, or adjust the settings, if needed.

Reduce the impact of deamination in low-quality FFPEs

Samples of low quality that exhibit deamination can be analyzed if you increase the minimum allele frequency parameters in an assay. For example, a minimum allele frequency value of 10% for the tumor mutational burden (TMB) calculation can reduce the impact of deamination on the reported TMB value. It is important to consider the tumor content of a given sample when you increase the minimum allele frequency of the TMB calculation.

- Samples with low tumor content can have many true somatic mutations that are excluded from the tumor mutational burden calculation when a higher minimum allele frequency filter is included in the assay.
- The tumor mutational burden values cannot be reliable if you adjust the minimum allele frequency parameters for samples with a high estimated SNP proportion consistent with deamination, (primarily an FFPE number) or a high deamination score, (for example >100).
- The mean depth and uniformity of the sample as determined by the coverageAnalysis plugin can also indicate sample quality. For more information, see “coverageAnalysis plugin in Genexus™ Software” on page 405.

IMPORTANT! Use the default parameter settings unless you are an advanced user.

QC step assay options

Role—System administrator, organization administrator and manager.

Administrators and managers can use the **QC** step to select appropriate Quality Control (QC) metric values for use with an assay that is created or copied and edited in Genexus™ Software.

The options that are available in the **QC** step depend on the type of assay.

CF-1 Control

Option	Description
Min Average Reads Per Lane	The number of average reads per chip lane. You can specify the minimum threshold value that is required for the CF-1 (templating Control Fragment-1) to pass the QC metric.
Min Base Call Accuracy	The total number of errors for all positions in CF-1/ total number of CF-1 base reads. You can specify the minimum threshold value that is required for CF-1 to pass the QC metric.
Min Mean AQ20 Read Length (bp)	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for reads of a library. You can specify the minimum threshold value that is required for CF-1 to pass the QC metric.

NTC QC - DNA

Option	Description
Max Average Base Coverage Depth	The average number of reads of all targeted reference bases. You can specify the maximum threshold value that is required for the DNA no-template control to pass the QC metric.
Max Mean Read Length (bp)	The average length, in base pairs, of called reads. You can specify the maximum threshold value that is required for the DNA no-template control to pass the QC metric.

NTC QC - RNA

Option	Description
Max Mapped Fusion Reads	The total number of fusion reads that are mapped to the reference genome in Genexus™ Software. You can specify the maximum threshold value that is required for the RNA no-template control to pass the QC metric. Mapped Reads can be configured in the QC step or in the Parameters step in the Fusions section as Minimum Total Valid mapped reads. The values configured in the Parameters step overwrite those configured in the QC step.
Max Mean Read Length (bp)	The average length, in base pairs, of called reads. You can specify the maximum threshold value that is required for the RNA no-template control to pass the QC metric. Mapped Reads and Mean Read Length can be configured in the QC step or in the Parameters step in the Fusions section as Minimum Total Valid mapped reads and Minimum mean read length for valid sample QC. The values configured in the Parameters step overwrite those configured in the QC step.

Purification QC – DNA

Option	Description
Min Sample Concentration DNA (ng/μl)	The minimum threshold value for the sample concentration of purified DNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.
Max Sample Concentration DNA (ng/μl)	The maximum threshold value for the sample concentration of purified DNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.

Purification QC – RNA

Option	Description
Min Sample Concentration RNA (ng/μl)	The minimum threshold value for the sample concentration of purified RNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.
Max Sample Concentration RNA (ng/μl)	The maximum threshold value for the sample concentration of purified RNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.

Purification QC – TNA

Option	Description
Min Sample Concentration TNA (ng/μl)	The minimum threshold value for the sample concentration of purified TNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.
Max Sample Concentration TNA (ng/μl)	The maximum threshold value for the sample concentration of purified TNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for an integrated purification and sequencing run” on page 237.

Run QC

Option	Description
Min Final Reads	The number of library reads required in a run. Only reads passing quality and other filters are included. You can specify the minimum required number of reads for a run to pass QC.
Min Key Signal	The average signal after software processing for all library ISPs that identically match the library key (TCAG). This value is a measure of the efficiency of template amplification. You can specify the minimum threshold value that is required for a run to pass QC for the key signal metric.
Min Percent Loading	The percentage of addressable wells on a chip lane that are loaded with an ISP. The percentage is derived from the number of wells with ISPs divided by the number of the total addressable wells in a run. You can specify the minimum threshold value that is required for a run to pass QC for the percent loading metric.
Min Raw Read Accuracy	The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as $100 \times (1 - (\text{sum(per base error)} / \text{sum(per base depth)}))$. You can specify the minimum threshold value that is required for a run to pass QC for the raw read accuracy metric.

Sample QC - DNA

Options	Description
MAPD	You can specify the maximum threshold value that is required for a DNA sample to pass the QC metric. This option is available only when a Copy Number Baseline for CNV Calling is selected in the panel step. For more information, see “MAPD” on page 597.
Min Deamination score	Deamination is reported as the estimated SNP proportion consistent with deamination (low allele frequency C:G>T:A SNVs). The deamination score can be used to determine the quality of the FFPE sample. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Mapped Reads	The number of reads aligned to the reference genome. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Mean AQ20 Read Length (bp)	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for reads of a library. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Mean Read Length (bp)	The average length, in base pairs, of called reads. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Number Of Bases Used In Calculating TMB	The number of exonic bases or all genomic bases that are covered by the panel. Only bases with sufficient base coverage are used in the calculation, as defined in the workflow parameters. By default, the TMB calculation uses total exonic bases with ≥ 60 bp coverage. In the TMB parameter settings, you can select between exonic bases or all genomic bases. You can also modify the sufficient coverage value. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Mean Red Cov	The mean molecular coverage of targeted CNV gene. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Median Mol Cov	The median molecular coverage of non-CNV reference loci. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.

Sample QC - DNA (continued)

Options	Description
Min Number of variant calls	The number of somatic variants that are identified in the sample. This value is reported in the statistic.txt file as Total Somatic Filtered Variants Count (numerator for TMB calculation) and Variant Count. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.
Min Uniformity of Base Coverage	The percentage of reads showing a depth of coverage $\geq 20\%$ of the mean base coverage. You can specify the minimum threshold value that is required for a DNA sample to pass the QC metric.

Sample QC - RNA

Option	Description
Min Mapped Fusion Reads	The number of reads aligned to the reference genome. You can specify the minimum threshold value that is required for an RNA sample to pass the QC metric. The minimum allowed value for mapped reads. Min Mapped Fusion Reads and Mean Read Length can be configured in the QC step or in the Parameters step in the Fusions section as Minimum Total Valid mapped reads and Minimum mean read length for valid sample QC. The values configured in the Parameters step overwrite those configured in the QC step. The best practice to achieve consistent results is to use the same values in both the QC step and the Parameters step.
Min Mean AQ20 Read Length (bp)	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for reads of a library. You can specify the minimum threshold value that is required for an RNA sample to pass the QC metric.
Min Mean Read Length (bp)	The average length, in base pairs, of called reads. You can specify the minimum threshold value that is required for an RNA sample to pass the QC metric.
Min RNA Expression Ctrls Detected	The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. Fusion panels include primer pairs that cover seven control housekeeping genes. For example, to pass QC, Oncomine™ Precision Assay GX requires that 2 out of 7 control genes for cfTNA samples be detected, and 5 out of 7 control genes for FFPE RNA samples be detected. You can specify the minimum threshold value that is required for an RNA sample to pass the QC metric. This option is available only for cfTNA panels.

Resequencing

Resequencing is repeat sequencing of the same DNA template a second time when the chip that was used in the first run remains on the Genexus™ Integrated Sequencer. The benefits of resequencing depend on the assay. Resequencing can improve sequencing metrics, increase the number of reads, improve sensitivity, or increase the number of samples that can be sequenced on a chip. Runs that include the resequencing option have longer run times.

When you copy and edit or create an assay in Genexus™ Software, that user-defined assay can include an option to sequence and resequence samples in the same run. The option to resequence samples is available only in assays that specify a 200-bp template size.

For more information about resequencing samples, see the Genexus™ Integrated Sequencer user guide, or the user guide for the assay or panel kit.

Resequencing settings are available in the **Primary Analysis** section of the **Parameters** step when you create an assay. Template size is specified in the **Templating** section of the **Reagent** step.

The way that resequencing is implemented depends on the library chemistry of an assay. For system-installed assays with Ion AmpliSeq™ library chemistry, resequencing is not needed to improve performance. Therefore, resequencing is disabled for system-installed Ion AmpliSeq™ assays. Resequencing is enabled for system-installed Ion AmpliSeq™ HD assays.

By default, resequencing is enabled for assays that use Ion AmpliSeq™ HD chemistry and disabled in assays that use Ion AmpliSeq™ chemistry. Assay parameters can require optimization if resequencing is enabled for Ion AmpliSeq™ assays.

Flow order

The flow order is the order in which a chip is exposed to each particular dNTP. The default **Ion samba** flow order consists of a 32-base sequence, repeated. This flow order resists phase errors by providing opportunities for out-of-phase molecules to catch up, and is designed to sample all dimer (nucleotide pair) sequences efficiently. **Ion samba** is the default flow order because it improves sequencing accuracy for longer reads by resisting phase errors.

The flow order option is in the **Primary Analysis** section of the **Parameters** step when you create, or copy and edit, an assay.

When you create or copy an assay and enable resequencing, the flow order for resequencing must be different from the flow order for sequencing. We recommend **Ion samba** for the flow order for sequencing and **Ion samba.contradanza** for the flow order for resequencing.

Edit assay parameters in a file

Role—System administrator, organization administrator and manager.

Administrators and managers can edit assay parameters in the **Parameters** step in Genexus™ Software.

Not all parameters appear in the software. You can download a JSON file that contains the parameters that appear in the software and more parameters that do not appear in the software. For more information, see “Download assay parameters” on page 265. If you are an advanced user and understand how the changes can affect results, you can edit the parameters file and then upload

the edited file to create an assay that captures the values of the JSON file. Contact your Field Bioinformatics Specialist (FBS) for assistance.

1. In the menu bar, click **Assays ▶ Create Assays**.
2. Select the type of assay you want to create.
3. In the **Panel** step, select a panel and other options, then click **Next**.
4. In the **Reagent** step, select the kits and chips, and then click **Next**.
5. In the **Parameters** step:
 - a. In the **Filter Parameters by** dropdown list at the bottom of the screen, select the section for the parameters you want to change or select **All** to download a file that contains all parameters.
 - b. Click **Download** to download a JSON file for the parameters.
6. Edit the parameter values in the file and save the file.
7. Click **Upload**.
 - a. Click **Select File**.
 - b. Navigate to the edited file.
 - c. Click **Open**.
 - d. In the **Upload Parameters** dialog box, click **Upload** to upload the file.

The parameter values in the uploaded file are added to the assay.

Manage assays

Administrators and managers can import an assay, export a locked assay, edit, lock, or delete a draft assay, edit the QC parameters of a locked assay, and obsolete a locked assay in Genexus™ Software.

Operators can view assay details, view the audit trail of an assay, and download assay parameters.

Find an assay

You can search, filter, and sort information to narrow the list of assays shown in the **Assays / Manage Assays** screen, then find a specific assay in Genexus™ Software.

For information to search for an assay by a unique identifier, such as the name of the assay, see “Search for assays” on page 255.

For information to filter assays by a common trait, such as the name of the creator of the assay, see “Filter and search in the list of assays” on page 256.

Search for assays

You can search for assays in Genexus™ Software with keywords or text that are included in the name or full name of an assay.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. Click **Assays ▶ Manage Assays**.
 2. In the **Manage Assays** screen, in the **Assay Full Name** column heading or **Assay Name** column heading, click  (**Filter**).
 - a. In the search field, enter a full or partial assay name of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - b. In the filter dialog box, select from the limited list.
 - Select a checkbox next to the name of an assay to select one or more assays.
 - Click **Select All** to select all assays listed in the filter dialog box.
-

Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

- c. In the filter dialog box, click **Filter**.
In the column heading,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of assays is shown in the **Manage Assays** screen.

Click  **Clear Filters** to remove all filters and view the full list of assays.

Filter and search in the list of assays

You can apply filters to narrow search results, or shorten the list of assays when you view the list of assays in Genexus™ Software.

You can also search for assays in Genexus™ Software with keywords or text that are included in an assay name.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for **demo** in analysis names returns a list of all analyses with a name that includes **demo**. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and therefore are not recommended. For example, a search for **demo 1** would return results only for data that includes the string **demo1**.
-

1. Click **Assays ▶ Manage Assays** to open the **Manage Assays** screen.
2. Click the **Filter Assays By** dropdown menu, then apply the filters.
 - Select **IRU Shortlist** to limit the assays shown to the assays that are tagged for use with the IonReporterUploader plugin.
 - Select **Status** to expand the list of options for status, then select a status:
 - Locked & Draft
 - Obsolete
 - Locked
 - Draft
 - Select **All** to return to the complete list of assays.

3. Select from the filter and search options in the column heading of interest.

Option	Description
Filter and search by name of an object.	Click  (Filter) in the column heading of interest, then select one or more of the search options. Enter at least 3 characters to search by the full or partial name for the object. Select one or more matches from the filter dialog box. To select all matches, click Select All. Next, click Filter. The filtered list of assays is shown in the Manage Assays screen.
Filter and search by research application.	Click  (Filter) in the Research Application column, then select one or more of the search options. - Enter at least 3 characters to search by the full or partial research application name. Select one or more matches from the filter dialog box. To select all matches, click Select All. Next, click Filter. - Select from the research applications shown, then click Filter. The filtered list of assays is shown in the Manage Assays screen.
Filter and search by the creator of the assay.	Click  (Filter) in the Created By column, then select one or more of the search options. - Enter at least 3 characters to search by the full or partial assay name. Select one or more matches listed in the filter dialog box. To select all matches, click Select All. Next, click Filter. - Select from the user names that are shown, then click Filter. The filtered list of assays is shown in the Manage Assays screen.
Filter by the exact or approximate date on which assays were created.	Click  (Filter) in the Created On column, then use the options to filter by a date or range of dates to filter the list based on the assay creation date.

4. (Optional) Click **Clear Filters** to return to the entire list of assays.

The list of assays is filtered according to the selections.

Sort the list of assays

You can sort the list of assays to make it easier to find assays in Genexus™ Software.

- Click **Assays** ▶ **Manage Assays** to open the **Manage Assays** screen.
- Click a column heading in the list of assays.
If the column is sortable, the contents of the column is sorted alphabetically, or chronologically.
- Click the column heading again to reverse the order.

View assay details

Role—System administrator, organization administrator and manager, operator.

You can view assay details in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, click the assay name of interest.

The assay details are shown.

If needed, you can click ... **(More Options)** to perform more actions, such as export, copy, edit, lock, delete, obsolete, edit QC, download parameters, or audit the assay. The options that are available depend on whether the assay is system-installed or is a user-defined assay. For user-defined assays, the options that are available also depend on whether or not the assay is locked.

The actions for each assay are also available in the **Manage Assays** screen when you place the pointer over the row of an assay.

Assay details

When you view assay details in Genexus™ Software, you can view information about the components of the assay. The information that is available depends on the type of assay.

You can view the following information in the section that contains the assay name.

- **Research Application**—The research application for the assay, such as **DNA** or **DNA and Fusions**.
- **Created By**—The sign-in name of the person who created the assay.
Assays that are included with the software are labeled as **System Installed**.
- **Analysis Pipeline Version**—The version for a collection of algorithmic modules that analyze data and identify variants including SNPs, INDELs and fusions, and biomarkers, and can include the annotation of variants.
Each assay in the software is associated with an analysis pipeline version. The annotation sets that are available in the software when you create or edit an assay are compatible with the same version of that analysis pipeline as the assay.
- **Created On**—The date on which the assay was created.
- **Assay Name**—The name and version of the assay.

Note: For system-installed assays or assays that you install in the software, the end of the assay name, for example w5.0.0, is the version number of the assay. The assay version number is not the same as the **Revision** that is shown in the assay details.

- **Revision**—Tracks revisions made to unlocked assays. For example, when you edit an assay for the first time, the version changes from **1** to **2**.

Assay detail ^[1]	Description
Panel	The panel section lists the reference files that are used in an assay and other information about options used for the panel. For example, the Minimum Read Count Per Sample, which is the minimum number of reads per sample that are needed to achieve the coverage depth required for the assay, is shown. The minimum read counts per sample is used to calculate the number of samples that are allowed in a chip lane when you plan a run. For more information and a complete list of components in the panel section, see “Panel step assay options” on page 240.
Reagents	The reagents section lists the kits and chips for the assay and other information about options selected for reagents. For more information and a complete list of information shown in the reagents section, see “Reagent step assay options” on page 245.
• NTC QC – RNA • CF-1 Control • Purification QC • NTC QC – DNA • Run QC • Sample QC	The QC sections list the Quality Control (QC) metric values for the assay. For more information and a complete list of metrics in the QC sections, see “QC step assay options” on page 248.

(continued)

Assay detail ^[1]	Description
Plugins	The plugins sections list the plugins that are associated with the assay. For more information about plugins, see Chapter 13, “Plugins for data analysis”.
Description	A short description of the assay, similar to the assay full name.

^[1] The details that are shown depend on the assay.

Transfer an assay to another sequencer

Role—System administrator, organization administrator and manager.

If you want to use a user-defined assay with another Genexus™ Integrated Sequencer, first use Genexus™ Software to export the assay to a folder on the computer on which the software is used. Then, use the software to import the assay to another sequencer.

You cannot export system-installed assays. Only user-defined assays that are locked can be transferred.

IMPORTANT! Before you transfer an assay to another sequencer, you must ensure that all user-defined genome reference files and presets that are associated with the assay, including an annotation set, a filter chain, a gene list, report template, and a copy number baseline, sequence variant baseline, and exon or tile assay baseline, are available on the sequencer to which the assay is transferred. You can either import the user-defined presets or create the presets on the target sequencer. The names of the presets that you import or create must be identical on both the source and the target sequencers.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Assays ▶ Manage Assays** screen, place the pointer over the row of a locked, user-defined assay, then click **Export**.
The assay parameter files are downloaded to the computer as a ZIP file and are available for import to another sequencer.
Panel reference files are not included in the exported file.
3. In the menu bar, click **Assays ▶ Manage Assays**.
4. In the **Manage Assays** screen, click  **Import Assay**.
5. In the **Import Assay** dialog box, click **Select Assay file**, go to the folder on the computer that contains the assay that was exported from a sequencer, then select the ZIP file for the assay.
6. Click **Upload**.
The assay appears in the list of assays in the **Assays ▶ Manage Assays** screen.

Edit a draft assay

Role—System administrator, organization administrator and manager.

When a user-defined assay is first created, the assay is in draft status. While the assay is in draft status, it can be edited. A locked assay cannot be edited. Each time you edit a draft assay, the software creates a revision of the draft assay. The revision number of the assay is shown in the assay details. For more information, see “View assay details” on page 258.

You can optimize the configuration of an assay when you edit an assay. You can plan a run with a draft assay, view the results, then change the configuration and plan new runs before you lock the draft assay.

You can view the history of changes made to a draft assay. For more information, see “Review and download audit records for an assay” on page 266.

IMPORTANT! Use the default parameter settings for assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a draft assay, then click **Edit**. The **Edit Assay** workflow reopens.
3. Edit the options on each assay step as desired. For more information, see “Assay options” on page 240.
4. Click **Save** to save changes to the assay.

The assay remains a draft until you lock the assay. For more information, see “Lock a draft assay” on page 261.

Lock a draft assay

Role—System administrator, organization administrator and manager.

When a manager or administrator first creates an assay, the status of the assay is **Draft**. With the exception of changes to QC parameters, you cannot edit a locked assay. A locked assay cannot be deleted.

1. In the menu bar, click **Assays ► Manage Assays** to open the **Manage Assays** screen. An assay full name followed by **Draft** indicates that the assay is in draft status.
2. In the **Manage Assays** screen, place the pointer over the row of a draft assay, then click **Lock**.

The assay is now locked and cannot be edited or deleted. **Draft** is removed from the assay full name in the **Assays ► Manage Assays** screen.

Delete a draft assay

Role—System administrator, organization administrator and manager.

When an assay is in draft status, an administrator or manager can delete it from the software. Only draft assays can be deleted. Draft assays are indicated by **Draft** next to the **Assay Full Name**.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a draft assay, then click **Delete**.
3. In the **Delete Assay** dialog box, click **Yes** to confirm the deletion.

The draft assay is removed from the software and is no longer available in the **Assays / Manage Assays** screen.

Edit the QC parameters of a locked assay

Role—System administrator, organization administrator and manager.

Administrators and managers can edit the QC parameters of a locked assay. The QC parameters can be adjusted according to research needs that can change over time. You can edit the QC parameters of any locked assay, including system-installed assays, if needed. When the QC parameters are changed, the edited QC parameters are applied to any new run that uses the assay. Edits to QC parameters do not affect runs that use the assay and are completed before edits are made. **Chip Kit** should not be changed.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a locked assay, then click **Edit QC**.
The **Edit QC** dialog window opens.
3. Edit the QC parameters as desired.
To reset the QC parameters to the original values in a subsection, such as **Run QC** or **Sample QC – DNA**, click **Reset** in the upper right corner of the subsection.
4. Click **Save**.

The new QC parameter values are saved and applied in any run that uses the assay after the changes are saved.

Oncology content packages for use in assays

Administrators and managers can use a new or updated Oncology content package that is installed in Genexus™ Software in an assay.

Oncology content packages can include new or updated gene lists, OncoPrint Variant Annotations rulesets, Variant Naming Services, and cancer type sample attributes. For more information see “Oncology content packages with gene lists” on page 348 and “Cancer types in Oncology content packages” on page 73.

Before you use updated Oncology content updates, you need to download and install the update package. Updated Oncology content packages are available from apps.thermofisher.com.

- If a Thermo Fisher™ Connect Platform account has been configured, download and install software directly from thermofisher.com/connect. See “Download and install software updates from the App Store” on page 370
- Alternatively, if the instrument is not connected to the internet, use a USB drive to install and update the software. See “Format a USB drive for a software update” on page 372 and “Download and install software updates with a USB drive” on page 371.

After an Oncology package update is installed, you can add a new or updated OncoPrint Variant Annotations ruleset, Variant Naming Service, or gene list to an assay in the **Panel** step with the following options when you create a new assay, or edit an assay.

- **OncoPrint Variant Annotations Ruleset**—Add an updated OncoPrint Variant Annotations ruleset to an assay.
- **Use Latest Variant Name**—Add an updated variant naming service to an assay.
- **Gene List**—Add an updated gene list to an assay.

For more information, see “Assay creation overview” on page 233, “Edit a draft assay” on page 261, and “Panel step assay options” on page 240.

Use the latest ClinVar annotation source to annotate results

Role—System administrator, organization administrator and manager.

Administrators and managers can enable an option that changes the version of the ClinVar annotation source that an assay uses to annotate the results. In Genexus™ Software, this option is called **Use Latest Annotations**. You can enable this option when you create or edit an assay, or enable the option for a locked assay that is available in the software. To use this option, you must use Genexus™ Software 6.8 or later. The **Use Latest Annotations** option affects only the ClinVar annotation source.

If this option is enabled, and the version of ClinVar in the assay is earlier than a more recent version that is currently included in the software version that you use, the most recent ClinVar version in the software is then used to annotate the results. For example, to sequence samples using the OncoPrint Comprehensive v3 - GX5 - DNA and Fusions - w5.0.2 assay, which by default includes ClinVar 20200329, and you instead want to use the most up-to-date ClinVar annotation source, then you can use this option to apply ClinVar 20220709 to annotate the results.

When enabled, the **Use Latest Annotations** option applies only either the ClinVar version that is included in the annotation set that is configured in the assay, or the latest version of ClinVar that is available in the software version that you use.

To enable the **Use Latest Annotations** option, follow these steps that correspond with the status and state of the assay.

Assay status	Steps
You can set the option in a <i>locked</i> assay that is available in the software.	- In the menu bar, click Assays ▶ Manage Assays. - Find a locked assay of interest in the Manage Assays screen. - Place the pointer over the row of a locked assay, then click Use Latest Annotations. - In the Use Latest Annotations dialog box, click Yes. The link in the row for the assay changes to Use Default Annotations.
You can set the option in a <i>draft</i> assay that was created, or copied and edited, in Genexus™ Software 6.6 or earlier. An assay full name followed by Draft indicates that the assay is in draft status.	- In the menu bar, click Assays ▶ Manage Assays. - In the Manage Assays screen, place the pointer over the row of a draft assay, then click Lock. - Place the pointer over the row of the assay that you locked, then click Use Latest Annotations. - In the Use Latest Annotations dialog box, click Yes. The link in the row for the assay changes to Use Default Annotations.
You can set the option when you create or edit a draft assay.	Enable Use Latest Annotations in the panel step. For more information, see “Assay creation overview” on page 233, “Edit a draft assay” on page 261, and “Panel step assay options” on page 240.

This setting persists so that every time a new version of ClinVar annotation source is installed in the software, that new version is used for any new run plan to which the assay is applied. When results are generated from the run plan, the most recent version of the ClinVar annotation source is used to annotate the results.

To revert to the ClinVar annotation source version that is included in the annotation set that is configured in the assay, see “Revert to the ClinVar annotation source included with the annotation set” on page 264.

Revert to the ClinVar annotation source included with the annotation set

Role—System administrator, organization administrator and manager.

If the **Use Latest Annotations** option has been enabled and saved to an assay, you can choose to revert to the ClinVar annotation source that is configured as part of the annotation set for the assay. This reversion affects only results from a new run plan to which the assays are applied.

For information on how to enable **Use Latest Annotations**, see “Use the latest ClinVar annotation source to annotate results” on page 263.

The ClinVar annotation source that is configured as part of the annotation set for the assay is considered to be the *default annotation source*. This ClinVar version is used for any new run plan to which the assay is applied, regardless of whether newer versions of the ClinVar annotation source is installed in the software.

Managers and administrators can enable the settings **Use Latest Annotations** and **Use Default Annotations**.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of the assay of interest, then click **Use Default Annotations**.
3. In the **Use Default Annotations** dialog box, click **Yes**.
The link in the row for the assay changes to **Use Latest Annotations**.

The ClinVar annotation source that is part of the annotation set is configured with the assay and is used for any new run plan to which the assay is applied.

Remove access to an assay

Role—System administrator, organization administrator and manager.

Administrators and managers can remove a locked, user-defined assay from use in the software by designating it obsolete. The assay is not deleted and a record of it is maintained in the audit trail. The results for any runs already performed with the assay remain available in the software. System-installed assays cannot be made obsolete.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a locked, user-defined assay, then click **Obsolete**.
3. Click **Yes** to confirm the operation.
The assay is no longer available for use in a run.

Download assay parameters

Role—System administrator, organization administrator and manager, operator.

Assay parameters can be downloaded and exported, for example to use that assay in another Genexus™ Integrated Sequencer.

Panel reference files are not included in the export.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a locked assay, then click **Download Parameters**.

The assay parameter JSON files are downloaded to the computer in a ZIP file and are available to review or import to another sequencer.

Review and download audit records for an assay

Role—System administrator, organization administrator and manager, operator.

Users can review and download the entire history of an assay in Genexus™ Software. An audit trail lists the records of actions made to the selected assay. Each time-stamped audit record shows the original assay values and any new values for each action.

The following user actions generate an audit record.

- Create a new assay
- Copy an assay
- Edit a draft assay
- Lock an assay
- Import an assay
- Export an assay
- Delete an assay
- Edit parameters in an assay

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Assays ► Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the assay of interest, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.
4. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the assay is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attribute mappings Manage Assays screen.



Manage presets

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Role—System administrator, organization administrator and manager.

Genexus™ Software includes system-installed presets to use in the software. Administrators and managers can create user-defined presets to use in the software. Manage presets in the tabs in the **Assays / Manage Presets** screen.

Annotation sets

Role—System administrator, organization administrator and manager.

Genexus™ Software includes preset annotation sets that can be selected for use in assays. If the annotation set that you need is not available for selection in the software, you can create an annotation set that includes the specific annotation sources that you need.

Annotation sets are compiled from various annotation sources to provide information about the biological meaning of variants and allow variant sorting and prioritization. Annotation sets are derived from public and private annotation databases for hg19 and GRCh38 human genome references.

- The Human Genome version 19 (hg19) reference is an early version of the human genome reference.
- The Genome Reference Consortium human genome assembly version 38 (GRCh38) is the latest version of the human genome reference.

Use the tools that are available in the **Assays / Manage Presets** screen in the **Annotation Sets** tab to create and manage annotation sets.

View annotation set details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details about an annotation set in Genexus™ Software.

1. Click **Assays ► Manage Presets**.
2. In the **Assays / Manage Presets** screen, in the **Annotation Sets** tab, click the annotation set name.
3. View the annotation set details.

Item	Description
Analysis Pipeline Version	The compatible version of the analysis pipeline, for which the annotation set can be used.
Last Modified By	For user-defined annotation sets, the sign-in name of the person who created or modified a user-defined annotation set.
Last Modified On	Date on which a user-defined annotation set was last modified.
Created By	User-defined annotation sets show the sign-in name of the person who created the annotation set. Annotation sets that are included with the software are labeled as System Installed .
Created On	Date on which a user-defined annotation set was created.
Status	Status of the annotation set, including: • Locked • Draft • Obsolete
Reference	Human genome reference that is available in the annotation set. • hg19 • GRCh38
Annotation Versions	A list of the annotation sources included in the annotation set, and the version of each annotation source.

Add a new annotation set

Role—System administrator, organization administrator and manager.

Administrators and managers can create user-defined annotation sets in Genexus™ Software. Annotation sets are available as an option when you create or copy an assay.

1. Click **Assays ► Manage Presets**.
2. In the **Manage Presets** screen, in the **Annotation Sets** tab, click **+ Add New**.
The **Create Annotation Set** screen opens.

3. In **Analysis Pipeline Version**, select the version of the analysis pipeline that you want to use for the new annotation set.
In most cases, you will select the **Analysis Pipeline Version** for the software version that you are currently using, which includes the latest annotation sources available in the software.
4. In **Name**, enter a name for the new annotation set, and an optional description in **Description**.
5. In the **Genome Reference** list, select a human genome reference to use for the new annotation set.
 - hg19
 - GRCh38
6. Add system-installed annotation sources to the annotation set. In the **Available Annotation Sources** list, select a system-installed annotation source and a version, then click **Add** ➤ to move it to the **Selected Annotation Sources** list.

The list of available annotation sources is based on the selected genome reference.

Available annotation source	Description
5000 Exomes	Population frequency information from the 5000 Exomes project.
ClinVar	An assessment of impact of the variants observed from NCBI ClinVar database. Source— http://www.ncbi.nlm.nih.gov/clinvar
COSMIC	A catalog of known somatic mutations in cancer tumor tissue. Source— https://cancer.sanger.ac.uk/cosmic
dbSNP	Single Nucleotide Polymorphism database. A database of genomic variants. The dbSNP annotation source in the software contains a flag carried by a subset of its SNPs that have been curated by UCSC to be "UCSC Common SNPs". For a variant to be annotated as a UCSC Common SNP, the variant is first annotated as being present in dbSNP, and it might also be classified as a UCSC Common SNP. Source— https://www.ncbi.nlm.nih.gov/snp/
DGV	Database of genomic variants—A curated database of human genomic structural variation. Source— http://dgv.tcag.ca/dgv/app/home
Disease Research Area	A hierarchically organized database of disease categories. Source— https://meshb.nlm.nih.gov
DrugBank	A database from which names of therapies that are used for researching variants are collated. Source— https://www.drugbank.ca
ExAC	Exome Aggregation Consortium—A database catalog of variant frequencies. ExAC is no longer part of the default annotation set. Source— https://gnomad.broadinstitute.org

(continued)

Available annotation source	Description
Gene Model	A set of genes that overlap with the variant. Sources— https://www.ncbi.nlm.nih.gov/refseq and http://www.ensembl.org
Gene Ontology	Standardized ontology for gene and gene projects (for example, functional role or localization). Source— http://geneontology.org
GNOMAD	The gnomAD v2.1.1 data set (Exomes). Different data sets are derived from Exome or whole genome sequencing, but GNOMAD v2.1.1 in Genexus™ Software uses Exome data only. Source— https://gnomad.broadinstitute.org
Named Variant	A list of known variants in the CFTR gene panel. This option is available in Genexus™ Software 6.8.2 and earlier, and not available in Genexus™ Software 7.0.
OMIM	Online Mendelian Inheritance of Man. Source— https://www.ncbi.nlm.nih.gov/omim
Pfam	A database of protein families that includes their annotations and multiple sequence alignments using hidden Markov models. Source— http://pfam.xfam.org
phyloP Scores	A measure of conservation of the protein across a wide range of organisms. Source— http://compugen.cshl.edu/phast (phast is a tool that generates the scores)
SIFT / PolyPhen	A prediction of the functional effect of a variant on a protein. Sources— https://sift.bii.a-star.edu.sg and http://genetics.bwh.harvard.edu/pph2

7. If the annotation source that you want does not exist in the software, import a new annotation source. The annotation source must be a TXT, BED or VCF file. For more information, see “Annotation source file structure” on page 271.

a. In the **Available Annotation Sources** list, select one of the custom categories.

- **Gene Set (Custom)**—Select this option to import a TXT file that contains a custom set of variants in a specific set of genes.
- **Genomic Regions (Custom)**—Select this option to import a BED file that contains a custom set of variants in specific gene regions.
- **Transcript Set (Custom)**—Select this option to import a TSV file that contains a custom set of transcript annotations.
- **VariantDB (Custom)**—Select this option to import a VCF file that contains a custom set of variant annotations.

b. Click **+ Add New Source** below the list.

c. Enter a name for the new annotation source.

- d. Enter a version for the new source.
 - e. Browse to select the new annotation source file.
 - f. Click **Upload**.
The new annotation source is added to **Available Annotation Sources** list.
 - g. Click **Add** ➤ to move it to the **Selected Annotation Sources** list.
8. Review the list of selected annotation sources, then add or remove sources, if needed. To remove a source from the **Selected Annotation Sources** list, select the checkbox, then click **Remove** ✕.
 9. Click **Save** to save the annotation set.
If an error occurs, check the annotation source files for format errors. Correct the text file, and then repeat this procedure with the corrected file. See “Annotation source file structure” on page 271.

The annotation set is listed in the **Annotation Sets** tab and is available to be added to an assay that you create or edit.

Annotation source file structure

Role—System administrator, organization administrator and manager.

If an annotation source that you need is not available for selection in the software, administrators and managers can create annotation source files and import the files into an annotation set.

You can create your own files for these four types of annotation sources. Use a text editor to create the file, then save the file in the correct file format that is appropriate for importing the file into the software.

Annotation source type	Description	File format for import
Genomic region	A set of regions	BED
Gene set	A set of genes	TXT
Transcript set	A set of transcripts	TSV
VariantDB	A set of variants	VCF

The types of annotation sources and the file formats and file content required for each annotation source are described as follows.

Genomic region annotation source file

You can provide user-defined annotation information for specific regions of interest in the genome in a genomic region annotation source file.

- The genomic region annotation source file must be a BED format file with four, tab-separated columns. Columns must list a chromosome number, start value, end value, and annotation values.
- Enter user-defined annotations in the **Annotation value** column of the file, as key-value pairs separated by semicolons.

An example file looks something like this.

#CHROM	Start	End	Annotation values
chr1	000000	000001	fld1=abc;fld2=123;fld3=this
chr1	000001	000002	fld1=abd;fld2=124;fld3=that
chr1	000002	000003	fld1=abe;fld2=125;fld3=this
chr1	000003	000004	fld1=abf;fld2=126;fld3=that
chr1	000004	000005	fld1=abg;fld2=127;fld3=this
chr1	000005	000006	fld1=abh;fld2=128;fld3=that

The hit level for a genomic region annotation source is not configurable and is always set to overlap.

Gene set annotation source file

You can provide annotation information for specific genes of interest in a gene set annotation source file. The file lists gene names and categories for those genes.

- The gene set annotation source file must be a TXT format file with two tab-separated columns. The gene name is in the first column and a corresponding category for the gene is in the second column. Entries for the columns must be comma-separated gene symbols, followed by a tab character and any annotation string to describe the gene category.
- The file can include an optional heading name for the columns. The second column name is used as the annotation key name. If the heading line is not provided, the default annotation key name of Name is used.

Here is an example file.

```
#Geneset5      Disease
Gene1, Gene2   disease1
Gene3          disease2
Gene4, Gene5, Gene6  disease3
Gene7, Gene8   disease4
```

Transcript set

You can use a transcript set to provide annotation information for specific transcripts of interest. A transcript set limits the annotations that are applied to your variant calls to annotations that match your list of preferred transcripts.

Use the following guidelines when you use or create a transcript set.

- The preferred transcript set must be a TSV format file.
- Ensure that the transcript set includes a header line that indicates the human genome on which it is based. If this reference header information is not included, the software processes the file as if it is based on the hg19 human genome reference. The options for the header line are:
 - **##reference=GRCh38**
 - **##reference=hg19**
- The contents of the file for the transcript set are in the format: **GENE_NAME transcript_accession_id1, transcript_accession_id2, ...**, with a tab character between the gene name and the first transcript ID.
- Gene names must be specified as HGNC gene symbols.
- A tab character is required after the gene name.
- A comma and a space character are required between transcripts.
- If gene name is missing in the file, all transcript annotations for that gene are preserved.

- Transcripts can be specified as RefSeq or Ensembl accession IDs that include a version. However, if the accession ID contains no decimal point version, then all versions of the transcript are matched. For example, Gene1 NM_00000006 is matched with all versions of this transcript. This annotation source uses a TSV format with tab characters.

The following is an example file with a header to indicate that the transcript set is based on the GRCh38 human genome reference.

```
##reference=GRCh38
GENE REFSEQ AND ENSEMBL TRANSCRIPTS
Gene1 NM_000000006.2, NM_000000005.3, ENST000000000007.3
Gene2 NM_000000001.1, NM_000000007.1, NM_0000000004.5,
NM_000000008.3, ENST000000000006.4
GENE REFSEQ AND ENSEMBL TRANSCRIPTS
Gene1 NM_000000006.2, NM_000000005.3, ENST000000000007.3
Gene2 NM_000000001.1, NM_000000007.1, NM_0000000004.5, NM_000000008.3,
ENST000000000006.4
```

VariantDB annotation source file

You can provide user-defined annotation information for specific variants of interest in a VariantDB file.

- The VariantDB annotation source file must be a VCF format file
- User-defined VariantDBs must have unique names. For example, name and version number should be combined to ensure unique names.

```
1 | Name: variantdbA_v1      Version: v1
2 | Name: variantdbA_v2      Version: v2
3 | Name: variantdbA_v3      Version: v3
```

- A tab-delimited file with a header line is required.

```
##fileformat=VCFv4.1
#CHROM    POS      ID      REF      ALT      QUAL    FILTER  INFO
chr1      124535436  COSM00001  TG      AA      .      .      AMPID=AMPL495041;TEMP_ID=0
chr1      124535494  COSM00002  G       T       .      .      AMPID=AMPL495041;TEMP_ID=1
chr1      128808434  COSM00003  T       A       .      .      AMPID=AMPL30014;TEMP_ID=2
chr1      124597624  .         T       G       .      .      AMPID=AMPL30014;TEMP_ID=3
chr1      136671158  .         TT      CA      .      .      AMPID=AMPL30014;TEMP_ID=5
chr1      141128903  COSM00006  TTG     CTT     .      .      AMPID=AMPL30014;TEMP_ID=6
```

- We recommend that the user-defined input file provided to VariantDB be left-aligned. Left alignment is used to normalize the positions of ambiguous INDELs that can be placed at multiple positions.

Further information on VCF format is as follows.

- **Official specification of VCF (Variant Call Format) version 4.1**

<http://www.1000genomes.org/wiki/Analysis/Variant%20Call%20Format/vcf-variant-call-format-version-41>

In VCF format files, missing values are represented by dots. The content must be tab-separated. Ensure that no extra or hidden characters are added to the VCF files, which may occur when they are opened in programs like Excel™ or Microsoft™ Word, or when emailed as an unzipped attachment.

- **Mandatory headers required when creating a VariantDB file**—The following three headers must be present in the first three lines of the VCF file (FORMAT and Sample columns are optional in VCF files):

```
##fileformat=VCFv4.1
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
#CHROM    POS      ID       REF      ALT      QUAL     FILTER    INFO     FORMAT
Sample
```

- **Hit-level information in a VariantDB file**—You can adjust the hit level of each VariantDB file individually by including this information in the header. The following hit-level parameters can be included in the VCF header.
 - **##HITLEVEL=overlap** matches all annotations whose loci overlap with variant.
 - **##HITLEVEL=locus** matches all annotations whose loci start at the variant locus.
 - **##HITLEVEL=allele** matches all annotations that are locus matches plus have at least one allele in common with variant.
 - **##HITLEVEL=genotype** matches all annotations that are 'allele' matches where the genotypes also match.
 - **##HITLEVEL=auto** matches the most specific hit level possible, which could be any of the hit levels listed above.
- **Mandatory columns required in the VCF file when creating a VariantDB**—Providing FORMAT and SAMPLE fields is not mandatory according to the official VCF specification. However, in order to perform a "genotype" hit level match in the software, you must specify a GT (genotype) for the variant in the FORMAT column.

An example of a variant with a GT field of 0/1 in the FORMAT field of a VCF file is given below:

```
chr1      141128903    COSM00006    TTG CTT . .
AMPID=AMPL30014;TEMP_ID=6    GT    0/1
```

If only an "overlap" or "locus" or "allele" match is needed, you do not need to specify a GT field. However, the missing values must be represented by dots in the appropriate columns. For example:

```
chr1      141128903    COSM00006    TTG CTT . .
AMPID=AMPL30014;TEMP_ID=6...
```

If the "auto" hit level match is chosen, the software tries to find the most specific hit level match possible. However, if no GT value is supplied, the most specific hit level possible is an allele match, as there is no GT value to do an allele or genotype level match.

- **How to filter on VariantDB**

- **Option 1**

The software automatically exposes a filter on the first INFO key of the VariantDB VCF file if such a key is specified and if the ID field of the VCF file is missing.

- Consider the following example.

```
##fileformat=VCFv4.1
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
#CHROM    POS      ID      REF      ALT      QUAL      FILTER      INFO
FORMAT    Sample
chr1      124535436  .      TG      AA      .      .
AMPID=AMPL495041;TEMP_ID=0...
chr1      128808434  .      T       A       .      .      AMPID=AMPL30014;TEMP_ID=2...
```

If the above VCF file with two variants is used to make a VariantDB in the software, you can filter on the AMPID field, since the AMPID key is the first INFO key present in the INFO field of the VCF file and the ID fields are missing (represented by dots).

- **Option 2**

If the INFO field is not populated, filtering is enabled on the ID column.

Consider the example below, in which the INFO field is missing and represented with a dot:

```
##fileformat=VCFv4.1
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
#CHROM    POS      ID      REF      ALT      QUAL      FILTER      INFO
FORMAT    Sample
chr1      124535436  COSM00001  TG      AA      .      .      ....
chr1      141128903  COSM00006  TTG     CTT     .      .      ....
```

If the VCF file above with two variants is used to make a VariantDB in the software, you can filter on the ID field, because the INFO field of the VCF file is not populated.

Obsolete annotation sets

Role—System administrator, organization administrator and manager.

Annotation sets that have been added or imported in Genexus™ Software can be made unavailable in the software. You cannot designate system-installed annotation sets as obsolete.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Annotation Sets** tab, place the pointer over the row of the user-defined annotation set that you want to make unavailable, then click **Obsolete**.

The annotation set status is **Obsolete** in the list of annotation sets.

Updated versions of annotations

Role—System administrator, organization administrator and manager.

In Genexus™ Software, for some annotations, administrators and managers can change the version of the annotation source that is used by an assay.

When you use an assay with the **Use Latest Annotations** option enabled, you can view the version of the annotation source that is applied when you plan a run, reanalyze a sample or a run, download files, view the run report screen, and view PDF reports.

You can enable or disable this setting when you perform the following actions in the software.

- Create an assay.
- Place the pointer over the row of a locked assay in the **Assays / Manage Assays** screen.

Option	Description
Use Latest Annotations is in the Panel step of the Assays / Create Assay screen when you create an assay.	• When this option is enabled, the latest version of the ClinVar annotation source that is available in the software is used by the run plan to which the assay is applied. • If this option is disabled, the ClinVar annotation source version that is associated with the assay is used regardless of whether newer versions of the annotation source are installed in the software. That is, if a newer version of the ClinVar annotation source is installed in the software and the setting is disabled, that version is not associated with the assay. This option is available only for the ClinVar annotation source in the software that is used to annotate data. For more information, see “Panel step assay options” on page 240.
Use Latest Annotations is shown when you place the pointer over the row of a locked assay in the Assays / Manage Assays screen.	The most recent version of the ClinVar annotation source that is available in the software is used by the assay for any new run plan. As a result, run plans created with this assay, before this setting is enabled, might use different versions of the ClinVar annotation source than run plans that are created after this setting is enabled, depending on which ClinVar annotation source is available at the time the run plan is created.
Use Default Annotations is shown when you place the pointer over the row of a locked assay in the Assays / Manage Assays screen.	The ClinVar annotation source that is included in the annotation set that is configured in the assay is considered to be the <i>default annotation source</i> . This default is used for any new run plan to which the assay is applied, regardless of whether newer versions of the annotation sources are installed in the software.

Get the latest annotation source

Role—System administrator.

A system administrator can get the latest additions, deletions, or changes to the ClinVar database in Genexus™ Software.

To download and install a new version of the ClinVar database from the Thermo Fisher Scientific App Store, you must first do the following.

- Configure a Thermo Fisher™ Connect Platform account.
For instructions, see “Configure a Connect Platform account with a QR code” on page 352.
 - Ensure that the Genexus™ Integrated Sequencer has the latest software version of Genexus™ Software.
 - Connect the sequencer to the internet.
1. In the menu bar, click  (**Settings**) ▶ **Software Updates**, then click **Software Updates** in the upper right corner.
The **Software Updates** screen opens with **App Store** enabled.
 2. (Optional) click the desired package name to view the installation details and version information in the **View Package Details** pane.
 3. Click **Download** in the **Actions** column in the row of the software package of interest.
ClinVar packages have a **Category** type of **Configurations**. ClinVar packages are named as **ClinVar <Release date>**.
After the download is complete, the **Download** link changes to an **Install** link.
 4. Click **Install**.
 5. In the **Install Annotation Source** dialog box, click **Yes**.
 6. When installation of the software package is complete, click **Installed Software** to verify the installation.

Filters and filter chains

Role—System administrator, organization administrator and manager.

Genexus™ Software includes system-installed filter chains to narrow the list of variants that are shown in sample results.

A filter chain is a set of filters that you can add to an assay to filter in or filter out variants of interest, then view the filtered results after a sequencing run. You can also apply a filter chain to dynamically change the view of the variants as you view sample results.

System-installed filters are based on public and proprietary annotation sources and data types that are included in the software. System-installed filter chains are locked and cannot be changed. You cannot edit system-installed filter chains.

If you associate a filter chain with an assay, the filters that are included in the filter chain are applied to sample results that use the assay. You can also apply a filter chain directly to sample results to dynamically change the view of the variants, then save the filter chain to the results. See “Show variant results based on a filter chain or a gene list” on page 174. When you view sample results, a more stringent filter yields fewer results, and a less stringent filter yields more results.

If system-installed filter chains do not meet research needs, administrators and managers can create filter chains to refine sample results. User-defined filter chains can be created from one or more system-installed filters. Alternatively, a system-installed filter chain can be copied, then edited, to create a user-defined filter chain. For more information, see “Add a new filter chain” on page 279.

When you create a filter chain, the filter chain is in draft status. You can apply a draft filter chain to sample results, or add the filter chain to an assay that is in draft status. If you lock the filter chain, you can save it to an assay, then lock the assay. Reports that are generated with a filter chain that is locked can be electronically signed.

Review filter chain details

Role—System administrator, organization administrator and manager.

Administrators and managers can review the details of filter chains. Filter chains are used to narrow the list of sample results that are shown in the software and in results files. If a filter chain does not meet your needs, you can create a new filter chain, or copy and edit an existing filter chain and adjust the settings. Then, the new or edited filter chain is available in the software to add to an assay before a sequencing run, or to apply to sample results after a sequencing run.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click a filter chain.
4. Review the following details.
 - The name of the filter chain.
 - The software version in which the filter chain originated. A filter chain that is compatible with an earlier software version can be associated with an assay, or applied to sample results after a sequencing run. You might use a filter chain from an earlier software version to troubleshoot results based on an earlier filter chain version, or to compare results between two versions of a filter chain.
 - Whether the filter chain is system-installed or user-defined.
 - The date that the filter chain was created.
 - A brief description of the filter chain.
 - The filters that are included in the filter chain and the query that is used to apply the filters.
 - The order in which the filters that are contained within the filter chain are applied.

Add a new filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can create new filter chains in Genexus™ Software. A filter chain is a set of filters that you add to an assay before a sequencing run, or apply to sample results after a run to narrow the list of sample results that are shown in the software and in results files.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name for the new filter chain.
5. Specify the **Genome Reference** to use.
The default reference is hg19.
6. *(Optional)* In the **Annotation Sets** list, select an annotation set to use with the filter.
7. *(Optional)* Click **Add description** to add information.
8. In the **Add Filter** dropdown list do the following.
 - a. Select a filter to include in the filter chain, then select the query options for the filter. For more information about the options for each filter, see “System-installed filters” on page 554.

Note: If the list does not populate with the filters that are available in the software, restart the sequencer.

 - b. Click  **Update** to add the filter to the filter chain.
9. Repeat step 8 for each filter that you want to add to the filter chain.
10. *(Optional)* To make final changes to the filters to include in the filter chain, place the pointer over a filter listed below **Query**, then make the following changes.
 - Click  **Create Group** to create a filter chain with groups. For more information, see “Create a filter chain with groups” on page 280.
 - Click  **Edit** to make changes to the filter.
 - Click  **Delete** to delete the filter.
11. Click **Save**.
The new filter chain is listed in the **Filter Chains** table and is available for use.

The filter chain is in draft status.

You can lock the filter chain if needed. Locked filter chains cannot be edited or deleted.

Create a filter chain with groups

Role—System administrator, organization administrator and manager.

Administrators and managers can create filter chains with groups in Genexus™ Software.

Filter chains are used to narrow the list of sample results that are shown in the software and in results files.

You can group filters together to combine a subset of filter criteria in a filter chain. This procedure is an example of how to create a filter chain with groups. When applied to a sample result, the filter chain in this example returns all variants that are CNV variants of the ERBB2 gene as well as all BigDel or BigDup variants of the BRCA1 gene.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name for the new filter chain.
5. (Optional) Specify the **Genome Reference** to use.
The default reference is hg19.
6. (Optional) In the **Annotation Sets** list, select an annotation set to use with the filter.
7. In the **Add Filter** dropdown list, select the **CNV Somatic Confidence Range** filter.
You must add a filter before you see the query options.
 - a. (Optional) Select the query options for the filter.
For more information about the options for each filter, see “System-installed filters” on page 554.
 - b. Click  **Update** to add the filter to the filter chain.

Information for the filter is shown below **Query**.

8. Add a filter to the group.
 - a. Place the pointer over the information for the newly added filter, then click  **Create Group**.
 - b. In the **Add Filter** dropdown list, select **Variant Type**.
 - c. In the list of annotations that appears, select **CNV**, then click  **Update** to add the filter to the group.
 - d. In the dropdown list of filters for the group, select **Gene Symbol**.
 - e. In the search field that appears, enter **ERBB2**, then select **ERBB2**.
 - f. Click  **Update** to group the gene symbol filter with the variant type.
The filter group of a CNV Variant Type AND ERBB2 Gene Symbol is added to the filter chain.

9. Add another filter group.

- a. In the dropdown list of filters for a new group, select **Gene Symbol**.

Ensure that you select the filter list that is associated with the group. The parentheses in the information for the groups indicate the start and the end of the groups. In addition, the indentation of the dropdown lists for the filters indicates the group.

- b. In the new pane that appears, click **OR**, then, in the search field, enter **BRCA1**, then select **BRCA1**, then click  **Update** to add the filter.

Information about the filter that you have created appears.

- c. Place the pointer over the information for the BRCA1 filter, then click  **Create Group**.

- d. In the new pane that appears, click **AND**, then, in the dropdown list of filters, select **Variant Subtype**.

- e. Select **BigDel** and **BigDup**, then click  **Update** to add the filter.

The filter group of an BigDel and BigDup variants of the BRCA1 gene is added to the filter chain.

10. Click **Save**.

The filter chain with groups is listed in the **Filter Chains** table and is available for use.

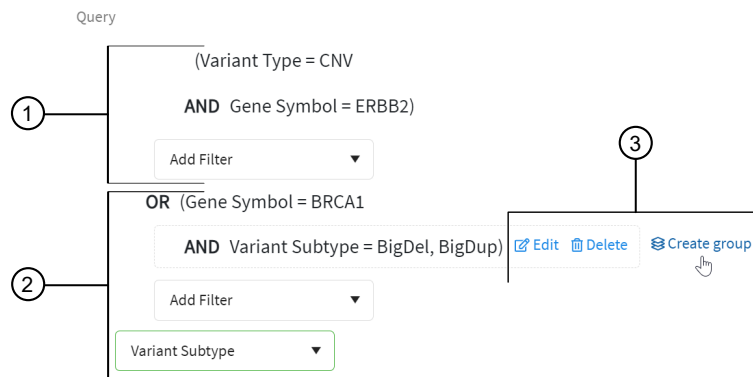


Figure 3 Example of a filter chain with groups

- ① A group of filters in the filter chain.
- ② Another group of filters in the filter chain.
- ③ Actions that are available when you place the pointer over the row of the filter.

Copy a filter chain from the Filter Chains tab

Role—System administrator, organization administrator and manager.

Administrators and managers can copy then edit a filter chain in Genexus™ Software to create a new filter chain. Use the filter chain to narrow the list of sample results that are shown in the software and in results files.

To copy and edit a system-installed filter chain, copy the locked system-installed filter chain, then make changes to the query options for the filter chain if needed.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Filter Chains** tab.
3. Place the pointer over the row of the filter chain of interest, then click **Copy**.
The **Copy Filter Chain** screen opens.
4. Change the software-generated name of the filter chain, then make changes if needed. You can edit the query options, add new filters, or change the annotation set.
5. Click **Save**.
The new filter chain is added to the **Filter Chains** tab.

The copied filter chain is listed in the **Filter Chains** table and is available for use.

Edit a filter chain from the Filter Chains tab

Role—System administrator, organization administrator and manager.

Administrators and managers can edit filter chains in Genexus™ Software that are not locked. Use the filter chain to narrow the list of sample results that are shown in the software and in results files.

Filter chains that are locked cannot be edited. System-installed filter chains are always locked and cannot be edited.

When you edit a filter chain that is applied to sample results after a sequencing run, the changes that you make apply to all sample results to which the filter chain is applied. Sample results that are not locked are immediately filtered based on the edited filter chain. As a result, the **Filtered In** count shown in the results may change.

If you want to edit a filter chain when you view sample results, see instead “Show variant results based on a filter chain or a gene list” on page 174.

1. Click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Filter Chains** tab.
3. In the **Filter Chains** table, place the pointer over the row of the filter chain of interest, then click **Edit**.
4. In the **Edit Filter Chain** dialog box, change the options for the filter.
For more information about the options for each filter, see “System-installed filters” on page 554.
5. Click **Save**.

The updated filter chain is listed in the **Filter Chains** table and is available for use.

Delete a filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can delete user-defined filter chains in Genexus™ Software. Only filter chains in draft status that are not locked can be deleted. You cannot delete a filter chain that is saved with a sample result.

System-installed filter chains are always locked and cannot be deleted.

1. Click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Filter Chains** tab.
3. In the **Filter Chains** table, place the pointer over the row of the filter chain that you want to delete, then click **Delete**.
The **Delete Filter Chain** dialog box opens with the message *Are you sure that you want to delete the filter chain?*
4. In the **Delete Filter Chain** dialog box, click **Yes** to delete the filter chain.

The filter chain is removed from the software and is no longer listed in the **Filter chains** table, and is no longer available for use.

To make a user-defined filter chain that is saved to a sample result unavailable for use, you can obsolete the filter chain.

Lock a filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can lock user-defined filter chains in Genexus™ Software. Filter chains are used to narrow the list of sample results that are shown in the software and in results files. Filter chains that are locked cannot be edited or deleted. System-installed filter chains are always locked.

1. Click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Filter Chains** tab.
3. In the **Filter Chains** table, place the pointer over the row of the filter chain that you want to lock, then click **Lock**.
4. In the **Lock Filter Chain** dialog box, click **Yes** to lock the selected filter chain.

The filter chain is now locked and cannot be edited or deleted.

Obsolete a filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can make user-defined filter chains that are locked unavailable in the software. You cannot designate as obsolete system-installed filter chains.

When you obsolete a filter chain, no change is made to sample results that already have the filter chain applied.

1. Click **Assays ▶ Manage Presets**.
2. In the **Filter Chains** table, place the pointer over the row of a filter chain that you want to make unavailable, then click **Obsolete**.

The filter chain status is **Obsolete** and is not available for use.

Review and download audit records for a filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can review and download the entire history of a filter chain in Genexus™ Software. Filter chains are used to narrow the list of sample results that are shown in the software and in results files.

An audit trail lists the records of actions made to the selected filter chain. Each time-stamped audit record shows the original filter chain values and any new attribute values for each action.

The following actions generate an audit record.

- Create a new filter chain
- Copy a filter chain
- Edit a filter chain
- Lock a filter chain
- Delete a filter chain
- Obsolete a filter chain

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Place the pointer over the row of the filter chain of interest, then click **Audit**.
4. Click a time stamp in the left of the screen to view details for each audited action.
5. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp. Depending on the browser settings, the software downloads the file or prompts you to open or save the file.

User-defined filter chain examples

Role—System administrator, organization administrator and manager.

Administrators and managers can create filter chains for use in Genexus™ Software. The following examples are provided.

- “Create a gene symbol filter chain” on page 286
- “Create a COSMIC ClinVar and Variant Impact filter chain” on page 286
- “Create a PValue/dbSNP/Variant Type filter chain” on page 288

Create a gene symbol filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can create a filter chain that is based on gene symbols. Filter chains are used to narrow the list of sample results that are shown in the software and in results files.

This procedure shows how to create a filter chain that narrows results to only specific genes. For more information, see “Gene Symbol filter” on page 570.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and optional description.
5. Select the **Genome Reference** for the filter chain.
The default reference is hg19.
6. (Optional) In the **Annotation Sets** list, select an annotation set to limit the list of filters.
7. In the **Add Filter** list, select the **Gene Symbol** filter.
8. Enter genes of interest in the search field, then in the list that appears, select the checkbox for each gene symbol that you want to add.
9. After you select the gene symbols that you want to include, click **🔗 Update**.
The selected genes are added to the filter chain.
10. Click **Save**.

The gene symbol filter chain is listed in the **Filter Chains** table and is available for use.

Create a COSMIC ClinVar and Variant Impact filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can create a filter chain that includes both annotation source filters and variant data type filters. Filter chains are used to narrow the list of sample results that are shown in the software and in results files.

This procedure shows how to create a COSMIC, ClinVar, Allele Fraction, and Variant Effect filter chain. In addition, this example shows how to include unannotated variants in the filter chain. Unannotated variants are variants that do not have annotation information in the annotation source. If you include

unannotated variants in a filter in the filter chain, variants that are not annotated by that filter are listed in the results when the filter chain is applied.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and for the filter chain.
5. Select the **Genome Reference** for the filter chain.
The default reference is hg19.
6. In the **Annotation Sets** list, select an annotation set to limit the list of filters, if needed.
7. Select the filters to add to the filter chain.
To include unannotated variants in the results, select **Include Unannotated** for each filter selection.
 - a. In the **Add Filter** list, select a version of the **COSMIC** filter.
 - b. Select the annotations that you want to include in the filter.
For more information, see “Add a new filter chain” on page 279 and “COSMIC filters” on page 561.
 - c. Click  **Update**.
 - d. In the **Add Filter** list, select a version of **ClinVar**.
 - e. Select the annotations that you want to include in the filter.
For more information, see “Clinvar filters” on page 557.
 - f. Click  **Update**.
 - g. In the **Add Filter** list, select **Allele Frequency**.
 - h. Enter a range value.
For more information, see “Allele Frequency filter” on page 556.
 - i. Click  **Update**.
 - j. In the **Add Filter** list, select **Variant Effect**.
 - k. Select the value that you want to include in the filter to detect the effect of variants on coding sequences.
For more information, see “Variant Effect filter” on page 579.
 - l. Click  **Update**.
8. Click **Save**.

The copied filter chain is listed in the **Filter Chains** table and is available for use.

Create a PValue/dbSNP/Variant Type filter chain

Role—System administrator, organization administrator and manager.

Administrators and managers can create a filter chain with multiple filters. Filter chains are used to narrow the list of sample results that are shown in the software and in results files.

This procedure shows how to create a filter chain with PValue, dbSNP, and Variant Effect filters.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and optional description.
The default reference is hg19.
5. In the **Add Filter** list, select the filters to add to the filter chain.
 - a. Select the **PValue** filter, select **Between**, **Greater than or equal to**, or **Less than or equal to**, enter a range, then click  **Update**.
 - b. Select a version of **dbSNP**, then click  **Update**.
 - c. Select **Variant Effect**, in **Select Specific Annotations**, select **In** or **Contains**, then select the annotations to include in the filter to detect the effect of variants on coding sequences, then click  **Update**.
6. Click **Save**.

The new filter chain is available for selection when you create or edit an assay, or view sample results. For more information, see Chapter 8, “Create and manage assays” or “Show variant results based on a filter chain or a gene list” on page 174.

Copy number baselines

Role—System administrator, organization administrator and manager.

A copy number baseline is a set of control samples that are used to create a baseline for the detection of copy number variants (CNVs). A copy number baseline is associated with a panel file in the software. CNV analysis is based on read counts for each amplicon.

Copy number baselines can be added only to assays that support CNV detection.

Known sources of variability include pool imbalance (when an assay has more than one pool of amplicons), total number of reads, per-amplicon attributes of GC proportion, and length of the amplicon insert. In practice, other systematic variability that is observed is modeled in the software.

Copy number estimates are made using a proprietary algorithm. A copy number baseline is created for each assay separately.

There are three types of copy number baselines available in Genexus™ Software for use with the following types of libraries.

- Ion AmpliSeq™ libraries
- Ion AmpliSeq™ HD libraries
- Low-coverage whole-genome libraries

Administrators and managers can create new copy number baselines, create copies of existing copy number baselines, and augment existing copy number baselines. When a baseline is augmented, an existing copy number baseline is copied and saved with a new name. Additional samples can then be added to the new baseline.

Somatic CNV calling

Somatic CNV calling uses a proprietary algorithm to make copy number estimates.

The Variability Correction Information Baseline (VCIB) algorithm uses a bioinformatics baseline, which is created using at least 48 diverse samples, to allow the analysis of corrected log2ratios of amplicons of identified CNV regions (usually genes) in sample data that is entered. This algorithm is followed by a correction algorithm for the percent tumor cellularity recorded for the sample to give copy number and confidence interval data for the identified CNV regions. The algorithms used to calculate the corrected log2 ratios and the correction for tumor fraction are proprietary.

Use the following guidelines when you select samples to use in a baseline of CNV detection type **Somatic (CNV VCIB)**.

- You must use a minimum of 48 samples. At least 6 of the samples must be normal germline samples to create a **Somatic (CNV VCIB)** baseline.
- Select a cohort of diverse sample types. This includes germline samples and cell-lines.
- Select samples with no known CNV aberrations. If this information is not available, a large cohort size can compensate any CNV aberrations.

For details about CNV call results in the **Sample Results** screen, see the user guide for the assay that you use.

Germline CNV calling

Germline CNV calling uses a proprietary algorithm to make copy number estimates.

In Ion AmpliSeq™™, Ion AmpliSeq™ HD, and OncoPrint™ assays, germline copy number estimates are performed in small units called tiles. Tiles are short segments of sequences that are about 100bp long. In the target region file, the algorithm defines a group of overlapping amplicons as a contig. Each contig is divided into tiles that are approximately 100bp long. The algorithm helps ensure that the tiles in the same contig are about the same size.

When Genexus Software calculates the mean base coverage for each tile, the same normalization that was done by the CNV baseline creation step is performed on the test sample. The software then compares the mean coverages to expected mean coverage for the tiles in the baseline samples. The software further derives the posterior likelihood for every tile, with posterior likelihood for every possible ploidy value (0 to 10), given the coverage observation.

The algorithm uses a model for the whole chromosome to determine the ploidy value of every tile, instead of deciding the most likely ploidy value for a CNV segment tile by tile. A transition probability between two adjacent tiles to model the likelihood that two adjacent tiles are having the same ploidy value is used. The likelihood of an assignment of ploidy values to a list of tiles is the multiplication of the likelihood of the assigned ploidy of every tile times all the transitional probabilities between adjacent tiles based on the assigned ploidy values. The software finds the most likely assignment, and that is reported as the CNV result.

For details about CNV call results in the **Sample Results** screen, see the user guide for the assay that you use.

View copy number baseline details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details for the copy number baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. In the **Copy Number Baselines** table, click the name of a copy number baseline and review the details.

The details that are shown depend on the setup of the copy number baseline. Annotation versions that were used to create copy number baselines are also shown but are not listed in this table.

Copy Number Baseline Details

Item	Description
Created By	Sign-in name of the person who created the baseline. System-installed baselines are included with the software.
Created On	Date on which the baseline was created.
Number of Normal Samples	The number of normal samples that are included in the baseline.
Gender	The biological sex of the sample: Female , Male , or Unknown .
Number of Samples^[1]	The total number samples that are included in the baseline.
Amplification Type	Library amplification technology that is used in the baseline: IonAmpliSeq , IonAmpliSeqHD , or Low-Coverage Whole Genome .
Panel Name^[2]	Name of the panel that is included in the baseline.
Genomic Reference	The reference genome for the baseline: hg19 or GRCh38 .
Reference File Name	The name of the reference genome file that is used in the baseline.

^[1] Applies only to baselines for use with Somatic (CNV VCIB) panels.

^[2] Applies only to baselines for use with **IonAmpliSeq** or **IonAmpliSeqHD** amplification types.

You can click ... **(More Options)** to perform actions such as obsolete, export, copy, edit, add or remove notes, audit the copy number baseline and view and download the log for the baseline. The options that are available depend on whether the copy number baseline is system-installed or is a user-defined copy number baseline.

The options for each baseline are also available in the **Copy Number Baselines** table when you place the pointer over the row of a copy number baseline.

Create a copy number baseline for use with Ion AmpliSeq™ libraries

Role—System administrator, organization administrator and manager.

Administrators and managers can create a copy number baseline for detecting copy number variants (CNVs) using Ion AmpliSeq™ panels.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. In the **Create Copy Number Baseline** screen, select **AmpliSeq**.
5. Complete the **Panel** step.
 - a. Enter a name in **Copy Number Baseline Name**.

The name is limited to 50 characters and no spaces are allowed.
 - b. Select a panel.
 - c. Select a **CNV Detection Type**.
 - **Germline (CNV Informatics Baseline)**—Use CNV germline settings when all cells in the samples are expected to have the same copy number state in a diploid background (for autosomes). Usually, the copy number is expected to be an integer value (0,1,2,3,4 and so on), although this is not strictly required.
 - **Somatic (CNV VCIB)**—Use CNV somatic settings for samples that may not have a pure population of cells, as with a mixture of normal and cancer cells. The software enables you to enter the percentage of cancer cells that are present in the sample. In addition, the copy number may not be an integer value. The software considers both the copy number in the cancer cells and the copy number that is observed in the sample, due to some non-cancerous cells that do not include the copy number change.
 - d. Leave **Augment an Existing Copy Number Baseline as Base** deselected.

Use this option only to augment an existing baseline, that is, copy and add samples to a baseline that uses a **CNV Detection Type** of **Somatic (CNV VCIB)**. For more information see “Augment a copy number baseline for use with Ion AmpliSeq™ or Ion AmpliSeq™ HD libraries” on page 297.

- e. (Optional) Enter information about the baseline into the **Description** field.
 - f. Click **Next**.
6. For baselines that use a **CNV Detection Type** of **Somatic (CNV VCIB)**, complete the **Parameters** step. Accept the default settings or adjust the appropriate parameters, then click **Next**.
 7. In the **Samples** step, click an option to view the samples for one of the following categories.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

8. In the **Samples** step, select the required number of samples for the baseline. For accurate baselines, it is recommended that you include samples that were analyzed or generated with the same panels.

For any genomic region for which you want to detect CNVs, it is recommended that samples include at least 10 amplicons. That is, if the goal is to detect CNVs in a gene, at least 10 amplicons should be in or adjacent to that gene, ideally 12 amplicons or more. If fewer than 10 amplicons are present, only CNVs with greater copy numbers may be detectable.

For robust detection of CNVs, most of the amplicons in a panel should be in normal diploid regions. If more than 30 percent of the amplicons are in a CNV region, it may be difficult to detect the CNV. In practice, if a panel contains at least 5 genes widely spread across the genome, the panel will usually meet these criteria.

- For baselines that use a **CNV Detection Type** of **Germline (CNV Informatics Baseline)**, include at least 20 samples that are not expected to have germline CNVs. Replicate samples can be used but it is recommended that at least 10 unique samples from diverse populations. More than 10 samples should be used, if available.
- For baselines that use a **CNV Detection Type** of **Somatic (CNV VCIB)**, you must use a minimum of 48 samples, and at least 6 of the samples must be normal, to create a **Somatic (CNV VCIB)** copy number baseline. Select a cohort of diverse sample types, made up of samples with no known CNV aberrations. If this information is not available, a large cohort size can compensate any CNV aberrations.

If you do not select the minimum number of samples that are required for the baseline, an error message appears, and you cannot proceed. If needed, you can increase the **Minimum number of samples** parameter if you augment baselines that use a **CNV Detection Type** of **Somatic (CNV VCIB)**.

- a. Select the checkbox next to each sample to use for the baseline.
 - b. Click **Next**.
9. In the **Review** step, review the selections, then click **Launch**.
The new copy number baseline is shown to the **Copy Number Baselines** tab. You can monitor the creation of the baseline on this tab.

The new copy number baseline is locked and shown in the **Copy Number Baselines** tab with a **State** of **Completed** and is available for use in the software. Copy number baselines can only be added to assays that support CNV detection.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab, or click **Download Log** to download the log. The log file can help you troubleshoot the failure.

Create a copy number baseline for use with Ion AmpliSeq™ HD libraries

Role—System administrator, organization administrator and manager.

Administrators and managers can create a copy number baseline for detecting copy number variants using Ion AmpliSeq™ HD panels.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

The somatic settings in this type of copy number baseline are appropriate when you have a sample that may not have a pure population of cells, as with a mixture of normal and cancer cells. The software allows you to enter the percentage of cancer cells that are present in the sample, and in some cases the software can predict that percentage. In addition, the copy number may not be an integer value. The software considers both the copy number in the cancer cells and the copy number that is observed in the sample, due to some non-cancerous cells that do not contain the copy number change.

Note: Copy number variant calling for use with Ion AmpliSeq™ HD libraries copy number baselines are not optimized for performance.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. In the **Create Copy Number Baseline** screen, select **AmpliSeq HD**.
The **Create Copy Number Baseline** screen opens to the **Panel** step.
5. Complete the **Panel** step.
 - a. Enter a name in the **Copy Number Baseline Name** field.
The name is limited to 50 characters and no spaces are allowed.
 - b. Select a **Panel**.
 - c. The **CNV Detection Type** of **Somatic (CNV VCIB)** is automatically selected.
 - d. Leave **Augment an Existing Copy Number Baseline as Base** deselected.
Use this option only to augment an existing baseline, that is, copy and add samples to a baseline that uses a **CNV Detection Type** of **Somatic (CNV VCIB)**. For more information see “Augment a copy number baseline for use with Ion AmpliSeq™ or Ion AmpliSeq™ HD libraries” on page 297.
 - e. (Optional) Enter information about the baseline into the **Description** field.
 - f. Click **Next**.

6. Complete the **Parameters** step. Accept the default settings or adjust the appropriate parameters, then click **Next**.
7. In the **Samples** step, click an option to view the samples for one of the following categories.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

8. In the **Samples** step, select samples for the baseline. For accurate baselines, it is recommended that you include samples that were analyzed or generated with the same panel or assay.
The minimum number of samples is listed at the top of the screen.
If you do not select the minimum number of samples that are required for the baseline, an error message appears, and you cannot proceed. If needed, you can change the **Minimum number of samples** parameter if you augment baselines.
For BAM and CRAM samples, you cannot view the assay column. You can only see the assay for sequencing samples.
 - a. Select the checkbox next to each sample to use for the baseline.
 - b. Click  in the **Normal Sample** column to designate a selected sample as "Normal" in the baseline.
The tube icon changes to green. A **Normal Sample** is designated with .
If you do not designate the minimum number of samples that are required to be designated as **Normal Sample** for the baseline, an error message appears, and you cannot proceed.
 - c. Click **Next**.
9. In the **Review** step, review the selections, then click **Launch**.
The new copy number baseline is shown to the **Copy Number Baselines** tab. You can monitor the creation of the baseline on this tab.

The new copy number baseline is locked and shown in the **Copy Number Baselines** tab with a **State** of **Completed** and is available for use in the software. Copy number baselines can only be added to assays that support CNV detection.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab, or click **Download Log** to download the log. The log file can help you troubleshoot the failure.

Create a copy number baseline for low-coverage whole-genome libraries

Role—System administrator, organization administrator and manager.

Administrators and managers can create a copy number baseline for use with whole genome libraries with whole genome amplification and low coverage, such as Aneuploidy. This type of copy number baseline is for use with Ion ReproSeq™ PGS Kits.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. In the **Create Copy Number Baseline** screen, select **Low-coverage Whole-Genome**.
The **Create Copy Number Baseline** screen opens to the **Panel** step.
5. Complete the **Panel** step.
 - a. Enter a name in the **Copy Number Baseline Name** field.
The name is limited to 50 characters and no spaces are allowed.
 - b. Select a reference genome.
 - **GRCH38**
 - **hg19**
 - c. Verify that **CNV Informatics Baseline** is the selected **CNV Detection Type**.
 - d. Leave **Augment an Existing Copy Number Baseline as Base** unselected.
Use this option to add samples to baselines that use a **CNV Detection Type** of **CNV Informatics Baseline**. For more information see “Augment a copy number baseline for use with low-coverage whole genome libraries” on page 299.
 - e. *(Optional)* Enter information about the baseline into the **Description** field.
 - f. Click **Next**.
6. In the **Parameters** step, accept the default settings or adjust the appropriate parameters, then click **Next**.
7. In the **Samples** step, click an option to view the samples for one of the following categories.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

8. In the **Samples** step, click an option to view the samples for one of the following categories.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

9. In the **Samples** step, select the required number of samples for the baseline. For accurate baselines, it is recommended that you include samples that were analyzed or generated with the same panels.

If you do not select the minimum number of samples that are required for the baseline, an error message appears, and you cannot proceed. If needed, you can increase the **Minimum number of samples** parameter if you augment baselines that use a **CNV Detection Type** of **Somatic (CNV VCIB)**.

- Select the checkbox next to each sample to use for the baseline.
- Click **Next**.

10. In the **Review** step, review the selections, then click **Launch**.

The new copy number baseline is shown in the **Copy Number Baselines** tab. You can monitor the creation of the baseline in this tab.

The new copy number baseline is locked and shown in the **Copy Number Baselines** tab with a **State** of **Completed** and is available for use in the software. Copy number baselines can only be added to assays that support CNV detection.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab, or click **Download Log** to download the log. The log file can help you troubleshoot the failure.

Copy a copy number baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can copy then edit a copy number baseline. For example, you can start with an existing copy number baseline, then change parameters, or add more samples. A baseline with a **State** of **Completed** can be copied and edited.

If you will add only new samples to an existing baseline, see instead “Augment a copy number baseline for use with Ion AmpliSeq™ or Ion AmpliSeq™ HD libraries” on page 297.

- In the menu bar, click **Assays ▶ Manage Presets**.
- In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
- Place the pointer over the row of the copy number baseline that you want to copy, then click **Copy**.

The **Copy Copy Number Baseline** screen opens to the **Panel** step. The settings and samples that are included can be modified for the new baseline.

4. (Optional) Proceed through the steps in the **Copy Copy Number Baseline** screen and modify settings.

For more information, see “Create a copy number baseline for use with Ion AmpliSeq™ libraries” on page 291 and “Create a copy number baseline for use with Ion AmpliSeq™ HD libraries” on page 293. For whole-genome libraries, see “Create a copy number baseline for low-coverage whole-genome libraries” on page 295.

5. In the **Review** step, review the selections, then click **Save**.

The new copy number baseline is added to the **Copy Number Baselines** tab. You can monitor the creation of the baseline in this tab.

To make the copy number baseline available to select when an assay is created or copied and edited, lock the baseline. Locked copy number with a **State** of **Completed** can be copied and edited. Copy number baselines can only be added to assays that support CNV detection.

Augment a copy number baseline for use with Ion AmpliSeq™ or Ion AmpliSeq™ HD libraries

Role—System administrator, organization administrator and manager.

Administrators and managers can augment an existing copy number baseline that uses a **CNV Detection Type** of **Somatic (CNV VCIB)**. When a baseline is augmented, an existing copy number baseline is copied and saved with a new name. Additional samples can then be added to the new baseline. The existing copy number baseline remains unchanged in the software.

Samples that are added cannot be designated as a **Normal Sample** and should be diverse to help ensure that likely systematic variation is included in the run.

This option is not available for copy number baselines that use a **CNV Detection Type** of **Germline (CNV Informatics Baseline)**.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. Select an option for the copy number baseline.
 - **AmpliSeq**—Copy and augment an existing copy number baseline for use with Ion AmpliSeq™ libraries.
 - **AmpliSeq HD**—Copy and augment an existing copy number baseline for use with Ion AmpliSeq™ HD libraries.
5. Complete the **Panel** step.
 - a. Enter a name in **Copy Number Baseline Name**.
The name is limited to 50 characters and no spaces are allowed.

- b. Select a **Panel** to use with the copy number baseline.
 - c. Select the existing copy number baseline to augment.
 - d. Select **Augment an Existing Copy Number Baseline as Base**. The **CNV Detection Type of Somatic (CNV VCIB)** is automatically selected.
 - e. *(Optional)* Enter information about the baseline into the **Description** field.
 - f. Click **Next**.
6. Complete the **Parameters** step. Accept the default settings or adjust the appropriate parameters, then click **Next**.
A minimum number of samples is required for the baseline. You can adjust the **Minimum number of samples required to add to an existing baseline** parameter in the **Copy Number Baseline Creation** section.
7. *(Optional)* In the **Samples** step, click an option to filter the samples.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

8. In the **Samples** step, select the required number of samples for the baseline. For accurate baselines, it is recommended that you include samples that were analyzed or generated with the same panels.
If you do not select the minimum number of samples that are required for the baseline, an error message appears and you cannot proceed. If needed, you can increase the **Minimum number of samples** parameter if you augment baselines that use a **CNV Detection Type of Somatic (CNV VCIB)**.
 - a. Select the checkbox next to each sample to use for the baseline.
 - b. Click **Next**.
9. In the **Review** step, review the selections, then click **Save**.
The new copy number baseline is added to the **Copy Number Baselines** tab. You can monitor the creation of the baseline in this tab.

Augmented copy number baselines that have a **State of Completed** are available for selection when an assay is created or copied and edited. Copy number baselines can only be added to assays that support CNV detection.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab. The log file can help with troubleshooting if the baseline creation job fails.

Augment a copy number baseline for use with low-coverage whole genome libraries

Role—System administrator, organization administrator and manager.

Administrators and managers can augment an existing copy number baseline for use with whole genome amplification and low coverage libraries, such as aneuploidy. When a baseline is augmented, an existing copy number baseline is copied and saved with a new name. Additional samples can be added to the new baseline. The existing copy number baseline remains unchanged in the software.

When you use the augmented baseline, the new samples that you add can make the baseline more accurate by capturing additional sequencing variability.

IMPORTANT! Do not change the default parameter settings. Augmented copy number baselines for use with whole genome amplification and low coverage libraries must match the parameter settings in the original baseline. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. Select **Low-coverage Whole-Genome**.
5. Complete the **Panel** step.
 - a. Enter a name in the **Copy Number Baseline Name** field.
The name is limited to 50 characters and no spaces are allowed.
 - b. Select a reference genome.
 - **GRCH38**
 - **hg19**
 - c. Select **Augment an Existing Copy Number Baseline as Base**, then select the baseline you would like to edit or augment. The **CNV Detection Type** of **CNV Informatics Baseline** is automatically selected.
 - d. *(Optional)* Enter information about the baseline into the **Description** field.
 - e. Click **Next**.
6. In the **Parameters** step, accept the default settings or adjust the appropriate parameters, then click **Next**.
7. Complete the **Parameters** step. Accept the default settings or adjust the appropriate parameters, then click **Next**.
A minimum number of samples is required for the baseline. You can adjust the **Minimum number of samples required to add to an existing baseline** parameter in the **Copy Number Baseline Creation** section.

8. (Optional) In the **Samples** step, click an option to filter the samples.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

9. In the **Samples** step, select samples for the baseline.

All of the samples that have been run with the selected panel are listed.

- a. Select the checkbox next to each sample to use for the baseline.

- b. Click **Next**.

10. In the **Review** step, review the selections, then click **Save**.

The new copy number baseline is added to the **Copy Number Baselines** tab. You can monitor the creation of the baseline in this tab.

Augmented copy number baselines that have a **State** of **Completed** are available for selection when an assay is created or copied and edited. Copy number baselines can only be added to assays that support CNV detection.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab. The log file can help with troubleshooting if the baseline creation job fails.

Export a copy number baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can export to a ZIP file a copy number baseline that has been created or imported into Genexus™ Software. A copy number baseline can be exported, for example, to use that copy number baseline on another Genexus™ Integrated Sequencer.

System-installed copy number baselines cannot be exported.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline that you want to export, then click **Export**.

The copy number baseline files are downloaded as a ZIP file and are available for import to another sequencer. Copy number baselines can only be added to assays that support CNV detection.

For more information, see “Import a copy number baseline” on page 301.

Import a copy number baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can import a copy number baseline to a Genexus™ Integrated Sequencer.

If you want to transfer a copy number baseline to another Genexus™ Integrated Sequencer, you must first export the copy number baseline from the sequencer that contains the baseline. For more information, see [Export a user-defined copy number baseline \(manager/administrator\)](#).

You cannot import system-installed copy number baselines.

Two types of files are supported.

- H5
- ZIP

For ZIP files, the panel must be available in the software before you import the copy number baseline. For more information, see [“View panel details” on page 327](#).

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click  **Import Copy Number Baseline** in the top right of the screen.

Option	Description
Import a ZIP file	a. In File Type, select ZIP. b. Click Select Copy Number Baseline file. c. Browse for and select the file. d. Click Upload.
Import an H5 file	a. In File Type, select H5. b. In the Panel list, select the panel. c. Click Select Copy Number Baseline file. d. Browse for and select the file. e. Click Upload.

When the file import is complete, a message confirms that the file import was successful.

Imported copy number baselines are available for selection when an assay is created or copied and edited. Copy number baselines can be added only to assays that support CNV detection.

View, add, or remove a note in a copy number baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can add notes to a copy number baseline or view existing notes. Use **Notes** to capture time-stamped written comments and observations for a copy number baseline. A copy number baseline can contain multiple notes from different users.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline of interest, click **Notes**, then review existing notes or add a new note.

Action	Description
View existing notes for a copy number baseline.	In the Add Notes dialog box, notes that are associated with the copy number baseline are listed by user, date, and time.
Add a new note to a copy number baseline.	a. In the Add Notes dialog box, click + Add Note. b. Enter the note, then click Save. The note is added to the copy number baseline and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.
Delete a copy number baseline note	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete) .

4. Click **Close** to return to the **Manage Presets** screen.

Download the log files for copy number baseline creation

Role—System administrator, organization administrator and manager.

Administrators and managers can download and view log files that show the details for copy number baseline creation. The log files are available after the copy number baseline creation job is complete.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of a user-defined copy number baseline, then click **Download Log**.

Delete a copy number baseline

Role—System administrator, organization administrator and manager.

If a copy number baseline fails during job creation, it can be deleted from the software by an administrator or manager.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.

3. Place the pointer over the row of a failed copy number baseline, then click **Delete**.
The **Delete Copy Number Baseline** dialog box opens with the message *Are you sure that you want to delete this Copy Number Baseline?*
4. Click **Yes** to delete the copy number baseline.

The failed copy number baseline is removed from the software and is no longer listed in the **Copy Number Baselines** table.

Remove a copy number baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can make copy number baselines unavailable in the software. You cannot designate as obsolete system-installed copy number baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline that you want to make unavailable, then click **Obsolete**.

The copy number baseline is **Obsolete** in the list of copy number baselines and is no longer available in for use in the software.

Sequence variant baselines (manager/administrator)

Role—System administrator, organization administrator and manager.

A sequence variant baseline is used to reduce the number of false positives for panels that detect somatic variants.

Variant calling filters in Genexus™ Software suppress and remove most sequencing errors. However, it is possible that some errors can pass through the filters and be reported as false positives. In somatic panels, sequencing errors that survive filtering are reported as false positives that require investigation.

A sequence variant baseline is a BED file that contains a list of alleles that are known sequencing errors. The baseline file includes detailed properties for each sequencing error. The errors and properties are based on known germline normal samples. When added to an assay in Genexus™ Software, the baseline is used by the software to filter out sequencing errors that can lead to false positive variant calls and improve variant finding.

Genexus™ Software includes system-installed sequence variant baselines.

You can also copy and edit an existing sequence variant baseline to create a sequence variant baseline.

View sequence variant baseline details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details for sequence variant baselines.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. In the **Sequence Variant Baselines** table, click the name of a sequence variant baseline.
4. Review the details in the **Sequence Variant Baselines** pane.

The details that are shown depend on the setup of the sequence variant baseline. Parameters used to create sequence variant baselines are also shown but are not listed in this table.

Sequence variant baseline details

Item	Description
Created By	Sign-in name of the person who created the baseline.
Created On	The date the baseline was created.
Number of Samples	The number of samples that are included in the baseline.
Amplification Type	Library amplification technology that is used in the baseline (Ion AmpliSeq™ or Ion AmpliSeq™ HD).
Panel Name	The name of the panel that is included in the baseline.
Chip Type	The semiconductor sequencing chip for the panel and baseline.
Genomic Reference	The reference genome for the panel and baseline.

5. (Optional) Click ... (**More Options**) to perform more actions, such as obsolete, export, copy, edit, or audit the sequence variant baseline.

The actions for each baseline are also available in the list of **Sequence Variant Baselines** when you place the pointer over the row of a sequence variant baseline.

Create a sequence variant baseline

Role—System administrator, organization administrator and manager.

Administrators and managers create a sequence variant baseline (SVB) if system-installed sequence variant baselines do not meet research needs. You can add the user-defined baseline to an assay to use as a baseline for a run.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.

3. Click **+ Add New**.
4. In the **Create Sequence Variant Baseline** screen, select the library chemistry for the panel that you want to use for baseline creation.
 - **AmpliSeq**
 - **AmpliSeq HD**
5. Complete the **Panel** step.
 - a. Enter a name in **Sequence Variant Baseline Name**.
The name is limited to 50 characters and no spaces are allowed.
 - b. Select or leave **Augment an Existing Sequence Variant Baseline as Base** unselected.
 - Select **Augment an Existing Sequence Variant Baseline as Base**, then select an SVB from the dropdown list to use the existing sequence variant baseline as a starting point for the new baseline.
 - Leave **Augment an Existing Sequence Variant Baseline as Base** unselected to create a new baseline.
 - c. Select the panel.
 - d. *(Optional)* Enter information about the baseline into the **Description** field.
 - e. Click **Next**.
6. In the **Parameters** step, accept the default settings or adjust the appropriate parameters, then click **Next**.
7. In the **Samples** step, select the required number of DNA, DNA & RNA, or TNA samples for the baseline. For accurate baselines, it is recommended that you include samples that were analyzed or generated with the same panels.
If you do not select the minimum number of samples that are required for the baseline, an error message appears, and you cannot proceed.
 - a. Select the checkbox next to each sample to use for the baseline.
 - b. Click **Next**.
8. In the **Samples** step, select the required number of DNA, DNA & RNA, or TNA samples for the baseline.
The samples that you select must have the same library chemistry, chip type, and flow order.
9. In the **Review** step, review the selections, then click **Launch**.
The new sequence variant baseline is added to the **Sequence Variant Baselines** table.

The new sequence variant baseline is locked and shown in the **Sequence Variant Baselines** tab with a **State of Completed**.

For assays that are compatible with the sequence variant baseline, you can select the sequence variant baseline file in the **Variant Finding** section of the **Parameters** step, in the **Analysis SVB File** dropdown list.

If the **State** of a sequence variant baseline is **Failed**, place the pointer over the row of the sequence variant baseline and click **View Log** to open the log in a separate browser tab. The log file can help with troubleshooting if the baseline creation job fails.

Copy a sequence variant baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can copy then edit a sequence variant baseline. For example, you can start with an existing sequence variant baseline, then change parameters, or add more samples. A user-defined baseline must not be locked and must be in a state of **Completed** before it can be copied. You cannot copy a system-installed or imported sequence variant baseline.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Manage Presets** screen, place the pointer over the row of the sequence variant baseline that you want to copy, then click **Copy**.
The **Copy Sequence Variant Baseline** screen opens to the **Panel** step with the panel and samples predefined. The settings and samples that are included can be modified for the new baseline.
3. Change the software-generated name of the baseline if needed, proceed through the workflow steps, and modify settings.
4. In the **Review** step, review the selections, then click **Launch**.

The new sequence variant baseline is added to the **Sequence Variant Baseline** list. When the **State** of the SVB is **Completed** and the **Status** is **Locked**, the sequence variant baseline is available for selection when an assay is created, or copied and edited.

For assays that are compatible with the sequence variant baseline, you can select the sequence variant baseline file in the **Variant Finding** section of the **Parameters** step, in the **Analysis SVB File** dropdown list. For more information, see Chapter 8, “Create and manage assays”.

Export a sequence variant baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can export sequence variant baselines that have been created, copied and edited, or imported in Genexus™ Software. System-installed sequence variant baselines cannot be exported.

A sequence variant baseline can be exported, for example to use that sequence variant baseline in another Genexus™ Integrated Sequencer.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the sequence variant baseline that you want to export, then click **Export**.

The sequence variant baseline files are downloaded as a ZIP file and are available for import to another sequencer.

For more information, see “Import a copy number baseline” on page 301.

Import a sequence variant baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can import a sequence variant baseline. Two types of files are supported:

- ZIP
- BED

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Click  **Import Sequence Variant Baseline** in the top right of the screen.

Option	Description
Import a ZIP file	a. In File Type, select ZIP. b. Click Select Sequence Variant Baseline file. c. Browse for and select the file. d. Click Upload.
Import a BED file	a. In File Type, select BED. b. Enter a name for the baseline. c. In the Panel list, select the panel. d. Click Select Sequence Variant Baseline file. e. Browse for and select the file. f. Click Upload.

A message confirms that the file import was successful when the file import is complete.

The imported sequence variant baseline is added to the **Sequence Variant Baseline** list. When the **State** of the SVB is **Completed** and the **Status** is **Locked**, the sequence variant baseline is available for selection when an assay is created, or copied and edited.

For assays that are compatible with the sequence variant baseline, you can select the imported sequence variant baseline file in the **Variant Finding** section of the **Parameters** step, in the **Analysis SVB File** dropdown list. For more information, see Chapter 8, “Create and manage assays”.

View, add, or remove a note in a sequence variant baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can view existing notes or add notes to a sequence variant baseline. Use notes to capture time-stamped written comments and observations for a sequence variant baseline. A sequence variant baseline can contain multiple notes from different users.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the sequence variant baseline of interest, click **Notes**, then review existing notes or add a new note.

Action	Description
View existing notes for a sequence variant baseline.	In the Add Notes dialog box, notes that are associated with the sequence variant baseline are listed by user, date, and time.
Add a new note to a sequence variant baseline.	a. In the Add Notes dialog box, click + Add Note. b. Enter the note, then click Save. The note is added to the sequence variant baseline and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.
Delete a sequence variant baseline note	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete) .

4. Click **Close** to return to the **Manage Presets** screen.

Download the log files for sequence variant baseline creation

Role—System administrator, organization administrator and manager.

Administrators and managers can download and view log files that show the details for sequence variant baseline creation. Log files include ERR, TXT and JSON files. The log files are available after the copy number baseline creation job is complete.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of a user-defined custom sequence variant baseline, then click **Download Log**.

Delete a sequence variant baseline

Role—System administrator, organization administrator and manager.

If a user-defined sequence variant baseline fails or is terminated during job creation, an administrator or manager can delete the sequence variant baseline from the software.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of a failed sequence variant baseline, then click **Delete**.
4. In the **Delete Sequence Variant Baseline** dialog box, click **Yes** to confirm the deletion.

The failed or terminated sequence variant baseline is removed from the software and is no longer included in the **Sequence Variant Baselines** table.

Remove a sequence variant baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can make user-defined sequence variant baselines unavailable in the software. You cannot designate as obsolete system-installed sequence variant baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the sequence variant baseline that you want to make unavailable, then click **Obsolete**.

The status of the sequence variant baseline is **Obsolete** in the list of sequence variant baselines and is unavailable for selection during assay creation.

Exon tile assay baselines

Role—System administrator, organization administrator and manager.

An exon tile assay baseline is used to model the amplicon expression variation within a gene. When added to an assay in Genexus™ Software, the exon tile fusion baseline normalizes a test sample, corrects the coverage in exon-tile amplicons, and identifies the presence or absence of expression imbalance in a gene. Exon tile assay baselines that you have created can be incorporated into only some assays. Contact a Field Bioinformatics Specialist (FBS) for guidance.

An exon tile assay baseline is a text file that contains the following items.

- The normalized expression values of exon-tile amplicons for genes with imbalance assays.
- The median normalized expression for each amplicon.

Exon tile assay baselines are associated with a panel BED file in the software.

Genexus™ Software includes predefined exon tile assay baselines.

You can also create an exon tile assay baseline in the software. You can later add the exon tile fusion baseline to an assay and use it as a baseline control for analyses.

An exon tile assay baseline must include at least 40 RNA, DNA & RNA, or TNA samples. Various parameters and thresholds used in the baseline computation determine the inclusion or exclusion of samples in the baseline for each of the genes, for example, ALK, RET, and NTRK1.

View exon tile assay baseline details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details for exon tile assay baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. In the **Exon Tile Assay Baselines**, click the name of an exon tile assay baseline.
4. Review the details in the **Exon Tile Assay Baselines** pane.

The details that are shown depend on the setup of the exon tile assay baseline. Parameters used to create exon tile assay baselines are also shown but are not listed in this table.

Exon Tile Assay Baselines

Item	Description
Created By	Sign-in name of the person who created the baseline. System-installed baselines are included with the software.
Created On	Date on which the baseline was created.
Number of Samples	The total number samples that are included in the baseline.
Amplification Type	Library amplification technology that is used in the baseline. Note: This applies only to Ion AmpliSeq™ or Ion AmpliSeq™ HD.
Panel Name	Name of the panel that is included in the baseline.
Genomic Reference	The file name of the fusion reference for the baseline.

5. (Optional) Click ... **(More Options)** to perform more actions, such as obsolete, export, copy, edit, or audit the exon tile assay baseline. The options that are available depend on whether the exon tile assay baseline is system-installed or is a custom exon tile assay baseline.
The actions for each baseline are also available in the list of **Exon Tile Assay Baselines** when you place the pointer over the row of an exon tile assay baseline.

Create an exon tile assay baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can create exon tile assay baselines for some assays. In some cases, exon tile baselines can be added only to the latest version of an assay. For more information about assay versions, see “Manage software updates” on page 369. Contact a Field Bioinformatics Specialist (FBS) for guidance.

By default, you must use at least 40 RNA, DNA & RNA, or TNA samples to create an exon tile assay baseline.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. In the **Exon Tile Assay Baselines**, click **+ Add New**.
4. In the **Create Exon Tile Assay Baseline** screen, select the library chemistry for the panel that you want to use for baseline creation.
 - **AmpliSeq**
 - **AmpliSeq HD**
5. In the **Panel** step, enter a name for the baseline, select the panel that you want to use, and enter an optional description for the baseline.

Only panels that are compatible with the selected library chemistry and exon tile assay baseline creation are listed. In some cases, exon tile assay baselines can be added only to the latest version of an assay panel.
6. In the **Parameters** step, do not change parameters from the default settings unless you understand how the change can affect the results.

Gene-specific parameters can be enabled with a TXT file.

 - a. Click **Download Example**.
 - b. Open the file with a text editor, enter new values if needed, then save the file with a new file name.
 - c. Click **Upload**, navigate to the file, then click **Open**.
 - d. In the **Gene specific threshold** file dropdown list, select the file to enable gene specific thresholds.
 - e. Click **Next**.
7. In the **Samples** step, click an option to view the samples for one of the following categories.

Option	Description
BAM/CRAM Samples	Shows a list of samples that are created with an uploaded BAM or CRAM file.
Sequencing Samples	Shows a list of samples for which a sequencing run is completed.

8. In the **Samples** step, select the required number of samples for the baseline. For accurate baselines, it is recommended that you include samples that were analyzed or generated with the same panels.

If you do not select the minimum number of samples that are required for the baseline, an error message appears, and you cannot proceed. If needed, you can increase the **Minimum number of samples** parameter if you augment baselines that use a **CNV Detection Type** of **Somatic (CNV VCIB)**.

- a. Select the checkbox next to each sample to use for the baseline.
- b. Click **Next**.

9. In the **Review** step, review the information.

10. Click **Launch**.

The new exon tile assay baseline is listed in the **Exon Tile Assay Baselines** table with a **State** of **Creating**. If there are other active jobs in the software, the **State** is **Pending** until the software starts exon tile assay baseline creation.

When the software completes the process of exon tile assay baseline creation, the sequence variant baseline is locked and shown in the **Exon Tile Assay Baselines** table with a **State** of **Completed**. The exon tile assay baseline is available to add to an assay. The new exon tile assay baseline is available in the **Fusion Baseline** dropdown list in the **Panel** step when you create an assay that contains a fusion panel that is associated with the new exon tile assay baseline. For more information, see “Panel step assay options” on page 240.

View, add, or remove a note in an exon tile assay baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can view existing notes or add notes to an exon tile assay baseline. Use the notes feature to capture time-stamped written comments and observations for an exon tile assay baseline. An exon tile assay baseline can contain multiple notes from different users.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.

3. Place the pointer over the row of the exon tile assay baseline of interest, click **Notes**, then review existing notes or add a new note.

Action	Description
View existing notes for an exon tile assay baseline.	In the Add Notes dialog box, notes that are associated with the exon tile assay baseline are listed by user, date, and time.
Add a new note to an exon tile assay baseline.	a. In the Add Notes dialog box, click + Add Note. b. Enter the note, then click Save. The note is added to the exon tile assay baseline and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.
Delete an exon tile assay baseline note.	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete).

4. Click **Close** to return to the **Manage Presets** screen.

Export an exon tile assay baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can export exon tile assay baselines that have been created in, or imported to Genexus™ Software. System-installed exon tile assay baselines cannot be exported.

An exon tile assay baseline can be exported, for example to add the exon tile assay baseline to an assay for use on a different Genexus™ Integrated Sequencer.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of the locked exon tile assay baseline that you want to export, then click **Export**.

The exon tile assay baseline files are downloaded as a ZIP file and are available to import for use with another sequencer.

For more information, see “Import an exon tile assay baseline” on page 314.

Import an exon tile assay baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can import an exon tile assay baseline. You can import baselines in ZIP format, and in some instances in TXT format. ZIP-format files contain a fusion panel file and a fusion FASTA file. For more information about the option to import an exon tile assay baseline in TXT format, contact a Field Bioinformatics Specialist (FBS).

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Click  **Import Exon Tile Assay Baseline** in the top right of the screen.

Option	Description
Import a ZIP file	a. In File Type, select ZIP. b. Click Select Exon Tile Assay Baseline file. c. Browse your computer and select the file. d. Click Upload.
Import a TXT file	a. In File Type, select TXT. b. In the Panel list, select the panel. c. Click Select Exon Tile Assay Baseline file. d. Browse your computer and select the file. Name is populated based on the file name. e. <i>(Optional)</i> To change the name from the software-generated name, enter a new name for the baseline. f. Click Upload.

When the file import is complete, a message confirms that the file import was successful. Imported exon tile assay baselines are locked by the software.

The imported exon tile assay baseline is available in the **Fusion Baseline** list in the **Panel** step when you create an assay that contains a fusion panel that is associated with the imported exon tile assay baseline. For more information, see “Panel step assay options” on page 240.

Download the log file for exon tile assay baselines

Role—System administrator, organization administrator and manager.

Administrators and managers can download and view a log file that shows the details for exon tile assay baselines creation. The log file is available after the exon tile assay baseline creation job is complete. The log file can help with troubleshooting if the exon tile assay baseline creation fails.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of a user-defined exon tile assay baseline, then click **Download Log**.

Delete an exon tile assay baseline

Role—System administrator, organization administrator and manager.

If an exon tile assay baseline fails or is terminated during job creation, an administrator or a manager can delete an exon tile assay baseline from the software.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. In the **Exon Tile Assay Baselines** table, place the pointer over the row of the draft exon tile assay baseline that you want to delete, then click **Delete**.
4. In the **Delete Exon Tile Assay Baseline** dialog box, click **Yes** to confirm the deletion.

The failed or terminated exon tile assay baseline is removed from the software and is no longer included in the **Exon Tile Assay Baselines** table.

Remove an exon tile assay baseline

Role—System administrator, organization administrator and manager.

Administrators and managers can make user-defined exon tile assay baselines unavailable in the software. You cannot designate as obsolete system-installed exon tile assay baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of the exon tile assay baseline that you want to make unavailable, then click **Obsolete**.

The **Status** of the exon tile assay baseline is **Obsolete** in the list of exon tile assay baselines. Exon tile assay baselines that have been made obsolete cannot be added to an assay when an assay is created or copied and edited.

Report templates

Role—System administrator, organization administrator and manager.

Administrators and managers create and manage report templates that can be used to generate PDF reports in Genexus™ Software. Users can select a report template and generate a report after a sequencing run is complete. You can also set an option in a run plan to automatically generate a report at the end of a sequencing run. When this option is enabled, a default template that is associated with that assay or application category.

A variant report may not contain information about variants. For example, results from immune repertoire assays, PGT-A assays, or microbiome assays do not include information about variants.

Preview a report template

Role—System administrator, organization administrator and manager.

Administrators and managers can view system-installed and user-defined report templates in Genexus™ Software.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** tab, place the pointer over the row of the report template that you want to view, then click **Preview**.
4. (Optional) In the **Report Template Preview** screen, click  **Download** to download a PDF file of the report template. Click  **Print** to print the report template.

Create a report template

Role—System administrator, organization administrator and manager.

Administrators and managers can create report templates for their organization in Genexus™ Software. The templates are used to generate reports in PDF format after a sequencing run. Report templates can be used to automatically generate reports after a sequencing run completes successfully, or manually generated when you view sample results.

As you create a report template, you can reorder sections, and add or remove sections. For more information, see “Sections of a variant report template” on page 321.

IMPORTANT! A user-defined report template must be locked before the template can be used to generate a report.

1. In the menu bar, click **Assays ► Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. Click  **Add New**.

4. In the **Report Template** screen, enter a unique **Report Template Name**, with only 50 or fewer alphanumeric characters (A-Z, a-z, 0-9), and the special characters (period (.), hyphen (-), underscore (_), and space, then complete the remaining sections at the top of the screen.
 - a. In **Application Category** select the application category that you want to associate with the report, or select **Other** for a user-defined application category.

The application category that you select affects the list of templates that are available to manually generate a report from sample results. For example, if you select **Solid Tumor**, only report templates that are associated with the **Solid Tumor** application category are available in sample results for solid tumor assays and samples.
 - b. In **Select Assay**, select the assay that you want to associate with the report template. Only assays associated with the selected application category are listed.

When you associate an assay and application category with a report template, that report template becomes available for use with samples for that application category that are processed with the associated assay.

If you select **None** for the assay, the report template is available to select when you generate a report from a result that was processed with any assay and samples that use the application category selected for the template. That is, a report template that is associated with **Select Assay of None** is available to select for all results when you generate a report.
 - c. (Optional) In **Start From Existing Report Template**, select or change the report template that is used as the basis for the new template.

The available templates are based on the selected application category and assay.

A default report template is selected if a default template is associated with the assay. If no default template is associated with the assay, the system-installed report template for the selected application category is selected.

If the application category that you selected has no default or system-installed template, a template named **Default** is selected.
 - d. In **Languages Setting**, select the language for the report.
5. Enable the **Set as Default** option to set the new report template to be a default report template.

When a report template is set as the default report template, a report is generated automatically after a sequencing run that completes successfully. The default report template can be associated with an application category, an assay, or both application category and assay.

Template association	Selections ^[1]
Default report templates associated with only an application category. Reports are generated automatically each time a sequencing run completes that includes samples with the selected application category.	Ensure that you selected None in the Select Assay dropdown list.
Default report templates associated with an assay. Reports are generated automatically after a sequencing run completes successfully for each report template that is associated with an assay, for run plans based on the assay.	Ensure that you select an assay in the Select Assay list.
Default report templates associated with both an application category and an assay. Reports are generated automatically after a sequencing run completes successfully when both the application category and assay are used together for the samples.	Ensure that you select an assay in the Select Assay list.

^[1] You must select an application category for all report templates.

Tip: You might want to set more than one report default template if, for example, you want to generate reports in multiple languages. You can set up to 3 default report templates that are associated with *both* an application category and an assay, and up to 3 default templates that are associated only with an application category.

Automatically generated reports for each default report template are shown in the **Reports** tab of the sample results table.

6. Disable the **Set as Default** if you do not want the report template to be set as a default template. Report templates that are not set as default, do not automatically generate reports after a sequencing run that completes. These report templates can however be used to manually generate reports after a sequencing run is complete in the **Reports** tab. For more information, see “Generate a variant report” on page 229.
7. Add information to the **Header** section.
 - a. Enter **Report Title** and **Address** of the organization.
 - b. To include a logo on the report, in **Logo**, click **Select File** to browse to the logo file. Acceptable formats are PNG, GIF, JPG, JPEG and TIFF.
 - c. In **Header Fields**, select a term to add to the **Header**.
 - d. Enable or disable the **Report Date** and **Assay Name** checkboxes to show the date that the report was generated and the name of the assay.
8. In the **Configuration** pane on the left, design the template.
 - In **Add Section**, select the section, then click **+ (Add)**.
 - To remove a section, in the box for the section, click **✕ (Remove)**.
 - To add or remove multiple sections, click **Bulk Add**, add or remove sections, then click **Apply**.
 - To rearrange sections of the report template, click-drag the section boxes to rearrange the order.
 - To apply the gene list to the report template, select **Apply Gene List**.

For more information about the sections that can be included in a report, see “Sections of a variant report template” on page 321.

9. Enter and customize the information for each section of the report template.

Some sections can be customized. Sections that are locked and cannot be customized are designated with  (locked).

- a. For each unlocked section of the template, in the **Configuration** pane on the left, click the section name of interest.
- b. In the **Section** pane on the right, add or remove information for the section.
You can click any customizable field to populate a list of available options for the field.

10. (Optional) Add an **Image** section. The image sections allow for the use of images in reports in the following ways.

- Automatically add a sample results image with the template. Use this option if you want to include an image for the sample results that are shown in the software with reports that are automatically generated after a sequencing run.
- Automatically add an image with the template that you select during template creation. Use this option if you want to include an image from your computer in all reports generated from this template, including reports generated for successfully completed sequencing runs, and reports generated manually after a run.
- Allow users who generate reports manually from the template to select one image for each image section that is added to the template. Use this option to give users the flexibility to add images to a report after the sequencing run is complete.

Note: This option requires that an assay be associated with the report template. Ensure that you select an assay in the **Select Assay** dropdown list.

- a. In **Add Section**, select **Image**, then click **+ (Add)**.
The **Image** section is added to the bottom of the **Configuration** pane.
- b. Select the **Image** section in the **Configuration** pane, then do one of the following.

Image details	Steps
Automatically add a sample results image with the template.	To add an image of sample results to an automatically generated report, select the Select Image From Analysis option, then in the Select Image From Analysis dropdown menu, select the name of the image for the sample results. This option is available only when an assay is selected for the template. [1]
Automatically add an image with the template that you select during template creation.	• Select Browse Image. • Browse and select the image that you want to include in all reports generated from the template from your computer.
Allow users who generate reports manually from the template to select one image for each image section that is added to the template.	Select Browse Image , then leave the Browse Image field blank.

[1] The list of images that are available in the dropdown menu depend on the assay that you select in the **Select Assay** option.

Tip: You can add multiple **Image** sections.

11. To preview the report layout, click **Preview Report**.
12. When you are finished, click **Save**.
13. Lock the draft template to make it available to generate reports in the software.
 - a. In the **Manage Presets** screen, click the **Report Templates** tab.
 - b. In the **Report Templates** table, place the pointer over the row of the report template that you want to lock, then click **Lock**.

The new report template is shown in the **Report Templates** list.

Sections of a variant report template

Role—System administrator, organization administrator and manager.

Administrators and managers can create variant report templates in Genexus™ Software. You can design a template to use the sections that provide information that meets laboratory requirements. The sections that are included in the variant report template relate to the application category and assay used in a run.

A variant report template can contain the following sections and information. Some sections can be used multiple times. Some sections can be customized. Some sections are locked and cannot be customized. Locked sections are designated with a  (locked) icon.

Section	Description
Default report sections.	
Header	Header Information is repeated on the top of each page of the PDF variant report. The header includes the title of the report, the organization address, and an optional logo. • In Header Fields, select up to 2 sample attributes to be displayed in the header on each page of the PDF variant report. The sample attribute list is populated based on the application category that is selected at the top of the template. • Select Show to print the report date in the header of the PDF variant report. • Select Show to print the assay name in the header of the PDF variant report.
Footer	Footer information is repeated on the bottom of each page of the PDF variant report. The footer includes the page numbering, regulatory information, and version of the Genexus™ Software. • Select Show to print the report date in the footer of the PDF report. • Select Show to print the electronic signatures in the footer of the PDF report.
Sections that can be used multiple times in various locations in the report.	
Custom Text	Use this section to allow the report user to add custom text to the variant report during report generation. Select the Editable on Report Generation option to allow the text to be edited when the report is manually generated. • Specify the title for the section. • Add a description. • Select Editable on Report Generation to allow the text to be edited when the report is manually generated.
Image	Use this section to add an image to a section in the variant report. The image can be generated from the sequencing run or a static image that you select. You can also allow users to add an image during report generation. The section allows images in PNG, GIF, JPG, TIFF, and JPEG formats. You can add one or more Image section in the template. See step 10 in “Create a report template” on page 316 for instructions.

(continued)

Section	Description
Page Break	Use this section to add one or more page breaks to improve the layout of the variant report in the PDF output.
Variant Details	Use this section to select to include details during report generation about variants that are detected or not detected in the run. - Specify the title for the section. - Choose which variants subsection to include in the variant report (SNVs/Indels, Fusions, CNVs). - In the Fusion subsection, choose the fusion format to use in the variant name columns. By default, the hyphen is used, or you can use a colon. - Add a description and footnote for each variant subsection. - Add and remove columns specific to the variant subsection. - Choose variants calls for each subsection (Present Calls, No Calls, Absent Calls). - Select columns of interest for each variant subsection.
Sections that can be used only one time in the report. The sections are listed here in alphabetical order.	
Complex Biomarkers	Use this section to provide information about complex biomarkers used in only some assays. You can add the following biomarkers: Tumor Mutational Burden, Microsatellite Instability, Genomic Instability, Homologous Recombination Repair, Aneuploidy score, and DNA Mismatch Repair.
Electronic Signatures	Use this section to provide a Report Signed By area that allows electronic signature. The section can include the name of the user who signs the report, the signature type, the date, and comments.
Key Metrics	Use this section to include amplicon information for the results. The following metrics can be added: Targeted Coverage, Amplicon Coverage, and Calculated Cellularity. The metrics that are available to include in this section depend on the assay that is associated with the report template.
Kit Barcodes	Use this section to include details about each barcode for the kit that is used in the run, including sample name, assay name, nucleic acid type, extraction kits and control kits.
Manual Signatures	A section for physical signatures on the printed report. Use this section to add subsections where up to 3 report users can physically add electronic signatures to the report. The section includes the title can include the name of the user who can sign the report, the signature type or title for the signer, the date signed, and comments.

(continued)

Section	Description
QC	Use this section to include quality control metrics in table format. Columns include the Metric Name, Value, Reference Range, and QC Status. The following quality control metrics may be included in the variant report PDF output. • Run QC—A summary of the run quality control metrics • NTC QC – DNA—A summary of the DNA no-template control quality control metrics • NTC QC – RNA—A summary of the RNA no-template control quality control metrics • Purification QC—A summary of QC metrics for the purification run. • Sample QC – RNA—A summary of RNA sample quality control metrics • Sample QC – DNA—A summary of DNA sample quality control metrics • Templating Control QC – CF-1—A summary of the CF-1 quality control metrics
Reagents	Use this section to provide details in table format about run consumables that are used in the run. Details include the consumable name, part numbers, expiration dates, and lot numbers.
Sample Details	Use this section to show the sample information that is entered into the software. • Arrange sample attributes vertically into 1, 2, or 3 columns. • Specify column titles. • Arrange sample attributes (system-defined and user-defined) under the columns. The sample attribute list is populated based on the application category and assay selections. • Select a maximum of up to 8 fields per column. • Add an optional footnote.
Sequencing Run Details	Use this section to show the sequencing run configuration. The following information may be included in the variant report PDF output. • Assay Name—The assay used in the run. • Assay Revision—The assay revision identification. • Completed On—The date and time when the run was completed. • Instrument—The name of the instrument. • Filter Chain—The filter chain applied to results. • Updated Annotations—The name and version of the latest annotation set that is applied to the sample result. • Panel—The panel used in the run. • Started On—The date and time that the run was started. • Operator—The user who sent the run to the instrument. • Serial Number—The serial number of the instrument. • Gene List—The gene list applied to the results.

(continued)

Section	Description
Variants Summary	Use this section to summarize the variants that are detected or are not detected in the run. The following details are included in the Variant details section. • SNVs/Indels—The number of variants for each variant type. • CNVs—The number of variants for each variant type, including Focal CNVs and ARM level CNVs. • RNA Variants—The number of exon skip and exon deletion variants, and intergenic variants for each variant type (fusions). The report includes subsections for Results for Sequence Variations Detected and Results for Sequence Variations Not Detected.
Variant Notes	Use this section to allow users to add notes to the variants that are detected in the run. The variant name and notes are shown in columns. Only the latest note for each variant is shown. • Variant Name and Notes are default columns in this section. • Add up to 5 columns to this section. • The Notes column cannot be removed from this section.

Edit a draft report template

Role—System administrator, organization administrator and manager.

Administrators and managers can edit a variant report for their organization in Genexus™ Software if it is in draft status. A locked report template cannot be edited.

1. In the menu bar, click **Assays ► Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to edit, then click **Edit**.
4. Edit the sections of the report template as desired, then click **Save**.

The report template remains a draft until you lock the report template. For more information, see “Lock a draft report template” on page 325.

Delete a draft report template

Role—System administrator, organization administrator and manager.

Administrators and managers can delete a draft report template from Genexus™ Software.

A locked report template cannot be deleted, but it can be made obsolete.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to delete, then click **Delete**.
4. In the **Delete Report Template** dialog box, click **Yes** to confirm.

Obsolete a locked report template

Role—System administrator, organization administrator and manager.

Administrators and managers can obsolete a locked report template in Genexus™ Software. A report template that has been made obsolete cannot be restored.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to make obsolete, then click **Obsolete**.
4. In the **Obsolete Report Template** dialog box, click **Yes** to confirm.

The draft report template status changes to **Obsolete** and the template is no longer available for use in the software.

Lock a draft report template

Role—System administrator, organization administrator and manager.

When a report template is first created in Genexus™ Software, the status of the report template is **Draft**. Administrators and managers can lock draft report templates to manage the report templates for their organization. After a report template is locked, it cannot be edited or deleted. System-installed report templates are locked by default.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to lock, then click **Lock**.

The report template is locked and cannot be edited or deleted.

View or add notes to a report template

Role—System administrator, organization administrator and manager.

Administrators and managers can view existing notes or add new notes to draft or locked report templates for their organization in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template of interest, then click **Notes**.

Action	Description
View existing notes for a report template.	In the Add Notes dialog box, notes that are associated with the report template are listed by user, date, and time.
Add a new note to a report template.	a. In the Add Notes dialog box, click + Add Note. b. Enter the note, then click Save. The note is added to the report template and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.
Delete a note from a report template	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete) .

4. Click **Close** to return to the list of report templates.

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Panels overview

Role—System administrator, organization administrator and manager.

Administrators and managers can manage panels in the **Panels** tab.

- Add new panel files to Genexus™ Software to use in sequencing runs on the Genexus™ Integrated Sequencer.
- Upload individual files for panels or a ZIP file that contains the panel file and related files. The files can be directly imported from Ion AmpliSeq™ Designer if your computer is connected to the internet, or uploaded from computer if you first import the files to your computer from Ion AmpliSeq™ Designer.
- View panel details, manage hotspot files, target region files, genome reference files, barcode sets, and control fragment files.
- Obsolete and reactivate panels and panel files.

For information about custom Ion AmpliSeq™ Designer panels, see the *Ion AmpliSeq™ & Ion AmpliSeq™ HD Custom Assay User Guide* (Pub. No. [MAN0028005](#)).

View panel details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details in Genexus™ Software about system-installed panels and panels that are imported or uploaded from Ion AmpliSeq™ Designer.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. View the panel details.

Import or add a custom panel

Role—System administrator, organization administrator and manager.

Before you sequence samples on the Genexus™ Integrated Sequencer with a custom Ion AmpliSeq™ or Ion AmpliSeq™ HD panel, you must import or add a ZIP file that contains files for the custom panel, or upload separate custom panel files to Genexus™ Software. Administrators and managers can complete these procedures. If you use a user-defined non-human genome reference file, ensure that you upload the reference FASTA file to the software *before* you import or add the custom panel files, which are contained in a ZIP file, to Genexus™ Software.

Note: RNA gene expression is not currently supported in Genexus™ Software for custom Ion AmpliSeq™ panels.

The first time that you use a custom panel, add the custom panel ZIP file to Genexus™ Software.

- Import the custom panel ZIP file directly from [AmpliSeq.com](https://www.ampliseq.com). For more information, see “Import a custom panel file from Ampliseq.com to Genexus™ Software” on page 329. These custom panels must have a status of "ordered" or "quote requested" on [AmpliSeq.com](https://www.ampliseq.com) to be available for import.
- Add the custom panel ZIP file from the computer. These custom panels can be designed with either a human or a non-human genomic reference.

The custom panel ZIP file that you import directly from [AmpliSeq.com](https://www.ampliseq.com), or add from a folder on the computer, contains the panel target regions (*designed.bed* file) and other files that are related to the custom panel.

- For DNA panels, you can import or add the ZIP file that contains the panel file and can include a hotspots file. To use versions of panel files that are not included in the Ion AmpliSeq™ custom panel, such as a user-defined hotspots (*hotspot.bed*) file, you must upload the files after the genome reference FASTA file and the custom panel files (*designed.bed* or ZIP file) are uploaded or added to Genexus Software.
- For fusion panels, the ZIP file from [AmpliSeq.com](https://www.ampliseq.com) contains the target region file (BED) and the fusion reference file (FASTA). The Ion AmpliSeq™ and Ion AmpliSeq™ HD custom fusion panel files are encrypted, so you cannot visualize or edit the file content or download file content after the fusion reference is uploaded in Genexus™ Software, but the files are used by the software only for analysis. Reverse transcription is included as part of the Genexus™ Software fusion panel workflow during library preparation.

Note: For a DNA and Fusions combined assay template, contact your Field Application Scientist (FAS) or Field Bioinformatics Specialist (FBS).

- To sequence and analyze custom panels that use user-defined genome references that are not hg19 or GRCh38, you can select between two assay types in the software.
 - **Generic Sequencing**, which only performs sequencing and mapping, but not variant calling.
 - Or, **Generic Sequencing + Variant Calling**, which includes both sequencing and mapping, and variant calling.

To use versions of panel files that are not included in the Ion AmpliSeq™ custom panel, such as a user-defined hotspots (*hotspot.bed*) file, you first must upload the files after the genome reference FASTA file and the custom panel files (*designed.bed* or ZIP file) are uploaded or added to Genexus Software.

- For custom panels that use user-defined genome references that are not hg19 and GRCh38, you can obtain the genome reference FASTA file from either of the following sources:
 - Use a reference that is included in the ZIP file from [AmpliSeq.com](https://ampliseq.com), along with a target regions BED file.
 - Use your own genome reference file (for example, if you have uploaded a genome reference file to [AmpliSeq.com](https://ampliseq.com) for a custom panel design). Alternatively, contact your Field Bioinformatics Specialist (FBS) for more information about genome reference files.

Note:

- If the reference FASTA file is in ZIP file format, the file must be extracted before it is uploaded to Genexus™ Software.
- If a user-defined reference file was previously uploaded to Genexus™ Software, the user-defined reference is ready to use and can be selected from the reference dropdown list when a panel is imported or added.

For more information, see “Add user-defined target regions files for custom panels to Genexus™ Software” on page 339.

To improve specificity and sensitivity and to improve the limit of detection, you can create a Sequence Variant Baseline. You can also adjust parameters, such as the Variant Finding parameters, to lower limits of detection (LOD). For more information, see “Create a sequence variant baseline” on page 304.

Import a custom panel file from Ampliseq.com to Genexus™ Software

Role—System administrator, organization administrator and manager.

Before you can import panel files directly into Genexus™ Software, you must have the following

- An account in Genexus™ Software.
 - An access code that enables you to access [AmpliSeq.com](https://ampliseq.com) from Genexus™ Software. You can generate a randomized access code on the website, or create an access code of your choice. You can also use an access code that you generated or created previously.
 - Custom panels with a status of "ordered" or "quote requested" on [AmpliSeq.com](https://ampliseq.com).
1. To generate an access code for [AmpliSeq.com](https://ampliseq.com) do one of the following actions.
Proceed to step 2 to use a previously saved access code for [AmpliSeq.com](https://ampliseq.com).

Option	Action
Generate a random access code on AmpliSeq.com for use in step 4.	- Go to AmpliSeq.com, and sign into your account. - Click My Account ▶ Manage Access Code. - In the Manage Access Code dialog box, click Generate to receive an access code, then click Save. - Click Copy to Clipboard to use the access code in step 4.
Create an access code of your choice for AmpliSeq.com for use in step 4.	- Go to AmpliSeq.com, and sign into your account. - Click My Account ▶ Manage Access Code. - Enter the code that you want to use as your access code into the Access Code field. Select an Access Code Expiration in the dropdown list, then click Save. - Click Copy to Clipboard to copy the access code for use in step 4. If desired, you can remember the access code for future use. Alternatively, you can generate an access code each time that you sign into AmpliSeq.com.

- In Genexus™ Software, in the menu bar, click **Assays** ▶ **Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
- Click  **Import from AmpliSeq**.
- In the **Import from AmpliSeq** dialog box, enter the required information.

Panel Type	Options
DNA and Fusion Panel	IMPORTANT! This option is not supported for custom panel file imports from AmpliSeq.com .
DNA Panel	Reference—Select a reference genome. Library Chemistry—Select AmpliSeq or AmpliSeqHD. AmpliSeq Panels—Select Custom Panel. AmpliSeq Username—Enter your username for AmpliSeq.com. AmpliSeq Access code—Enter the access code that you created previously, or that you copied to the clipboard in step 1.
Fusion Panel	Library Chemistry—Select AmpliSeq or AmpliSeqHD. AmpliSeq Panels—Select Custom Panel. AmpliSeq Username—Enter your username for AmpliSeq.com. AmpliSeq Access code—Enter the access code that you copied to your clipboard, or that you created previously.
RNA Panel	IMPORTANT! This option is not currently supported in Genexus™ Software.

- Click **List My AmpliSeq panels**, then select the custom panel to import.

6. Click **Import**.

Note: Large files take time to import. You can see the file listed in the **Panels** tab only when the import is complete.

After the import is complete, the new panel is available to be associated with a new software assay that you create or copy in Genexus™ Software. Hotspots files for the panel are listed in the **Hotspots** tab and are also available to add to a user-defined assay. For ZIP file imports, the name of the hotspots is the name of the panel followed by **_Hotspot**. Fusion references are listed in the **Panels** tab with more details available in the **Fusion References** tab.

Add user-defined files for custom panels to Genexus™ Software

Role—System administrator, organization administrator and manager.

Administrators and managers can add user-defined panel files individually to the software.

You can upload user-defined files for use with custom Ion AmpliSeq™ and Ion AmpliSeq™ HD panels, including hotspots and target regions files in BED format, and genomic reference files. You can also create or import a sequence variant baseline to improve the specificity of variant calling, which is especially useful when you want to use the software to detect somatic variants so that variants can be sensitively and specifically detected at lower LOD. You must add these files for custom panels to Genexus™ Software *before* you sequence samples on the Genexus™ Integrated Sequencer. When you add the files to Genexus™ Software, the files are available to select when you create or copy the assays to use for the sequencing runs, and in some cases, to help ensure that the files are used correctly by the software.

Export panel files

Role—System administrator, organization administrator and manager.

Administrators and managers can export existing panel files from Genexus™ Software for use on the same or another Genexus™ Integrated Sequencer. For example, you might want to export a panel file to do the following.

- Export a panel file to your computer to edit the panel, then later import the panel from your computer to Genexus™ Software to use in a sequencing run.
 - Move a panel file from one sequencer to another. First export the file to your computer, then import the panel file to another sequencer.
1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
 2. Place the pointer over the row of the panel that you want to export, then click **Export** to download the panel BED file.

Obsolete panels

Role—System administrator, organization administrator and manager.

Administrators and managers can apply a status of obsolete to panels that have been added or imported in Genexus™ Software. A panel file that is obsolete cannot be used in a new assay. An obsolete panel can be used in a sequencing run only if the panel is included in an existing assay.

System-installed panels cannot be designated as obsolete.

1. In the menu bar, click **Assays ► Manage Panels**.
2. In the **Panels** tab, place the pointer over the row of the panel that you want to make unavailable in the software, then click **Obsolete**.

The panel is listed in the **Panels** table with a **Status** of **Obsolete**. Panels that have been made obsolete cannot be added to a new assay. Administrators and managers can reactivate panels that have been made obsolete. For more information, see “Reactivate panels” on page 332.

Reactivate panels

Role—System administrator, organization administrator and manager.

Administrators and managers can reactivate panels that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ► Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Place the pointer over the row of the panel that you want to reactivate, then click **Reactivate**.

The panel is listed in the **Panels** table with a **Status** of **Locked** and is available to add to a new assay.

Manage hotspot files

Role—System administrator, organization administrator and manager.

Genexus™ Software includes system-installed hotspots files as presets to use in assays. User-defined hotspots files can be added when administrators or managers import or add a panel.

System-installed hotspots files are locked and cannot be changed.

Administrators and managers can do the following tasks in the **Hotspots** tab.

- Manage and view details about the system-installed hotspots files.
- Add new hotspot BED files.
- Export the files to a computer.
- Make a hotspots file obsolete.
- Reactivate the obsolete file in the **Assays / Manage Panels** screen.

View hotspot file details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details about system-installed hotspots BED files and hotspots BED files that have been uploaded in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. In the **Hotspots** table, view the details for the hotspot files, including the panel with which the file is associated, whether the file is **Locked**, if the file is **Active**, who created the file, and the date on which the file was created. You can also view the genomic coordinates and alleles that are included in the hotspot file.

Add a new hotspots file

Role—System administrator, organization administrator and manager.

Administrators and managers can upload new a hotspot file in Genexus™ Software.

A hotspots file can also be added when you add or import a panel file.

For more information about custom Ion AmpliSeq™ Designer panels, see the *Ion AmpliSeq™ & Ion AmpliSeq™ HD Custom Assay User Guide* (Pub. No [MAN0028005](#)).

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. Click **+ Add New**.
4. In the **Add Hotspot File** dialog box, enter a name, select a panel, then select the BED file that you want to upload.
5. Click **Upload**.
Large files take time to upload and may not appear immediately.

The hotspot file is listed in the **Hotspot** table and is available to select when you create or copy and edit some user-defined assays.

Export a hotspots file

Role—System administrator, organization administrator and manager.

Administrators and managers can export a hotspots file from Genexus™ Software. For example, you might want to export a hotspots file to do the following.

- Export a hotspots file to your computer to edit the panel, then later import the hotspots file from your computer to Genexus™ Software to use in a sequencing run.
 - Move a hotspots file from one sequencer to another. First export the file to your computer, then import the hotspots file to another Genexus™ Integrated Sequencer.
1. In the menu bar, click **Assays ► Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
 2. Click the **Hotspots** tab.
 3. Place the pointer over the row of the hotspots file that you want to export, then click **Export** to download the hotspots BED file to your computer.

Obsolete a hotspots file

Role—System administrator, organization administrator and manager.

Administrators and managers can apply a status of obsolete to hotspots files that have been added to Genexus™ Software. A hotspots file that is obsolete cannot be used in a new assay. You cannot designate system-installed hotspots files as obsolete. An obsolete hotspots file can be used in a sequencing run only if the hotspots file is included in an existing assay.

1. In the menu bar, click **Assays ► Manage Panels**.
2. In the **Panels** tab, click the **Hotspots** tab.
3. Place the pointer over the row of the hotspots file that you want to make unavailable in the software, then click **Obsolete**.

The hotspot file is listed in the **Hotspots** table with a **Status** of **Obsolete**. Hotspots files that have been made obsolete cannot be added to a new assay. Administrators and managers can reactivate hotspot files that have been made obsolete. For more information, see “Reactivate hotspot files” on page 334.

Reactivate hotspot files

Role—System administrator, organization administrator and manager.

Administrators and managers can reactivate hotspot files that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ► Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. Place the pointer over the row of the hotspot file that you want to reactivate, then click **Reactivate**.

The hotspot file is listed in the **Hotspots** table with a **Status** of **Locked** and is available to add to a new assay. Hotspot files are only available to select for some assays.

Manage fusion reference files (manager/administrator)

Role—System administrator, organization administrator and manager.

Genexus™ Software includes system-installed fusion reference files as presets to use in assays.

System-installed fusion reference files are locked and cannot be changed.

Fusion panels from [AmpliSeq.com](https://www.ampliseq.com) include a custom fusion reference file (FASTA). These fusion reference files must be uploaded as part of the custom Ion AmpliSeq™ panel. For more information, see “Import or add a custom panel” on page 328.

Administrators and managers can do the following tasks in the following tasks in the **Fusion References** tab.

- Manage and view details about fusion reference files.
- Upload new fusion reference files.
- Export the files to a computer.
- Make a fusion reference file obsolete.
- Reactivate the file in the **Assays / Manage Panels** screen.

View fusion reference file details (manager/administrator)

Role—System administrator, organization administrator and manager.

Administrators and managers can view details about system-installed fusion reference files and fusion reference files that have been uploaded Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. In the **Fusion References** table, view the details for the fusion reference files.

Add a new fusion reference file (manager/administrator)

Role—System administrator, organization administrator and manager.

Administrators and managers can upload a new fusion reference file in Genexus™ Software.

For more information, see the *Ion AmpliSeq™ & Ion AmpliSeq™ HD Custom Assay User Guide* (Pub. No. [MAN0028005](#)).

1. In the menu bar, click **Assays ▶ Manage Panels**.
2. In the **Manage Panels** screen, select the **Fusion References** tab.
The **Panels** tab in the **Manage Panels** screen opens.

3. Click the **Fusion References** tab.
4. Click **+ Add New**.
5. In the **Add Fusion Reference** dialog box, enter a name, select a reference genome, then select the FASTA file that you want to upload.
Large files take time to upload and may not appear immediately.
6. Click **Upload**.

The fusion reference file is listed in the **Fusion References** table and is available to select when you create or copy and edit some user-defined assays, or when you add a new panel.

Export a fusion reference file (manager/administrator)

Role—System administrator, organization administrator and manager.

Administrators and managers can export a fusion reference file from Genexus™ Software.

For example, you might want to move a fusion reference file from one sequencer to another. First export the file to your computer, then upload the fusion reference file to another Genexus™ Integrated Sequencer.

1. In the menu bar, click **Assays ► Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. Place the pointer over the row of the fusion reference file that you want to export, then click **Export** to download the fusion reference FASTA file.

Obsolete fusion reference files (manager/administrator)

Role—System administrator, organization administrator and manager.

Administrators and managers can apply a status of obsolete to fusion reference files that have been added to Genexus™ Software. A fusion reference file that is obsolete cannot be used in a new assay. An obsolete fusion reference file can be used in a sequencing run only if the fusion reference file is included in an existing assay.

You cannot designate system-installed fusion reference files as obsolete.

1. In the menu bar, click **Assays ► Manage Panels**.
2. In the **Panels** tab, click the **Fusion References** tab.
3. Place the pointer over the row of the fusion reference file that you want to make unavailable in the software, then click **Obsolete**.

The fusion reference file is listed in the **Fusion References** table with a **Status** of **Obsolete**. Fusion reference files that have been made obsolete cannot be added to a new assay. Administrators and managers can reactivate fusion reference files that have been made obsolete. For more information, see “Reactivate fusion reference files (manager/administrator)” on page 337.

Reactivate fusion reference files (manager/administrator)

Role—System administrator, organization administrator and manager.

Administrators and managers can reactivate fusion reference files that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. Place the pointer over the row of the fusion reference file that you want to reactivate, then click **Reactivate**.

The fusion reference file is listed in the **Fusion References** table with a **Status** of **Locked** and is available to add to a new assay. Fusion reference files are available to select for only some assays.

Manage reference genome files

Role—System administrator, organization administrator and manager.

The Genexus™ Software includes system-installed reference genome files as presets to use in assays. User-defined genome reference files can be added when you import or upload a panel.

System-installed genome reference files are locked and cannot be changed.

Administrators and managers can do the following tasks in the **References** tab.

- Manage and view details about system-installed genome reference files.
- Add new genome reference files.
- Export the files to a computer.
- Make a genome reference file obsolete.
- Reactivate the file in the **Assays / Manage Panels** screen.

View reference genome file details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details about system-installed reference genome files and reference genome files that have been uploaded into Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. In the **References** table, view the details for the reference genome files.

Add a new reference genome file

Role—System administrator, organization administrator and manager.

Administrators and managers can upload a new DNA reference genome file so that the reference genome is available for use in Genexus™ Software. New reference genomes are uploaded as a FASTA file.

For more information, see the *Ion AmpliSeq™ & Ion AmpliSeq™ HD Custom Assay User Guide* (Pub. No. [MAN0028005](#)).

1. In the menu bar, click **Assays ▶ Manage Panels**.
2. In the **Manage Panels** screen, click the **References** tab, then click **+ Add New**.
3. In the **Add Reference Genome** dialog box, enter the required and optional information, then select the FASTA file to upload the reference sequence information.
Large files take time to upload and may not appear immediately.
4. Click **Upload** when you have finished.

The reference genome file is listed in the **References** table and is available to select when you add or import a panel or a fusion reference file.

Obsolete reference genomes

Role—System administrator, organization administrator and manager.

Administrators and managers can apply a status of obsolete to user-defined reference genomes that have been added to Genexus™ Software. A reference genome that is obsolete cannot be used in a new assay. You cannot designate as obsolete system-installed reference genomes.

1. In the menu bar, click **Assays ▶ Manage Panels**.
2. In the **Manage Panels** screen, click the **References** tab.
3. Place the pointer over the row of the reference genome that you want to unavailable in the software from the software, then click **Obsolete**.

The reference genome is listed in the **References** table with a **Status** of **Obsolete**. Reference genomes that have been made obsolete cannot be added to a panel or a new fusion reference file. Administrators and managers can reactivate reference genomes that have been made obsolete. For more information, see “Reactivate reference genomes” on page 339.

Reactivate reference genomes

Role—System administrator, organization administrator and manager.

Administrators and managers can reactivate reference genomes that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. Place the pointer over the row of the reference genome that you want to reactivate, then click **Reactivate**.

The reference genome is listed in the **References** table with a **Status** of **Locked** and is available to add to a new panel or fusion reference.

Add user-defined target regions files for custom panels to Genexus™ Software

Role—System administrator, organization administrator and manager.

Administrators and managers can upload individual target regions files in BED format for use with custom DNA panels. You must add the files for an Ion AmpliSeq™ custom panel to Genexus™ Software before you sequence samples on the Genexus™ Integrated Sequencer.

Use this procedure only to upload reference and target region files separately, for example, to edit a target regions file, or use a version of a file that is not included in the Ion AmpliSeq™ custom panel that you download from [AmpliSeq.com](https://www.ion Torrent.com).

1. In Genexus™ Software, in the menu bar, click **Assays ▶ Manage Panels**.
2. The **Panels** tab in the **Manage Panels** screen, click **+ Add New**.
3. In the **Add Panel** dialog box, enter the following required information.
 - a. Enter the following information.
 - **Name**—Enter a name for the new panel.
 - **Panel Type**—Select **DNA Panel** or **RNA Panel**.

Note: When you select **RNA Panel**, reverse transcription is performed on the Genexus™ Integrated Sequencer as part of the custom panel workflow during library preparation. This option can be used, for example, for viral RNA custom panels.

Reference—Select the reference genome for the panel.

- b. Under **File Type** select **BED File**, then click **Select Target Regions file**.
- c. Find the target regions file to use with the Ion AmpliSeq™ custom panel file on the computer, then click **Open**.

4. Click **Upload**.

Note: Large files take time to upload. You can see the file listed only when the upload is complete.

The new target regions file is listed in the **Panels** tab and is available to add to a user-defined assay in Genexus™ Software.

Manage barcode sets

Role—System administrator, organization administrator and manager.

Genexus™ Software includes system-installed barcode sets as presets to use in assays. User-defined barcode sets can be added when you import or upload a panel.

System-installed barcode sets are locked and cannot be changed.

Administrators and managers can view details about system-installed barcode sets, add new barcode sets, and make a barcode sets obsolete in the **Assays / Manage Panels** screen, in the **Barcode Sets** tab.

For information about system-installed barcode sets, see “System-installed barcode sets” on page 550.

View barcode set details

Role—System administrator, organization administrator and manager.

Administrators and managers can view details for system-installed barcode sets and barcode sets that have been uploaded in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
2. In the **Panels** tab in the **Manage Panels** screen, click the **Barcode Sets** tab.
3. In the **Barcode sets** table, view the list of barcode sets.
4. Click the name of a barcode set in the list to view the barcode IDs, sequences, and other details for the barcode set.
5. Click **✕ (Close)** to return to the complete list of barcode sets.

Add a new barcode set

Role—System administrator, organization administrator and manager.

Administrators and managers can upload a new barcode set in Genexus™ Software. You can upload barcode set files in XLS or XLSX format. An example barcode set file is available in the software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.

3. Click **+ Add New**, enter the required information in the **Add Barcode Set** dialog box, and browse to and select the barcode set file.

Click the **Click here** link to download an example barcode file.

4. Click **Upload**.

The barcode set is listed in the **Barcode Sets** table and is available to select in the reagent step when you create an assay. For more information, see “Reagent step assay options” on page 245.

Obsolete barcode sets

Role—System administrator, organization administrator and manager.

Administrators and managers can apply a status of obsolete to barcode sets that have been added to Genexus™ Software. Barcode sets that are obsolete in Genexus™ Software can no longer be used in the software. You cannot designate system-installed barcode sets as obsolete.

IMPORTANT! You cannot reactivate barcode sets that have been made obsolete.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.
3. Place the pointer over the row of a user-defined barcode set that you want to make unavailable in the software, then click **Obsolete**.
4. In the confirmation dialog box that appears, click **Yes** to confirm that you want to apply the obsolete status the barcode set.

Control fragments

Role—System administrator, organization administrator and manager.

Control fragment sequences are system-installed in Genexus™ Software and are included in reagents. These internal controls are predefined and cannot be modified by users.

Administrators and managers can view control fragment information in the **Assays / Manage Panels** screen, in the **Control Fragments** tab.

Column	Description
Name	The control fragment name.
Sequence	Single-letter nucleotide sequence of the control fragment.
Control	The process in which the control fragment is used.
Status	Indicates whether the control fragment information is locked and cannot be modified by the user.

■ Gene lists	342
■ View details for a gene list	343
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■ Set gene list as default for a cancer type	346
■ Lock a gene list	347
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Gene lists

Role—System administrator, organization administrator and manager.

A gene list is a collection of genes that is used to refine the list of variants shown in sample results in Genexus™ Software.

System-installed gene lists are available in the software, and administrators and managers can create gene lists that meet the research needs.

A gene list is applied in two ways to sample results.

- A gene list can be associated with an assay. When the assay is used in a sequencing run, the gene list will filter for variants of the genes.
- When you view sample results in the software, you can change the gene list that is applied to the sample results, or you can apply a gene list to the sample results for the first time, if no gene list is applied to the sample results. When you apply or change a gene list for sample results that are shown in the software, only variants that are based on the gene list are shown in the sample results. For more information, see “Show variant results based on a filter chain or a gene list” on page 174.

You can create a gene list for a specific type of cancer, or you can create a gene list that is not specific to a type of cancer.

- You can create multiple gene lists for a cancer type that is available in the software, but only one of the gene lists can be set as the default gene list for that cancer type.
- If you have a gene list for a specific cancer type that you want to apply to all sequencing runs that use a specific assay, one gene list for a specific cancer type can be dynamically applied to the sample results based on the cancer type of the sample when you select the **Default Cancer Type Gene List** option in an assay.

Note: When the option **Default Cancer Type Gene List** is set in an assay, the **View Gene List** pane lists this setting as **Default for Cancer Type**.

A gene list without a cancer type will always get applied to all sample results that are generated using the assay in which the gene list is configured.

A record of how each gene list is used, removed, or changed, and by whom, is maintained in the audit trail of a gene list. For more information, see “Review and download audit records for a gene list” on page 349.

View details for a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can view details for a gene list in Genexus™ Software, including the genes that are in the gene list.

Gene lists that include a cancer type can be set as a default gene list with the setting **Set as Default**. A default gene list for a specific cancer type can be dynamically applied to the sample results based on the cancer type of the sample when you select **Default Cancer Type Gene List** option in an assay.

IMPORTANT! If a gene list is updated through software upgrades or through an off-cycle software update, and a cancer type is obsoleted, all system-installed gene lists that include that cancer type will be obsoleted.

A ☆ (**Star**) is shown after the gene list name in the **Manage Gene Lists** screen for gene lists that are designated as default for a cancer type. When the **Default Cancer Type Gene List** option is set in an assay, the **View Gene List** pane lists the default gene list for the designated cancer type as **Default for Cancer Type**.

Note: When the option **Default Cancer Type Gene List** is set in an assay, the **View Gene List** pane lists this setting as **Default for Cancer Type**.

1. Click **Assays ▶ Manage Gene Lists** to open the **Manage Gene Lists** screen.
2. In the **Name** column, click the gene list name of interest.
Details for the gene list are shown. For more information, see “The View Gene List pane” on page 344.
3. When you are finished viewing the gene list details, click **✕ (Close)** to return to the **Manage Gene Lists** screen.

The View Gene List pane

Role—System administrator, organization administrator and manager.

Administrators and managers can select a gene list in the **Manage Gene Lists** screen in Genexus™ Software to view details for the gene list in the **View Gene List** pane.

Option	Description
Name	The name of the gene list.
<i>(Optional)</i> Cancer Type	A value that is shown when the gene list is created for a specific cancer type. Only one Cancer Type can be applied to a gene list.
Content Version ^[1]	The version of a system-installed gene list.
Default for Cancer Type	Gene lists that are set as default for a cancer type in an assay are applied to all sequencing runs that use an assay configured with the setting Default Cancer Type Gene List. A gene list that is designated as default for a cancer type must include a cancer type. A gene list that does not have a cancer type cannot be set as default. Note: When the option Default Cancer Type Gene List is set in an assay, the View Gene List pane lists this setting as Default for Cancer Type.
Created On	The date the gene list was created in the software.
Created By	The person who created the gene list. System-installed gene lists are shown as created by System Installed.
Updated On	The date on which the gene list was updated in the software.
Updated By	The person who last updated the gene list.
Relevant Genes	The genes that are included in the gene list.

^[1] Applies only to system-installed gene lists.

Create a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can create a new gene list in Genexus™ Software.

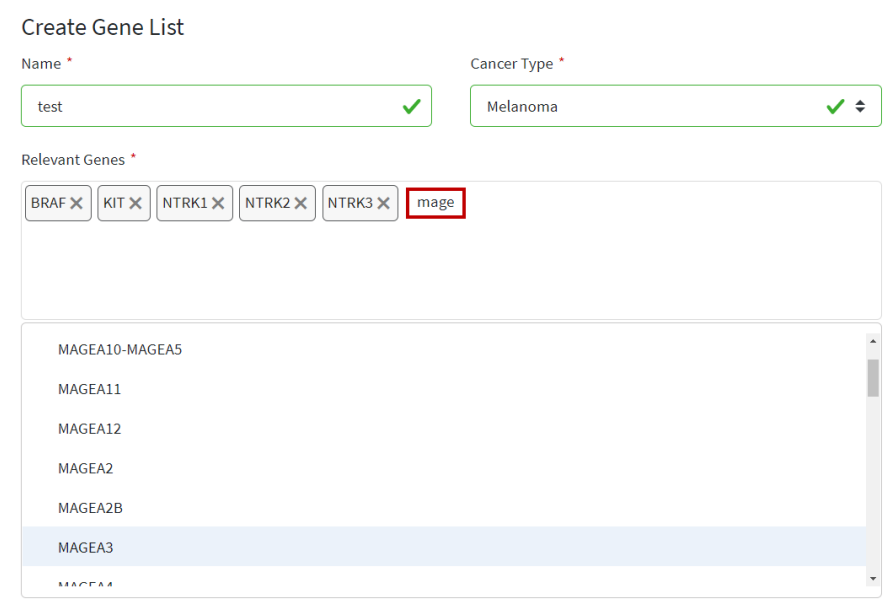
1. Click **Assays ► Manage Gene Lists** to open the **Manage Gene Lists** screen.
2. Click **+ Add New**.
3. In the **Create Gene List** screen, enter a unique gene list name, then complete the remaining sections.
 - a. *(Optional)* In **Cancer Type**, select the cancer type for the new gene list.
A gene list filters sample results that have the **Cancer Type** sample attribute that matches the cancer type that is used by the gene list.

- b. (Optional) To set the new gene list as default for the cancer type, enable the **Set as Default** toggle switch.

If you enable this option, this gene list will be applied to all sequencing runs that use an assay that includes the **Default Cancer Type Gene List** option, which is in the **Panel** step.

Note: When the option **Default Cancer Type Gene List** is set in an assay, the **View Gene List** pane lists this setting as **Default for Cancer Type**.

- c. In the **Relevant Genes** field, add genes to the gene list.

Option	Description
Add a gene to the gene list.	a. Enter a few characters of a gene name to see a list of genes with similar names. If you selected the Cancer Type, option, a list of prepopulated genes is shown, based on the selected cancer type. b. Select a gene from the populated list. For example, if you enter MAGE in the Relevant Genes field, you can select the MAGEA3 gene from the list of genes that is shown. 
(Optional) Remove a gene from the gene list	Click X in the gene box for the gene you want to remove. 

4. Click **Save**.

The new gene list is added to the **Manage Gene Lists** screen, and can be associated with an assay or applied to sample results to refine the list of variants that is shown in sample results.

Edit a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can add or remove the genes that are included in user-defined gene lists in Genexus™ Software. Only user-defined gene lists that are not locked can be edited. System-installed gene lists are always locked and cannot be edited.

1. Click **Assays ► Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to edit, then click **Edit**.
3. In the **Edit Gene List** screen, edit the **Relevant Genes** field.

Option	Description
Add a gene to the gene list.	a. Enter the name of partial name of a gene to see a list of genes with similar names. b. Select a gene from the populated list.
Remove a gene from the gene list.	Click X .

4. Click **Save**.

The new gene list is added to the **Manage Gene Lists** screen, and can be associated with an assay or applied to sample results to refine the list of variants that is shown in sample results.

Set gene list as default for a cancer type

Role—System administrator, organization administrator and manager.

Administrators and managers can set a gene list as default for a cancer type in Genexus™ Software.

For each type of cancer, one gene list can be set as the default gene list. A default gene list for a specific a cancer type can be dynamically applied to the sample results based on the cancer type of the sample when you select **Default Cancer Type Gene List** option in an assay. When the assay is used in a sequencing run, the default gene list filters for variants of the genes.

Gene lists must include a cancer type to be designated as a default gene list.

More than one gene list for a specific cancer type must be available before you can change the default gene list for that cancer type.

1. Click **Assays ► Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to designate as default, then click **Set as default**.
3. Click **Yes** to confirm that you want to change the default gene list.

The gene list is set as default in the **Manage Gene Lists** screen and is applied to sample results from sequencing runs when both of the following are true.

- Sample results include samples with the **Cancer Type** sample attribute that matches the cancer type that is used by the gene list.
- Sample results use an assay that has the gene list option **Default Cancer Type Gene List** selected.

Default gene lists are designated with a ☆ (**Star**) after the gene list name in the **Manage Gene Lists** screen.

Note: When the option **Default Cancer Type Gene List** is set in an assay, the **View Gene List** pane lists this setting as **Default for Cancer Type**.

Lock a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can lock user-defined gene lists in Genexus™ Software. User-defined gene lists that are locked cannot be edited or deleted. System-installed gene lists are always locked.

1. Click **Assays ▶ Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to lock, then click **Lock**.
3. In the **Confirm** dialog box, click **Yes** to lock the selected gene list.

The gene list is locked and cannot be edited or deleted.

Delete a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can delete gene lists that are in draft status in Genexus™ Software. Only gene lists that are not locked can be deleted. System-installed gene lists are always locked and cannot be deleted.

1. Click **Assays ▶ Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to delete, then click **Delete**.
3. In the **Confirm** dialog box, click **Yes** to delete the gene list.

The gene list is removed from the software and is no longer listed in the **Manage Gene Lists** screen.

Obsolete a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can apply a status of obsolete to make a locked user-defined gene list unavailable for use in Genexus™ Software. System-installed gene lists cannot be made obsolete.

Obsolete gene lists are shown in the **Manage Gene Lists** screen with a **Status** of **Obsolete** but are not available for use. You cannot reactivate obsolete gene lists.

You can view details for the gene list and the audit trail for obsolete gene lists.

1. Click **Assays ► Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to remove, then click **Obsolete**.
3. In the **Confirm** dialog box, click **Yes** to obsolete the gene list.

The gene list is listed in the **Manage Gene Lists** screen with the status of **Obsolete** and is not available for use.

Oncology content packages with gene lists

Administrators and managers can use a new or updated gene list from an Oncology content package that is installed in Genexus™ Software.

Oncology content packages can include new or updated gene lists, OncoPrint Variant Annotations rulesets, Variant Naming Services, and cancer type sample attributes. For more information see,

Before you use updated Oncology content updates, you need to download and install the update package. Updated Oncology content packages are available from apps.thermofisher.com.

- If a Thermo Fisher™ Connect Platform account has been configured, download and install software directly from thermofisher.com/connect. See “Download and install software updates from the App Store” on page 370
- Alternatively, if the instrument is not connected to the internet, use a USB drive to install and update the software. See “Format a USB drive for a software update” on page 372 and “Download and install software updates with a USB drive” on page 371.

After an Oncology content package is installed, you can use a new or updated gene list. For more information, see Chapter 11, “Manage gene lists”.

Review and download audit records for a gene list

Role—System administrator, organization administrator and manager.

Administrators and managers can review, export, or print the entire history of a gene list in Genexus™ Software. An audit trail records actions that are made to the selected gene list. Each time-stamped audit record shows the original gene list values and any new values for each action.

The following user actions generate an audit record.

- Create a new gene list.
- Edit a gene list.
- Lock a gene list.
- Delete a gene list.
- Obsolete a gene list.
- Set a gene list as default.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Assays ▶ Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the gene list of interest, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.
4. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the gene list is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attribute mappings Manage Gene Lists screen.

System administration and management

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Role—System administrator, organization administrator.

System administration is performed primarily by the system administrator. An organization administrator can perform some system administration tasks.

System administration in Genexus™ Software

Managers can use tools in the ⚙️ (**Settings**) menu to perform these tasks.

- Create and link user accounts to get software updates on the Thermo Fisher™ Connect Platform on the Thermo Fisher Scientific App Store, and to use Ion Reporter™ Software.
- View system services information and settings.

Administrators can use tools in the  **(Settings)** menu to perform these tasks.

- Design and implement strategies to back up data, monitor disk space usage, and clean up disk space.
- Create organizations for different tasks and for different users.
- Create and manage user account settings and policies, and audit these accounts.
- Create and configure software accounts to get software updates or to upload sample results to other sequencers or software.
- Manage system services and active jobs.
- Manage the retention of log files.
- Perform software updates.
- Configure email settings and electronic signatures, set up laboratory information, and audit these settings.
- View and export audit records.

Linked accounts for use with Genexus™ Software

Role—System administrator, organization administrator and manager.

Administrators and managers can configure software accounts in Genexus™ Software.

Account Type	Description
Thermo Fisher Connect	When a Thermo Fisher™ Connect Platform account is configured and active, system administrators can perform the following tasks in the software. • Download the latest software updates. • Download more plugin software. • Download assay definition files. • Download software configuration packages. The first Connect Platform account that is added in the software is designated as the administrator (Admin) account and cannot be unlinked or removed.
Genexus	Upload sample results to another Genexus™ Integrated Sequencer.
Ion Reporter Cloud	Upload sample results to an Ion Reporter™ Software on Connect account, and then use the Ion Reporter™ Software to further analyze those results.
Ion Reporter Server	Upload sample results to an Ion Reporter™ Server, and then use the Ion Reporter™ Software to further analyze those results.

Create a Connect Platform account

Role—System administrator, organization administrator and manager.

Before you can configure a Thermo Fisher™ Connect Platform account in Genexus™ Software, you must first create a valid Connect Platform account on the <http://apps.thermofisher.com> website.

1. Navigate to <http://apps.thermofisher.com>.
2. Click **Create account**.
3. Enter the information to create a new account.
4. Click **Create account** at the bottom of the screen to complete the registration.

Configure a Connect Platform account with a QR code

Role—System administrator, organization administrator and manager.

Administrators and managers can link their Thermo Fisher™ Connect Platform account in Genexus™ Software with a QR code that is generated in the software.

When a Connect Platform account at <http://apps.thermofisher.com> is configured and active, administrators can perform the following tasks in the Genexus™ Software.

- Download the latest software updates
- Download more plugin software
- Download assay definition files
- Download software configuration packages

Before you link a Connect Platform account to create a **Thermo Fisher Connect** account, you must have a valid Connect Platform account. You must also have the **Thermo Fisher Connect** mobile app to link the account with a QR code that is scanned on a mobile device. For more information, see the Thermo Fisher™ Connect Platform help system.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. Click **+ Link Account to Device**.
The **Link Account to Device** dialog box opens and a QR code image and QR code text are shown.
3. Link the Connect Platform account with the software.

Option	Description
Link a Connect Platform account with the Thermo Fisher Connect mobile app	a. Open the Thermo Fisher Connect mobile app, then sign in. b. In the app, click +. c. Click QR Code, then scan the QR code shown in the Link Account to Device dialog box in Genexus™ Software.
Link the account through the Thermo Fisher™ Connect Platform website	a. Click the link in the QR Code to open the Thermo Fisher Connect website. b. In the Connect Platform website, click  InstrumentConnect in the left sidebar. c. Click Link instrument in the upper right corner of the screen. d. Enter the QR code text shown in the Link Instrument screen. e. Click Link.

The linked account is listed in the **Settings / Thermo Fisher Account** screen. A successfully authenticated account has **Active** listed in the **Status** column.

When the **Thermo Fisher Account** is active, software updates are available in Genexus™ Software. Administrators can download and install the updates. For more information, see “Manage software updates” on page 369.

Unlink a Connect Platform account

Role—System administrator, organization administrator and manager.

Administrators and managers can unlink Thermo Fisher™ Connect Platform accounts in Genexus™ Software.

The first Connect Platform account that is added is designated as the administrator (Admin) account and cannot be unlinked or removed in the software.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. Place the pointer over the row of the account that you want to unlink, then click **Unlink**.

The Connect Platform account is no longer linked in Genexus™ Software.

Register Genexus™ Software accounts

Role—System administrator, organization administrator and manager.

Administrators and managers can register Genexus™ Software accounts. After the accounts are registered, you can transfer results from one Genexus™ Integrated Sequencer to another.

1. In Genexus™ Software, click  (**Settings**) ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, click  **Register Account**.
3. In the **Register Account** dialog box, enter the information that is required to register the account.

Item	Description
Account Type	Select Genexus .
Name	Enter an account name to identify the account in the Thermo Fisher Account Settings screen in Genexus™ Software. The account name can contain only alphanumeric characters (0-9, Aa-Zz), periods (.), underscores (_), or hyphens (-). For example, enter Lab_Admin .
User Name	Enter the user name that is used to sign in to the Genexus™ Software account that you want to link.
Password	Enter the password for the user name for the Genexus™ Software account that you want to link.
Server	Enter the name of the sequencer to use for file uploads.
Port	Enter the port ID of the sequencer to use for file uploads.
Version	Click  Get Analysis Pipeline Version , then select the Genexus™ Software version.
Set as Default Account	(Optional) Click the toggle switch to make this the default account to use to upload results files.

4. Click **Submit**.

The registered account is listed in the **Settings / Thermo Fisher Account** screen. The **Account Type** is **Genexus**. A successfully authenticated account has **Active** listed in the **Status** column.

Register an Ion Reporter™ Software on Connect account

Role—System administrator, organization administrator and manager.

Administrators and managers can register an Ion Reporter™ Software on Connect account in Genexus™ Software.

Ion Reporter™ Software on Connect, the cloud-based platform, is available at <https://ionreporter.thermofisher.com>.

When the account is registered, you can upload results files from Genexus™ Software directly to the Ion Reporter™ Software on Connect and use Ion Reporter™ Software for further data analysis, annotation and reporting. Unmapped BAM files for the sample results are uploaded.

Access to Ion Reporter™ Software on Connect is controlled by your thermofisher.com user name and password. Before you register the Ion Reporter™ Software on Connect account, you must have a valid Connect account on the thermofisher.com website. Users who use the account to access Ion Reporter™ Software on Connect must also have a valid Thermo Fisher account.

1. Click  (**Settings**) ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, click  **Register Account**.
3. In the **Register Account** dialog box, complete the information required to register the account.

Option	Description
Account Type	Select Ion Reporter™ on Connect .
Name	Enter a name to identify the account in the list of Thermo Fisher Account Settings .
User Name	This must be the user name (email address) for a valid Ion Reporter™ Software on Connect account.
Password	Enter the password for the Ion Reporter™ Software on Connect account.
Version	Click  Get Analysis Pipeline Versions , then select the software version.
(Optional) Set as Default Account	Click the toggle switch if you want to make the account the default account that is used to upload results files.

4. Click **Submit**.

The registered Ion Reporter™ Software on Connect account is added to the **Thermo Fisher Account Settings** table. The account can be selected in the **Setup** step of a run plan to automatically upload results files to Ion Reporter™ Software when the run is complete.

Results files can also be uploaded to Ion Reporter™ Software from the **Results / Run Results** screen. For more information, see “Upload results files to Ion Reporter™ Software” on page 196.

Register an Ion Reporter™ Server account

Role—System administrator, organization administrator and manager.

Administrators and managers can register a link to an Ion Reporter™ Server account in Genexus™ Software. When the server is registered, you can upload results files from Genexus™ Software directly to the Ion Reporter™ Server and use Ion Reporter™ Software for further data analysis, annotation and reporting. The uploads contain unmapped BAM files.

1. Click  **(Settings)** ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, click  **Register Account**.
3. In the **Register Account** dialog box, complete the information required to register the account.

Option	Description
Account Type	Select Ion Reporter™ Server
Name	A name to identify the account in the list of Thermo Fisher Account Settings
User Name	This must be a username for a valid Ion Reporter™ Server account.
Password	Enter the password for the Ion Reporter™ Server account.
Server	Enter the name of the Ion Reporter™ Server that you want to use to upload results files.
Version	Click  Get Ion Reporter Software Versions , then select the software version.
Port	Enter the port of the Ion Reporter™ Server.
(Optional) Set as Default Account	Enable the toggle switch to make this account a default account to upload results files.

4. Click **Submit**.

The registered Ion Reporter™ Server account is added to the **Thermo Fisher Account Settings** screen. A successfully authenticated account has **Active** in the **Status** column. The software version for the Ion Reporter™ Software is listed in the **Version** column.

The account can be selected in the **Setup** step of a run plan to automatically upload sample results files and launch analyses in Ion Reporter™ Software. Results files can also be uploaded to Ion Reporter™ Software from the **Results / Run Results** screen. For more information, see “Upload results files to Ion Reporter™ Software” on page 196.

Manage registered accounts

Role—System administrator, organization administrator and manager.

Administrators and managers can edit, unlink, delete, obsolete, and view changes made to the accounts that are registered in Genexus™ Software. Registered accounts can be used to automatically upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer that uses Genexus™ Software 7.0, upload BAM files to another sequencer that uses an earlier version of the software, or automatically upload data for further analysis with Ion Reporter™ Software.

Edit a registered account

Role—System administrator, organization administrator and manager.

Administrators and managers can edit accounts that are registered in Genexus™ Software. Registered accounts can be used to automatically upload results files in BAM or CRAM format to another Genexus™ Integrated Sequencer that uses Genexus™ Software 7.0, upload BAM files to another sequencer that uses an earlier version of the software, or automatically upload data for further analysis with Ion Reporter™ Software. For example, edit a registered account when the password of the account has changed.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to edit, then click **Edit**.
3. In the **Edit Registered Account** dialog box, enter the new information, then click **Submit** to save the changes.

Obsolete a registered account

Role—System administrator, organization administrator and manager.

Administrators and managers can obsolete accounts that are registered in Genexus™ Software. Registered accounts that have been used in a run plan that automatically uploads results files in BAM or CRAM format, or that automatically uploads data for further analysis with Ion Reporter™ Software can be obsoleted. Such accounts cannot be deleted. A registered account that is obsolete can no longer be used.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to obsolete, then click **Obsolete**.

Delete a registered account

Role—System administrator, organization administrator and manager.

Administrators and managers can delete accounts that are registered in Genexus™ Software, but have not yet been used in a run plan that automatically uploads results files in BAM or CRAM format to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to delete, then click **Delete**.
3. In the confirmation message, click **Yes** to delete the account.

The account is removed from the **Thermo Fisher Account Settings** screen.

Review and download audit records for a connected account

Role—System administrator, organization administrator and manager.

Administrators and managers can review, export, or print the entire history of changes that are made to connected accounts in Genexus™ Software. An audit record lists the creation of, and modifications to, the selected connected account. Each time-stamped audit record shows the connected account name associated with the change, and the original settings and any new settings for each action.

Changes to a connected account generates an audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click  **(Settings)** ► **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details of the audited actions.
4. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

Manage organizations

Genexus™ Software provides the ability to create organizations for different application categories, and to assign members to organizations based on their work assignments. An organization determines which application categories are available to members, and gives members access to specific software components, such as assays, presets, and results, which apply to an application. Members do not have access or visibility to other organizations, application categories, or components.

A system administrator can create organizations for the business. A system administrator can assign an organization administrator to an organization, or the system administrator can administer an organization.

After an organization is created, an organization administrator can create members in the organization that they administer. A system administrator can create members in any organization.

Members are created in organizations based on their work assignments and with organization-specific user credentials. Users can be a member of more than one organization, but with different user credentials.

When a member is created in an organization, both the member and the organization administrator receive an email notification that a member is added to their organization.

If you upgrade to Genexus™ Software 7.0, existing users become member of **GNX Org** by default.

Create a new organization

Role—System administrator.

The system administrator can create organizations and can also assign organization administrators. The organization administrator can add new users to their own organization. A system administrator can create users in any organization.

1. In the menu bar, click  **(Settings)** ▶ **Organization Management**.
2. In the **Settings / Organization Management** screen, click  **Create New Organization**.
3. Complete the **Create New Organization** screen.
 - a. In **Organization Name**, type a unique name for the organization.
 - b. In **Application Categories**, select one or more application categories to include in the new organization.
 - c. Click **Save**.

Assay and presets that are appropriate to the application categories are shown.

Assign an organization administrator to an organization

Role—System administrator.

The system administrator creates organizations and can assign organization administrators and grant electronic signature permissions. An organization administrator is not required. The organization administrator can create users and grant electronic signatures to users in their own organization.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **User Management**.
2. In the **User Management** screen, click  **Add New**.
3. In the **Add New User** dialog box, enter the user account information.

User names can contain alphanumeric characters and underscores, periods, and hyphens.
4. From the **Role** list, select **Administrator**.
5. From the **Organization** list, select the organization.
6. (Optional) Grant permissions for this administrator to electronically sign reports. Select an **Electronic Signature Type** to associate with the user account. For example, select **Approved** or **Final Signoff and Lock**.
7. Click **Save**.

The new user account is added to the list in the **User Management** screen. The software sends an email notification containing a temporary password to the email address of the new user.

Edit an organization

Role—System administrator.

The system administrator can edit an organization to add new application categories.

1. In the menu bar, click  **(Settings)** ▶ **Organization Management**.

The **Settings / Organization Management** screen displays a list of all of the organizations in the system.

2. In the **Settings / Organization Management** screen, place the pointer over the row of the organization that you want to edit, then click **Edit**.

3. Complete the **Edit Organization** screen.

- a. In **Application Categories**, select one or more application categories to include in the organization.

You cannot deselect an application category after it has been added to an organization and saved.

- b. Click **Save**.

- c. Click **Yes** to confirm that you want to save the edits to the application categories.

Assay and presets that are appropriate to the application categories are shown.

Review and download audit records for an organization

Role—System administrator, organization administrator.

In Genexus™ Software, administrators can review and download the history of changes that are made to an organization. An audit trail records the actions that are made to the configuration of an organization. Each time-stamped audit record shows the original organization values and new values for each action.

The following user actions generate an audit record for an organization.

- Create the organization
- Edit the organization

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **Organization Management**.
2. In the **Organization Management** screen, place the pointer over the row of an organization, then click **Audit**.
3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details of the audited actions.
4. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

Manage user accounts

Role—System administrator, organization administrator.

Administrators can perform user management actions in the **Settings / User Management** screen.

- Add new user accounts
- Assign user privileges (roles)
- Assign an organization
- Edit user information
- Reset passwords
- Set user account security policies
- View user account audit trails

Add a new user account

Role—System administrator, organization administrator.

Administrators can add new user accounts to the software, assign users to organizations, and grant user permissions to electronically sign reports. A new user receives an email notification to set up a password.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **User Management**.
2. In the **User Management** screen, click  **Add New**.
3. In the **Add New User** dialog box, enter the user account information.
User names can contain alphanumeric characters and underscores, periods, and hyphens.
4. From the **Role** list, select the appropriate user-access level (**Administrator**, **Manager**, **Operator**, or **Reporter**).
5. From the **Organization** list, select an organization.
6. In **Electronic Signature Type**, select a type to grant permissions for a manager, administrator, or operator user to electronically sign reports. For example, **Approved**.
The **Reporter** role has electronic signature permissions enabled by default.
Select **N/A** to remove electronic signature permissions.
7. Click **Save**.
The new account is added to the list in the **User Management** screen. The software sends an email notification containing a temporary password to the email address of the new user.

Edit user details

Role—System administrator, organization administrator.

Administrators can edit user details for users that are in the organization that they administer. Administrators can also suspend or disable user accounts. A history of user account changes is maintained by the audit feature.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  (**Settings**) ▶ **User Management**.
2. In the **User Management** screen, place the pointer over the row of the user account, then click **Edit**.
3. In the **Edit User** dialog box, enter changes in the appropriate account information fields.
 - In the **Role** list, select the user-access level (**Administrator**, **Manager**, **Operator**, or **Reporter**).
 - To grant electronic signature permissions for administrator, manager, or operator users to sign reports, select an **Electronic Signature Type**. For example, **Approved**. Select **N/A** to remove electronic signature permissions.
The **Reporter** role has electronic signature permissions by default.
A  (**Lock**) is shown when you select the **Electronic Signature Type** that is configured to lock a report upon sign off.
 - In the **Status** list, select the appropriate state (**ACTIVE**, **SUSPENDED**, or **DISABLE**).
4. Click **Save** to make the changes.

Changes that are made to the user account are shown in the **User Management** screen, and shown in the **Audit Details** screen for the user.

Change a user password

Role—System administrator, organization administrator.

Periodically, the software sends a system-generated email notification to a user that their password is about to expire. The account holder follows the instructions in the email to change a password. A user can also request that an administrator change their password. Administrators can change a password immediately. After the password is changed, Genexus™ Software sends an email to the user with a system-generated temporary password.

Administrators can change passwords for any user account, except for the system administrator account. The system administrator, and all other users, can change the password for their own user account through their profile in Genexus™ Software.

A history of user password changes is maintained by Genexus™ Software.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. To change a password immediately, do one of the following actions, depending on the account with which you sign in and the type of user account requires a password change.

Sign-in requirements for password change	Action
To change the password for any user account, except for the system administrator account, you must be signed in with an administrator account.	a. Click  (Settings) ▶ User Management, then place the pointer over the row of the user account to change, then click Reset Password. b. In the Change Password dialog box, enter the information in Current Password, New Password, and Confirm Password, then click Change.
To change your own password, including the system administrator account, you must be signed in to the software.	a. Go to  (Profile) ▶ My Profile, then click the Change Password link. b. In the Change Password dialog box, enter the information in Current Password, New Password, and Confirm Password, then click Change.

A confirmation message indicates the password reset.

2. To send an email to a user who requests to have a password changed, click  **(Settings) ▶ User Management**, place the pointer over the row of the account to change, then click **Reset Password**. In the **Change Password** dialog box, click **Send Password in Email**. A confirmation message indicates that the email is sent to the user.

Set user account security policies

Role—System administrator.

A system administrator can set security policies for Genexus™ Software user accounts. User account security policies include the permissible number of failed sign-in attempts, password lifetime, and the length of inactivity before users are automatically signed out of the software. These settings apply to all users in the software across all organizations.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **User Management**.
2. In the **User Management** screen, click  **Policies**.
3. In the **Policies** dialog box, select the appropriate values to set the user-account number of possible sign in attempts and time for attempts, suspension period, and password and session timeout policies.
4. Confirm that the **Enabled** toggle switch is set to enable the session policy settings.
5. Click **Save**.

Review and download audit records for a user account

Role—System administrator, organization administrator.

Administrators can review and download the entire history of changes that are made to user accounts in Genexus™ Software. An audit trail lists the records for the creation of, and modifications to, the selected user account. Each time-stamped audit record shows the user account name that is associated with the change, the original user account settings, and any new user account settings for each action.

The following user actions generate an audit record.

- Add or remove a user account
- Edit a user account
- Reset password

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **User Management**.
2. In the **User Management** screen, place the pointer over the row of the user of interest, then click **Audit**.
3. Review the list of audit records.
4. In the **Audit Details** screen, click a time stamp in the left of the screen to view details of each audited action for the selected user.
5. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

Create an SSH key

Role—System administrator.

The system administrator can add one or more SSH keys in their user profile in Genexus™ Software. SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives the system administrator a secure way to access a remote computer. SSH keys are created on the client systems, and then copied and pasted in Genexus™ Software to add in the authorized keys list.

IMPORTANT! Use only an SSH key that is password protected. The software does not enforce the use of password protected SSH keys, but the use of a password for the SSH keys is strongly recommended.

Do not share the SSH private key or the credentials for the SSH private key. The SSH private key should be used only by the user who creates the SSH private key.

Creating an SSH key is optional, however, best practice is to create one or more SSH keys. SSH keys are needed to access the system at the operating system level to configure some system-level settings, for example mounting an archive directory on the system. More than one SSH key may be needed.

The SSH keys feature is available only to the system administrator. A support representative can also configure SSH keys.

1. Before creating an SSH key, use the Keygen command on your local system to generate keys. When the public and private keys are generated, copy the text string from the public key to use in this procedure.
2. In the menu bar, click  **(Profile)** ▶ **My Profile**.
3. In the **Profile / My Profile** screen, in the **SSH Keys** section, click  **Add SSH Key**.
4. Complete the **Add SSH Key** dialog box.
 - a. In **Label**, type a unique name for this SSH key.
 - b. In **SSH Key**, paste or type the public SSH key string generated by using the Keygen command on your local system.
 - c. Click **Save**.

The SSH key is added to the list in the **SSH Keys** section in the **Profile / My Profile** screen and is available for use.

Configure email notifications

Role—System administrator.

The system administrator configures the software to allow email notifications to be sent to users for events such as to notify users when their account is set up or when their password is expiring, or to notify administrators about backup events. The settings can be edited by a system administrator. An audit record is created for any change.

This procedure can also be done by a support representative.

1. Obtain the SMTP server URL and port information from your IT department.
2. Click  **(Settings) ▶ Configuration**.
3. In the **Configuration** screen, in the **Email Settings** section, enter the STMP server URL and port information supplied by IT, and the Genexus™ Software user name, password, and email address of the system administrator.
4. Click **Update**.
5. Click **Send Test Email** to send a test email to the logged in user.
If the test message arrives successfully, email notifications are enabled for the software.

Configure laboratory information

Role—System administrator.

The system administrator configures contact information for the laboratory. This information provides points of contact for support requests or if a problem occurs with the connected Genexus™ Integrated Sequencer.

IMPORTANT! Administrators should review the contact information periodically to ensure that it is current and accurate.

1. Click  **(Settings) ▶ Configuration**.
2. Complete the **Lab Information** section.
 - **Lab Contact**—Enter the name, email address, and phone number of the person in your organization who should be notified during a support request of problems related to the sequencer.
 - **IT Contact**—Enter the name, email address, and phone number of the person in your organization who should be notified during a support request or for problems related to the sequencer or the network environment.

Edit laboratory information (administrator)

Role—System administrator.

The system administrator can edit the laboratory information in Genexus™ Software. The changes are maintained in the audit records. For more information, see “Review and download audit records for laboratory contact information (administrator)” on page 367.

1. Click  **(Settings)** ▶ **Configuration**.
2. In the **Lab Information** section, below the **Lab Contact** or **IT Contact** pane, click **Edit**.
The fields for the **Lab Contact** or **IT Contact** are now available to edit.
3. Enter the updated contact information.
4. Click **Save** to save the new selections or click **Reset** to revert to the original information.

Review and download audit records for laboratory contact information (administrator)

Role—System administrator.

The system administrator reviews and downloads the history of changes made to the laboratory contact information in Genexus™ Software. Audit records show changes made for each laboratory contact or IT contact.

1. Click  **(Settings)** ▶ **Configuration**.
2. In the **Configuration** screen, under **Lab Information**, next to the contact type, click **Audit**.
 - **Lab Contact**
 - **IT Contact**

A list of time-stamped audit records that includes information for each change to the **Lab Contact** or **IT Contact** is shown in the **Audit Details** screen.

3. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.
4. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the sample is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the Configuration screen.

Configure and manage electronic signatures

Role—System administrator.

The system administrator can configure and manage electronic signatures in Genexus™ Software. Electronic signatures are used to sign off variant reports.

Electronic signature types designate how an electronic signature is used. Different signature types, such as review, approve, and lock, can be required to sign off on a variant report. Designate one of the signature types to lock the report during sign-off.

Administrators can add new electronic signature types. A user account can be associated with just one type. Electronic signature types can be renamed, deleted, made obsolete, or restored. Changes to electronic signature types are shown in the audit trail for the signature type.

Electronic signature types and settings are available to all organizations.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click  **(Settings)** ► **Configuration**.
2. In the **Configuration** screen, in the **Electronic Signature** section, manage signatures.

Option	Description
Add new electronic signature types.	In the Bulk add list of comma delimited meaning of signature field, enter new signature types, separated by commas. For example, enter <i>Review,Inconclusive</i> to add two new signature types. Signature types that are added can be associated with users who are granted permission to electrically sign reports. For more information, see “Add a new user account” on page 361 and “Edit user details” on page 362.
Designate a signature type to lock a report.	In the Select a type of signature to lock report when signing off section, select the signature type to lock a report during report sign-off. For example, create a signature type of <i>Final sign off and lock</i> to lock a report. A report is locked when a user with that signature type electronically signs the report. Only one electronic signature type can lock a report.
Review the history of the electronic signature type.	Click  to view a list of time-stamped audit records that includes information for each action performed on the assay is shown in the Audit Details screen.
Rename a user-defined electronic signature type.	Click  Edit , enter a new name for the signature type, then click Save .
Delete a user-defined electronic signature type.	Click  Delete to delete a signature type. A user-defined electronic signature type can be deleted if is not associated with a user, is not set as lock-level, and is not used in electronic sign-offs for any reports. System-installed signature types cannot be deleted.

(continued)

Option	Description
Obsolete a user-defined electronic signature type.	Click  Obsolete to remove the signature type as an option during variant report sign-off. A signature type that has been used to sign off a report cannot be deleted but can be made obsolete. If an obsolete signature type is associated with a user account, an alert is added to the User Management screen next to Electronic Signature Type . An alert is also shown if no signature type is associated with a user account that includes permissions for electronic signatures. The signature type must be edited by an administrator before the user can electronically sign reports. For more information, see “Edit user details” on page 362.
Restore a user-defined electronic signature type.	Click  Reactivate to reactivate a signature type that has been made obsolete.

Manage software updates

Role—System administrator.

The system administrator can update Genexus™ Software. When software updates are available for download from the Connect Platform, users with a Thermo Fisher™ Connect Platform account see  **(Notifications)** in the Genexus™ Software banner on the top of the screen.

When an update of any software package is available to install, system administrators can view the available versions for assay definition files, plugins, configuration packages, and software updates in the **Software Updates** table in **Settings / Software Updates** screen.

- Assays that are available to install are listed in the **Category of Assay Definition Files** in the **Software Updates** table. The software files that contain the assay settings and parameters are packaged in a ZIP file called the assay definition file (ADF). You can install and use multiple versions of assays in the software.
- The currently installed software versions for each software module, assay definition files, plugin, configuration package or security patch update that is installed are shown when the **Installed Software** link is selected.
- New software updates that are available to be downloaded and installed are shown when the **Software Updates** link is selected.
- Security patch updates and security configuration updates for Genexus™ Software do not include any software features. These update packages are identified by fourth digit in the software version number. For example, Genexus™ Software x.x.x.1.

After the system update is installed, entries are logged in the installation history and audit history in Genexus™ Software.

Updated software packages are available from apps.thermofisher.com.

- If a Connect Platform account has been configured, download and install software directly from thermofisher.com/connect. See “Download and install software updates from the App Store” on page 370
- Alternatively, if the instrument is not connected to the internet, use a USB drive to install and update the software. See “Format a USB drive for a software update” on page 372 and “Download and install software updates with a USB drive” on page 371.

Download and install software updates from the App Store

Role—System administrator.

The system administrator can download and install software updates from the App Store. Before you download and install software updates from the Connect Platform on the Thermo Fisher Scientific App Store at thermofisher.com/connect, confirm these conditions.

- Configure a Connect Platform account. For instructions, see “Configure a Connect Platform account with a QR code” on page 352.
- Connect the Genexus™ Integrated Sequencer to the internet.
- Ensure that the Genexus™ Integrated Sequencer has the latest software version installed.
- Ensure that the sequencer is idle, no runs are in progress or are queued to run, and the main menu screen is shown on the instrument.

When software updates are downloaded to a computer from the Thermo Fisher App Store on Connect, the checksum information for the downloaded software updates are verified by Genexus™ Software before the software package is installed.

1. In the menu bar, click  (**Settings**) ▶ **Software Updates**, then click **Software Updates** in the upper right corner.

The **Software Updates** screen opens with **App Store** option selected.

2. (Optional) Click the desired package name, view the installation details and version information in the **View Package Details** dialog box, then click **Close**.

3. Click **Download** in the **Actions** column in the row of the software package of interest.
After the download is complete, the **Download** link changes to an **Install** link.

4. Click **Install**, then click **Yes** in the confirmation window.

Depending on the software package that you are installing, a command prompt window may open to show the progress of the installation. Some software packages take a long time to install.

5. When installation of the software package is complete, view the **Installed Software** screen to verify the installation.

Download and install software updates with a USB drive

Role—System administrator.

If the Genexus™ Integrated Sequencer is not able to connect to the Thermo Fisher Scientific App Store, you can download software updates onto a desktop computer, transfer the updates to a USB drive, then connect the USB drive to the sequencer to perform the software updates. The USB drive must be formatted properly.

To download and install software updates from the App Store, confirm these conditions.

- Create or access a Thermo Fisher™ Connect Platform account. For instructions, see “Configure a Connect Platform account with a QR code” on page 352.
 - Ensure that the sequencer is idle, no runs are in progress or are queued to run, and the main menu screen is shown on the instrument.
1. Format a USB drive for the software update.
Follow the instructions in “Format a USB drive for a software update” on page 372.
 2. Insert the USB drive in computer.
 3. Insert the USB drive in your computer, then sign in to the Thermo Fisher Scientific App Store on Connect at apps.thermofisher.com.
 4. On the left side of the App Store screen, click  (**AppConnect**).
A list of assets is shown.
 5. On the right side of the screen, under **Resource Libraries**, click **Ion Torrent™ Genexus™**.
 6. Click the **Software Update** filter option.
A list of software packages is shown.
 7. In the Ion Torrent™ Genexus™ Software table, select the row that contains the software package of the version that you want to install.
 8. Click  (**Download**) to download the package in a ZIP file.
 9. Copy the downloaded package to the `/media/usbinstall` folder in the USB drive.
 10. After completion, unmount the USB drive, then insert the drive into one of the USB ports at the back of the sequencer.
 11. In the menu bar of Genexus™ Software, click  (**Settings**) ▶ **Software Updates**, then click **Software Updates** in the upper right corner.
 12. Select **USB**.
 13. On the software package that you want to install, click **Install**, then click **Yes** to confirm.

When installation of the software package is complete, click **Installed Software** to verify the installation.

Format a USB drive for a software update

Prerequisites:

- A desktop computer with disk utility software that supports Ext3 file system format.
- A USB drive with sufficient storage capacity for the software update. A capacity of 128 GB or more is recommended.

IMPORTANT! Ensure that the USB drive does not contain any viruses or malware before you insert the USB drive into the Genexus™ Integrated Sequencer.

1. Insert the USB drive into the computer.
2. On the computer, open the disk utility software by navigating to **System ▶ Administration ▶ Disk Utility**.
3. If the USB drive has partitions, delete the partitions.
 - a. Under **Peripheral Devices from Storage Devices**, click the **USB Flash Drive (SMI USB DISK)** to select the USB drive.
 - b. Highlight the volumes (New Volume FAT >60 GB).
 - c. Click **Unmount Volume**, then click **Delete Partition**.
 - d. Click **Delete** to confirm.
4. Select the free USB volume, then click **Create Partition**.
The **Create partition on SMI USB DISK** window opens.
5. Deselect the **Take ownership of file system** checkbox.
6. Select **ext3** from the **Type** menu, then select the entire size of the partition. Name the partition **usbinstall**.
The partition name must be all lowercase.
7. Click **Mount**.
The mount point of the USB drive is `/media/usbinstall`.

Proceed to “Download and install software updates with a USB drive”.

Review and download audit records for a software update

Role—System administrator.

The system administrator can review and download the entire history of a software update in Genexus™ Software. An audit trail lists the records of actions related to software update packages. Each time-stamped audit record shows changes to software packages that were made with the update, including version numbers, organization names, date of the update, and any changes to the assets, such as, software components.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **Audit Records**.
2. In the **Audit Records** screen, select **Software Update** in the data object list, then click **Search**.
3. In the **Audit Records** screen, in the **Record** column for the software update of interest, click **Audit**.
4. Review the list of audit records.
5. In the **Audit Details** screen, click a time stamp in the left of the screen to review details for each audited action.
6. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

Manage backups

Role—System administrator.

The system administrator can perform these backup tasks in Genexus™ Software.

- Create strategies for backing up software data (**System Data**) and for backing up the sequencing data and run and sample results (**Analysis Results**).
- Mount one or more backup drive locations for network or cloud storage to fulfill backup strategies.
- Assign one or more organizations to specific backup drives.
- Assign one or more organization administrators to specific backup drives.
- Monitor backup drive space usage.
- Monitor sequencer disk space usage.
- Configure backup schedules for backing up the software.
- Manage drive cleanup settings.
- Configure settings for email notifications about backup events.
- View and download the audit trails of backup events.

Backup overview

Role—System administrator.

IMPORTANT! The first backup procedure or a new backup procedure takes a considerable amount of time. The amount of time depends on the number of runs to back up and the amount of disk space on the Genexus™ Integrated Sequencer. For more information, see “Disk space usage example” on page 387.

A sequencer restart also takes time. If a backup procedure is in progress when the sequencer restarts, the backup procedure resumes where it left off.

The following types of data are backed up daily at midnight and cannot be changed.

- **System Data**—Includes the Genexus™ Software—The database, reference files, assay definition files, annotation content, and custom and system-installed panel files, hotspot files, fusion reference files, and software presets.
- **Analysis Results**—Includes the final run results that are generated after sequencing. Run results include run details, BAM and CRAM files, filtered results, graphs, run reports, metrics, plugin results, annotated variant data, and VCF files.

Additional files are backed up daily or weekly based on backup settings. Additional files include 1.wells files and other intermediate files.

- The first backup drive that is mounted is the default drive. The default drive is the first drive shown in the backup directory list in the **Backup and Restore** screen.
- In previous versions of Genexus™ Software, only one mount drive was used for backup and system restore procedures. For an upgrade to Genexus™ Software 7.0, that single drive is the default drive that stores all backup data. After an upgrade, you can add additional drives and designate any drive as the default backup drive.
- You can have more than one backup drive in Genexus™ Software 7.0. Data from sequencing runs, and additional files can be assigned to different organizations, and can be backed up on separate drives.
- If only one backup drive is used, software system data (**System Data**), sample results (**Analysis Results**), and additional files are stored together on that drive. The drive must have the capacity to store all of this data.
- Software system data are backed up on the default drive. The system software data are the content necessary for essential system function and usage. This includes the software database, reference files, assays, annotation sources, templates, and plugins. Data that is specific to user-defined assays and any other system-installed objects are also backed up on the default drive. The data that is backed up on the default drive is used if the software needs to be restored or reimaged.
- Sample results (**Analysis Results**) and additional files for runs can be backed up on the default backup drive, or on additional backup drives. Sample results include run details, BAM and CRAM files, run reports, and plugin results.

- Each organization can have its own backup drive. The administrator for that organization can specify whether to back up additional files. Data for other organizations is not backed up on that drive.

IMPORTANT! Data for organizations that are not associated with a backup drive is not backed up. You must associate each organization with a backup drive and set up the drive for automatic backup procedures, or back up the data manually.

- If you change an additional drive to become the default drive, the next backup procedure puts the software data on the new default drive. In that case, there is more than one copy of the software data. However, a software restore procedure uses the data from the new default drive.
- WORM (Write Once Read Many) policies are supported for backups and restore procedures of data and run archives to Network Attached Storage (NAS) devices and to Amazon™ Web Services (AWS) directories. For more information, talk to your internal IT team.

During the restoration of data or archives, the following occurs.

- The software always restores the latest copy of the data.
- The software adds version numbers to the archived data on the storage device.
- The versions are displayed in parenthesis in the run name column on the **Restore Run** screen.
- The software always creates a copy of the backup and tries deleting the older copy.
- When the WORM policy is not enabled, the software overwrites the older version.
When the WORM policy is enabled, the software does not overwrite the older version and makes a new additional updated version of the backup.

Consult a local Field Service Engineer (FSE) if you need help to set up a backup system. For more information, see “Customer and technical support” on page 591.

Add a network backup drive

Role—System administrator.

The system administrator can use a Network File System (NFS) to mount one or more external storage devices to be used by Genexus™ Software as a backup drive.

The first mount that is added is the default backup drive. Only one default backup drive is allowed. The default backup drive stores the software system data.

Sample results and additional files are specific to organizations and are in organization-specific folders on the backup drive. An organization can be assigned to the default drive, or to a different drive.

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux operating system external storage devices for Genexus™ Software.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, in the **Backup Settings** section, click **Add Mount**.
3. Complete the **Add Mount** dialog box.
 - a. For **Connection type**, select **Network**.

- b. In **Mount Point directory**, enter a unique name of the backup drive.
Do not include information from a host name in the backup drive name. For example, if the host name is **gnx001**, do not use **gnx001** in the backup drive name.
- c. In **Network Storage Source**, enter the path to the storage device.
- d. (Optional) Select **Enable Encryption** to help maintain data privacy and integrity.
- e. (Optional) Select **Automount** to allow backup drives to be mounted when the sequencer is restarted.
When the option is not selected, you must manually mount a backup drive after the sequencer is restarted.
- f. (Optional) If this is the default backup drive, select **Set as Default Backup Directory**.
- g. Click **Save**.

4.

The drive is added to the **Backup Directory** list.

Manually configure a backup server

Role—System administrator.

A system administrator can manually configure an external storage device to be used as a backup server to store software data (**System Data**) and sample results (**Analysis Results**).

Alternatively, a system administrator can set up a backup server using Genexus™ Software. For more information, see “Add a network backup drive” on page 375.

IMPORTANT! We recommend Network File System (NFS) version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices.

1. Use a command-line tool to mount the NFS to the Genexus™ Integrated Sequencer.
The following example is for users who have an existing NFS with read and write access. Mount the NFS to the sequencer using the **fstab** file so that the mount point is persistent (NFS mounts automatically after rebooting).

Example:

- For NFS share: `cqtsop4:/md1200`
- Mount point: `nfs1`

2. Create a mount point to mount the external device on nfs share.
 - a. Create the directory `/nfs1` using the following command:


```
sudo mkdir /nfs1
```

- b. Append the mount point entry in `/etc/fstab`.

Command example:

```
sudo vi /etc/fstab
cqtsop4:/md1200 /nfs1 nfs defaults 0 0
```

- c. Save, then exit by pressing **Esc** on your keyboard, then type **:wq**

3. Create a permanent or temporary mount on nfs share:

- Permanent mount command: `sudo mount -a`
- Temporary mount command (example): `sudo mount cqtsop4:/md1200 /nfs1`

4. Verify that the NFS share is mounted using the `df -kh` command.

```
ionadmin@eternity1:~$ df -kh
Filesystem Size Used Avail Use% Mounted on
/dev/sda9 253G 5.9G 235G 3% /
none 4.0K 0 4.0K 0% /sys/fs/cgroup
udev 63G 4.0K 63G 1% /dev
tmpfs 13G 1.3M 13G 1% /run
none 5.0M 0 5.0M 0% /run/lock
none 63G 0 63G 0% /run/shm
none 100M 0 100M 0% /run/user
/dev/sda5 19G 2.9G 15G 17% /var
/dev/sda1 938M 37M 854M 5% /boot
/dev/sda6 9.1G 1.7G 7.0G 20% /home
/dev/mapper/raid-serverdata 8.4T 4.3T 4.1T 52% /serverdata
/dev/mapper/raid-servershare 1.0T 9.0G 1015G 1% /servershare
/dev/mapper/raid-scratch 1.0T 838M 1023G 1% /scratch
/dev/sda7 92G 61M 87G 1% /tmp
pluto:/md1200 37T 1.1T 36T 3% /nfs2
penguin-ts1:/local 11T 9.3T 978G 91% /nfs
ddn4.itw:/gs1/ion-data/data 1.1P 1001T 115T 90% /ddn
cqtsop4:/md1200 37T 37T 77G 100% /nfs1
```

The mounted directory is listed in the **Backup Directory** dropdown list in the **Backup & Restore** screen.

For more information, see help.ubuntu.com/its/serverguide/network-file-system-configuration.

Add a cloud backup drive

Role—System administrator.

The system administrator can mount a cloud backup directory to use as a backup drive.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, in the **Backup Settings** section, click **Add Mount**.
3. Complete the **Add Mount** dialog box.
 - a. For **Connection type**, select **Cloud**.
 - b. Enter a unique name of the **Mount Point directory**.
 - c. Select the **Cloud Provider**.
 - **AWS-S3**—Amazon Web Services™ (AWS) is a comprehensive and broadly adopted cloud storage.
 - **Azure**—Azure™ is a specific brand of cloud computing platform that helps organizations build, deploy, and manage applications and services. Azure™ uses ID drives (transient capacity), and page blobs VM-based volumes are stored in block storage. Object storage uses square blobs and files.,
 - **Google Cloud**—Google™ Cloud™ is a public cloud vendor.
 - d. Enter the **Configuration** information for the selected cloud provider.
Information must be entered correctly to help ensure that the mount works properly.
Information is to be provided by an administrator for the network.
 - e. (Optional) Select **Enable Encryption**.
 - f. (Optional) Select **Automount** to allow backup drives to be mounted automatically when the sequencer is restarted.
When this option is unselected, you need to manually mount a backup drive after the sequencer is restarted.
 - g. (Optional) If this is the default backup drive, select **Set as Default Backup Directory**.
The first time you mount a cloud backup directory, the option **Set as Default Backup Directory** is disabled, and the mount is automatically set as the default backup drive. The option is available when you set up subsequent backup drives.
The first mount added is the default backup drive. Only one default backup drive is allowed. The default backup drive stores the Genexus™ Software data. All other backup drives store sample results and additional files.
 - h. Click **Save**.

The drive is added to the **Backup Directory** list.

Add an organization to a backup directory

Role—System administrator.

After a backup directory is mounted, the system administrator can assign one or more organizations to a backup directory in Genexus™ Software.

Each organization can have its own backup directory, or organizations can share a backup directory.

Run data for the assigned organization is backed up to the assigned backup directory. Run data consists of run results, sample results (**Analysis Results**), and intermediate files or sequencing output files. If an organization is not assigned to a backup directory, the data for that organization is not backed up.

After one or more organizations is assigned to a directory, the organization administrator can specify what type of run files are backed up and when the files should be deleted from the server and no longer backed up. For example, the administrator decides whether to back up sequencing output data or intermediate files, or both, and how long to keep those files on the server before permanently deleting them.

IMPORTANT! Data for organizations that are not associated with a backup drive is not backed up. You must associate each organization with a backup drive and set up the drive for automatic backup procedures, or back up the data manually.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, in the **Backup Settings** section, click the **Backup Directory** name, then click  **Edit**.
3. In the **Configure Organization for Mount** dialog box, in the **Associate Organization(s)** box, select one or more organizations from the list.
4. (Optional) In the **Include Additional Files for Individual Org** section, select the files that should be backed up for the assigned organizations—**Sequencing Output** or **Intermediate Files** or both.
5. Click **Save**.

Start a manual backup

Role—System administrator.

After a new backup drive is mounted, the system administrator can manually start a backup procedure in Genexus™ Software instead of waiting for the nightly backup. The first backup process to a new drive can take a considerable amount of time.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, ensure that the **Backup & Cleanup Enabled** toggle switch is turned on.
3. Click **Start Now**.
 - Software data and run results are backed up.
 - Sequencing output and intermediate files are backed up based on backup settings.
 - The files are cleaned up based on cleanup settings.

Configure a schedule to back up additional files

Role—System administrator.

The system administrator can configure a schedule in Genexus™ Software to back up additional files to external data storage or to a cloud storage space. Additional files include 1.wells files and other intermediate files. These files may not need to be backed up daily.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup settings** tab, in the **Backup Settings** section, in the **Back up additional files** section, edit the backup schedule for additional files.

Select **Daily** or **Weekly**, then select the day and time for the backup.

A backup procedure of additional files takes time. Consider scheduling the backup procedure of additional files on a weekend or at a time when the Genexus™ Integrated Sequencer is not in use.
3. Enable the **Backup & Cleanup Enabled** toggle switch in the top right of the screen. This starts the backup process at the scheduled time.
4. Click **Save**.

Set up a schedule to delete run result files

Role—System administrator.

The system administrator can set up a schedule in Genexus™ Software to permanently delete backed up run result files, .wells files, and other intermediate files from the sequencing runs. This helps to ensure that there is sufficient disk space on the Genexus™ Integrated Sequencer.

Intermediate files include BAM files, log files, and sequencing output files. These files are not backed up by default. When you set a schedule to delete these files, by default, the files are deleted when they are not backed up and when the sequencer disk space falls below 4 terabytes (TB). You can change the disk space threshold to delete these files from the sequencer.

Run results that have been backed up and deleted in the software are listed in the **Run Results** screen and show the archived symbol (📁) after the run name. These can be restored by an administrator.

Run results that are designated as retained are never deleted, even when you set a schedule in the software to permanently delete run results. For more information, see “Retain run results” on page 209.

IMPORTANT!

- By default, when the **Disk Cleanup Settings** are active, the software is set to delete results 12 months after the run was last updated and backed up. You can change this setting.
- Intermediate and .wells files are not backed up unless an administrator sets a schedule to back up these files. By default, these files are deleted when the free disk space threshold is met, even when these files are not backed up. If you select to keep the default auto-delete setting for intermediate and .wells files and the files are deleted, the impact is minimal.
You cannot perform the following actions when these files are deleted.
 - Reanalyze a run from basecalling.
 - Upload run results to Ion Reporter™ Software.
 - Run the CustomerSupportArchive plugin.

1. In the menu bar, click ⚙️ (**Settings**) ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, update the **Disk Cleanup Settings** section.

Option	Procedure
Do not automatically delete run results at any time.	In Delete Analysis Results , click No .
Change the schedule to permanently delete run results.	Run results are deleted only if the results have been backed up. 1. In Delete Analysis Results, click Yes. 2. Select a schedule to delete the files. • Weekly • Bi-Weekly • 3 months • 6 months • 9 months • 12 months • 24 months—This is the default setting. • 36 months
Do not automatically delete .wells files and intermediate files at any time.	In Delete additional files , click No .
Change the schedule to permanently delete .wells files and intermediate files.	In Delete additional files , click Yes , then select the threshold of free disk space on the sequencer, from 1 to 10 TB, that initiates deletion of these files. The software deletes .wells files and intermediate files when the disk space falls below the selected threshold. If you have specified the backup of intermediate files and .wells files, these additional files must be backed up before they can be deleted.

3. Click the **Backup & Cleanup Enabled** toggle switch to the enabled position to start the backup and cleanup process at the scheduled time.
4. Click **Save**.

Configure email notification settings for backup events

Role—System administrator.

The system administrator can configure Genexus™ Software to send email notifications to administrators for backup events, such as when a backup completes or when the Genexus™ Integrated Sequencer has limited disk space remaining. The email notification goes to all users with an administrator role.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, in the **Email Notifications Settings** section, select the option or options for which to send an email notification to administrators.
 - **Backup fails or has insufficient storage space**
 - **Backup completes**
 - **System disk space falls below**—Select the minimum amount of sequencer disk space that is available for when the software sends an email notification.
 - **Backup disk space falls below**—Select the minimum amount of backup disk space that is available for when the software sends an email notification.
 - **Cloud used space exceeds**—Select the maximum amount of cloud backup disk space that is used for when the software sends an email notification. This option is available only when you select a cloud storage mount drive for backups.
3. Click **Save**.

The email notification settings take effect immediately.

Review and download audit records for backup settings

Role—System administrator, organization administrator.

Administrators can review the audit trail in Genexus™ Software for changes made to backup events. Changes to backup events include backup settings, disk cleanup settings, and email notification settings.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup Settings** tab, click **Audit** in a section.
 - **Email Notification Settings**
 - **Disk Cleanup Settings**
 - **Backup Settings**
3. Review the list of audit records.
4. In the **Audit Details** screen, select a time stamp in the left of the screen to view the audit record.
5. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

Disk space usage

Role—System administrator, organization administrator.

Administrators can monitor disk space usage for the Genexus™ Integrated Sequencer and can change data backup settings in Genexus™ Software. Disk space usage information is available in the **Settings / Backup & Restore** screen. You can view the date and time of the last successful backup for data and run results and the date and time of the last successful backup of additional files, such as sequencing output and intermediate files, based on the backup settings.

A best practice to maintain sufficient disk space is a systematic plan that backs up sample results and additional files that you want to store to an external file storage system. A system administrator or Field Service Engineer (FSE) sets up a data backup strategy and determines best disk cleanup settings.

Organization administrators can configure the backup schedule for their organizations if they have their own backup directory. For more information, see “Configure a schedule to back up additional files” on page 380.

Before data can be backed up or archived, you must add one or more mount points. The mount point can be an external storage device or a cloud-based storage. After data backup, files are saved in a folder in the external storage that is named `AssayDev`. If multiple sequencers are connected to the software, the software generates a parent folder for each instrument.

When files are backed up, the files are copied to an external storage drive or to a cloud drive. The files remain on both the sequencer and the external storage drive.

When files are cleaned up, the files are deleted from the sequencer, but remain on the external storage drive.

Contact a system administrator to back up and delete data manually. An administrator can also change the schedule to back up and delete data.

Disk backup and cleanup settings

Role—System administrator, organization administrator.

Administrators use the **Backup Settings** screen to configure optimal backup and cleanup settings for their organizations. Frequently review the system disk space that is used, and update the settings, if needed.

Cleanup settings do not apply for run results that you retain. When you retain run results, the run data, including the run additional files, 1.wells files, and intermediate files, are retained on the instrument and in the backup drive. Run results that are retained are not deleted and are shown in the software.

Option	Description
Delete Analysis Results (clean up run results)	If enabled, run results are deleted from the Genexus™ Integrated Sequencer after a specified interval of time, for example, 12 months after a run is completed. Runs that are backed up and deleted are marked as Archived. An administrator can restore the runs to access the data. Archived runs are listed in the Run Results screen with the archived symbol (📁) after the run name. By default, archived runs are not listed. To see the archived runs, select Archived in the Filter runs by list.
Delete additional files (intermediate files and 1.wells files)	If enabled, intermediate files and 1.wells files are deleted when the disk space falls below a specified threshold, such as 4 TB. If you have not configured the mount point, or if you have not configured the backup settings for additional files, files are cleaned up when the space threshold is met.
Backup additional files (intermediate files and 1.wells files)	If enabled, intermediate files and 1.wells files are backed up at a specified time. For example, every Friday at 10:00 p.m. Because 1.wells files are 10 times larger than the run results files (BAM files), more time is needed to back up 1.wells files than to back up software data and run results.

Free up sequencer disk space

Role—System administrator, organization administrator.

Genexus™ Software checks that sufficient disk space is available for sequencing runs. If the Genexus™ Integrated Sequencer has less than 1 terabyte (TB) of unused disk space, no new sequencing runs can be started on the sequencer.

If there is insufficient disk space on the sequencer, an email notification is sent to both system administrators and organization administrators. Planned sequencing runs cannot proceed until data are backed up and deleted to create sufficient disk space.

Administrators can perform the following actions to free up disk space, if needed.

- In the **Backup & Restore** screen, click **Start Now** at any time to start a backup and cleanup event. See “Start a manual backup” on page 380.
- In the **Run Results** screen, manually delete one or more run results. See “Delete a run result” on page 385.
- In the **Run Results** screen, manually archive one or more run results. See “Free up sequencer disk space” on page 384.

The settings in the **Backup and Restore** screen are ignored when you manually delete or archive runs in the **Run Results** screen.

Delete a run result

Role—System administrator, organization administrator.

Administrators can delete run results, sample results, and additional files for a sequencing run from the Genexus™ Integrated Sequencer. When you delete a run result, data and files for the run are deleted from the sequencer and are no longer shown in Genexus™ Software.

If the run results are backed up before the deletion, an administrator can restore the run result and access the data.

IMPORTANT! This action cannot be reversed if you proceed. If you delete a run result that is not backed up, it cannot be restored. Ensure that you do not need the run result before you delete it.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to delete.
3. Click  **Delete**.
4. In the **Confirm delete data for selected result(s)** dialog box, confirm that the data are backed up, if needed, and select the files to delete.
 - **Sequencing Output**
 - **Intermediate Files**
 - **Analysis Results**

If you delete the run results files, the sequencing output and intermediate files are also deleted.

5. Click **Delete**.

IMPORTANT! When you click **Delete**, a confirmation message is not displayed, and the run results are immediately deleted. Ensure that you want to delete the run results before you click **Delete**.

The run results, sample results, and additional files for the sequencing run that you select are deleted from the sequencer and are no longer shown in the software.

You can view the audit trail for a run result that you have deleted in the **Settings / Audit Records** screen. For more information, see “Search audit records” on page 400.

Types of files for backup

Three categories of sequencing files are backed in Genexus™ Software.

Category	Types of files	Cumulative size per run	Reanalysis use
Sequencing output	1.wells	250–500 GB	Reanalysis from the basecalling step
Intermediate files	Troubleshooting	150–180 GB	Not applicable
Run results	• BAM • VCF • Reports • Plugin results	150–180 GB	Reanalysis from the alignment step

Disk space usage example

The amount of disk space used for a sequencing run depends on the nucleic acid sample type and the assay that is used for sequencing. The Genexus™ Integrated Sequencer has 20 TB of data storage and can store 40 to 50 whole chip data runs. If the sequencer has less than 1 terabyte (TB) of available storage space, no new runs can be started.

The following table is a representative example of assay data usage. This table lists chip specifications and example data usage for a sequencing run with the Ion Torrent™ GX5™ chip.

Assay	Oncomine™ Precision Assay GX		Oncomine™ Myeloid Assay GX v2	Oncomine™ Comprehensive Assay v3 GX	Oncomine™ TCR Beta-LR Assay GX	Oncomine™ Comprehensive Assay Plus GX
Sample type	cfTNA	FFPE DNA+RNA	DNA+RNA	DNA+RNA	RNA	DNA+RNA
Number of reads per chip (4 lanes)	48–60 million	48–60 million	48–60 million	48–60 million	48–60 million	80–100 million
Number of reads per lane	12–15 million	12–15 million	12–15 million	12–15 million	12–15 million	20–25 million
Signal processing input size per chip ^[1]	~2 terabytes	~2 terabytes	~3.5 terabytes	~2 terabytes	~3.5 terabytes	~2 terabytes
Basecalling input size per chip ^[2]	200–300 gigabytes	200–300 gigabytes	400–600 gigabytes	200–300 gigabytes	400–600 gigabytes	230–330 gigabytes
Data size of intermediate and results files and logs ^[3]	120–180 gigabytes	120–180 gigabytes	160–240 gigabytes	120–180 gigabytes	160–240 gigabytes	220–280 gigabytes
Data size per lane of intermediate and results files and logs	30–45 gigabytes	30–45 gigabytes	40–60 gigabytes	30–45 gigabytes	40–60 gigabytes	55–70 gigabytes

(continued)

Assay	Oncomine™ Precision Assay GX		Oncomine™ Myeloid Assay GX v2	Oncomine™ Comprehensive Assay v3 GX	Oncomine™ TCR Beta-LR Assay GX	Oncomine™ Comprehensive Assay Plus GX
Data size of backed up files and logs	30–45 gigabytes	30–45 gigabytes	40–60 gigabytes	30–45 gigabytes	40–60 gigabytes	55–70 gigabytes
Data size of results files per lane ^[4]	~10–20 gigabytes	~8–24 gigabytes	~32–48 gigabytes	~9–15 gigabytes	~16–48 gigabytes	~32–48 gigabytes
Recommended number of samples per lane	1 sample per lane	4 samples per lane	8 samples per lane	1.5 samples per lane	4 samples per lane	1 sample per lane
Data size of the results per sample	~10–20 gigabytes	~2–6 gigabytes	~4–6 gigabytes	~6–10 gigabytes	~2–6 gigabytes	~32–48 gigabytes

^[1] The signal processing input on the sequencer is deleted after the run is complete and successful.

^[2] Administrators can schedule when basecalling input files are deleted. By default, these files are not retained. For more information, see “Set up a schedule to delete run result files” on page 380.

^[3] Includes QC, plugins, logs, results, and other assay and variant database files.

^[4] For more information about results files, see “Results files” on page 178.

Archive and restore run results and sample results

Role—System administrator, organization administrator.

Administrators can archive run data that are no longer needed. Run data consists of run results, sample results, and intermediate files or sequencing output files. This helps to make room for data storage on the Genexus™ Integrated Sequencer.

Archived run data is removed from the sequencer, but both run results and sample results that are associated with archived run data continue to be shown in Genexus™ Software until those sequencing runs are scheduled for cleanup. When a cleanup of archived run data is performed, older run results and sample results are automatically copied to the backup directory, based on settings that administrators configure. If there are multiple backup directories, the archived run data are copied to a backup directory that is assigned to the organization that performed the run. Archived run data continues to be retained on the backup directory after cleanup and can be restored to the sequencer from the backup directory.

Administrators can restore archived run data, provided that the run data has not been permanently deleted from the backup directory. For information about how to restore archived run data, see “Restore archived run results for a single run” on page 390 or “Restore multiple archived run results” on page 390.

Archive a run result

Role—System administrator, organization administrator.

Administrators can archive one to five run results at a time that are no longer needed on the Genexus™ Integrated Sequencer. Run data that is archived consists of run results, sample results, and intermediate files or sequencing output files.

Archived run data is removed from the sequencer, but run and sample results that are associated with archived data continue to be shown in Genexus™ Software until those archived runs are scheduled for cleanup.

When you archive run data, the run results files are archived by default. You can specify to archive 1.wells sequencing output files or intermediate files, or both.

If you archive run data at the time of a scheduled backup, the disk cleanup is skipped for that instance.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, select the checkbox of one or more run results that you want to archive, then at the top of the screen, click  **Archive Data**.
3. In the **Archive Confirmation** dialog box, select **Sequencing Output** to include 1.wells or select **Intermediate files** to include intermediate files, or select both.
4. Click **Archive**.

Archived runs results are listed in the **Run Results** screen with the archived symbol () after the run name.

To restore run results, see “Restore archived run results for a single run” on page 390 or “Restore multiple archived run results” on page 390.

Restore archived run results for a single run

Role—System administrator, organization administrator.

Administrators can restore run data in Genexus™ Software for a single sequencing run that is archived. Run data consists of run results, sample results, and intermediate files or sequencing output files. Archived run data for the sequencing run is retained on the backup directory, even after cleanup, and is restored to the sequencer from the backup directory.

Run results that are associated with archived run data are shown in the **Results / Run Results** screen with  (archived) after the run name. The **Restore** link is available when you place the pointer over the row of the archived run.

If you restore a run that has an assigned sample tag that has been deleted in the software, the deleted sample tag is recreated when the run result is restored and you can reassign the tag, if needed. For more information, see “Manage sample tags” on page 69.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, select **Archived** to filter the view of run results.
3. Place the pointer over archived run name of interest, then click **Restore**.
4. In the **Confirm** dialog box, click **YES**.
After the archived run data is restored, you can view the run results, sample results and additional files for the sequencing run in the software.
5. In the **Run Results** screen, click the run name to view the restored run results.

Restore multiple archived run results

Role—System administrator, organization administrator.

Administrators can restore run data in Genexus™ Software for multiple sequencing runs that are archived. Run data consists of run results, sample results, and intermediate files or sequencing output files. Archived run data for the sequencing runs is retained on the backup directory, even after cleanup, and is restored to the sequencer from the backup directory.

Administrators can restore multiple run results that have been archived in the **Restore Runs** screen in Genexus™ Software. You can search the list of runs that can be restored by the run name or by a sample tag name.

Run results that are associated with archived run data are shown in the **Results / Run Results** screen with  (archived) after the run name.

If you restore run data that has an assigned sample tag that has been deleted in the software, the deleted sample tag is recreated when the run data is restored and you can reassign the tag, if needed. For more information, see “Manage sample tags” on page 69.

1. In the menu bar, click  (**Settings**) ▶ **Backup & Restore**.
2. In the **Backup & Restore** screen, click the **Restore Runs** tab and select from the following options.
 - a. For run type, select **Archived runs** in the list box.

- b. Select the mounted drive that contains the run data that you want to restore, then click **Browse**.
 - c. In the **Select backup folder to list runs** list, navigate to the storage directory or drive, select the subfolder that contains the run data that you want to restore, then click **Select**.
Archived run data from that folder are listed on the **Restore Runs** tab.
 - d. In the **Restore Runs** tab, select the checkbox in the row for each sequencing run that you want to restore.
Filter the run list by run name or tag name or use the **Search** box.
3. Click **Restore Runs**.
The process to restore run data takes time. After the archived run data is restored, you can view the run results, sample results and additional files for the sequencing runs in the software.
 4. (Optional) After the archived run data are restored, click **Check Log** to view whether the process was successful.

Import archived sample results from external storage to a sequencer

Role—System administrator.

The system administrator can import archived run data from external data storage into a Genexus™ Integrated Sequencer. Run data consists of run results, sample results, and intermediate files or sequencing output files.

- The external data storage that contains the archived run data must be connected to the sequencer to which you import the archived run data.
- The assay for the archived run data that you want to import must exist in Genexus™ Software. The name of the assay, the barcode kit, and the extraction method type, must be identical between the source sequencer and the target sequencer.

If the assay does not exist in the software, import it. For information about how to import an assay, see “Transfer an assay to another sequencer” on page 260.

Note: Samples that do not exist in the software are included as part of the import and do not need to be recreated before the import.

After run data are imported, you can access **Run Results** and **Sample Results** in the target sequencer. Sequencer details are also listed with the instruments in the target sequencer.

1. In the menu bar, click ⚙️ (**Settings**) ▶ **Backup & Restore**.
2. In the **Backup & Restore** screen, click the **Restore Runs** tab and select from the following options.
3. In the type of runs dropdown list, keep the selection of **Archived runs**.
4. In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click **Browse**.

5. In the **Select backup folder to list runs** dialog box, navigate to the folder for a different system, then click **Select**.
6. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to import, then click **Restore Runs**.
Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags.
The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.
7. (Optional) After the archived run is imported, click **Check Log** to view if the process was successful or if there are any problems, such as if a sample has not been created in the software.

Restore all run results after a system crash

Role—System administrator.

Use this procedure only if there is a system crash and you have worked with a Field Service Engineer (FSE) to restore the software data on a reimaged system. After the software data is restored, the system administrator can restore all backed up run results.

1. In the menu bar, click  **(Settings) ▶ Backup & Restore**.
2. In the **Backup & Restore** screen, click the **Restore Runs** tab.
3. Select **All runs** from the list box.
4. Select the mounted drive that contains the runs that you want to restore, then click **Browse**.
5. In the **Select backup folder to list runs** dialog box, navigate to the folder that contains the runs that you want to restore, then click **Select**.
6. When a list of runs appears on the **Restore Runs** tab, select the checkbox in the row for each run that you want to restore. You can select specific runs, or you can restore all backed up runs after a system restore.
Use the search field to search for samples by a run name or by sample tags.
7. Click **Restore Runs**.
The process to restore runs takes time. After the runs are restored, you can view the run results in the software.
8. (Optional) After the runs are restored, click **Check Log** to view if the process was successful or if there are problems.

Manage plugins

Role—System administrator.

The system administrator manages plugins.

- System-installed plugins

System-installed plugins are included with Genexus™ Software. New plugins can be installed as part of a software package, such as an ADF, or during a software upgrade. For more information about system-installed plugins, see Chapter 13, “Plugins for data analysis”.

- User-defined plugins

The system administrator can install user-defined plugins in Genexus™ Software. User-defined plugins can be downloaded from the Thermo Fisher Scientific App Store. For more information, see “Download and install software updates from the App Store” on page 370.

The system administrator can import a user-defined plugin ZIP file that is downloaded from the App Store (see “Import a plugin” on page 394).

To import a user-defined plugin file, the file must contain valid content, must be configured correctly, and must be in the proper file format. The import fails if the plugin file is not valid.

A user-defined plugin can be installed using a USB drive. For more information, see “Download and install software updates with a USB drive” on page 371.

A software upgrade does not overwrite a user-defined plugin.

View plugins

Role—System administrator.

The system administrator can view the plugins that are installed in Genexus™ Software.

1. In the menu bar, click  **(Settings)** ▶ **Manage Plugins**.
2. In the **Settings / Manage Plugins** screen, review the list of plugins that are installed in the software.
3. *(Optional)* Use the **Filter Plugin Status** filter to show only those plugins that you need.
4. Click a plugin name to review the details.

Import a plugin

Role—System administrator.

The system administrator can install user-defined plugins in Genexus™ Software. Plugins with the same name and different versions can be imported. Only one version of a plugin can be the active version. An older version is marked as obsolete when a newer version is imported.

1. In the menu bar, click  **(Settings) ▶ Manage Plugins**.
2. In the **Settings / Manage Plugins** screen, click **Import Plugin**.
3. In the **Install Plugin** dialog box, click **Select File**, navigate to select the plugin file, then click **Upload**.

The file is validated during the import. If valid, the imported plugin appears in the plugin list.

Uninstall a plugin

Role—System administrator.

The system administrator can uninstall user-defined plugins in Genexus™ Software.

Uninstalled plugins cannot be used in an assay. If an uninstalled plugin is associated with an existing assay, a warning message on the assay details indicates that the plugin cannot be executed.

A plugin that is used in a locked assay cannot be uninstalled. If an assay using that plugin is obsoleted and no runs are pending that use that assay, then the plugin can be uninstalled.

Uninstalling a plugin does not activate its older version. The older version can be manually activated.

System-installed plugins cannot be uninstalled.

1. In the menu bar, click  **(Settings) ▶ Manage Plugins**.
2. In the **Settings / Manage Plugins** screen, place the pointer over the name of an active user-defined plugin, then click **Uninstall**.
3. In the **Uninstall plugin** dialog box, click **Yes** to confirm.

The plugin is removed from the plugin list.

Obsolete a plugin

Role—System administrator.

The system administrator can obsolete an active user-defined plugin so that it is no longer available for use in an assay in Genexus™ Software. An obsoleted plugin is still used if previously configured in an assay.

System-installed plugins cannot be obsoleted.

1. In the menu bar, click  **(Settings) ▶ Manage Plugins**.
2. In the **Settings / Manage Plugins** screen, place the pointer over the name of a user-defined plugin, then click **Obsolete**.
3. In the **Obsolete Plugin** dialog box, click **Yes** to confirm.
The plugin status changes to **INACTIVE** in the plugin list.

Activate an inactive plugin

Role—System administrator.

The system administrator can reactivate an inactive user-defined plugin that has been obsoleted in Genexus™ Software.

1. In the menu bar, click  **(Settings) ▶ Manage Plugins**.
2. In the **Settings / Manage Plugins** screen, place the pointer over the name of a user-defined plugin with a status of **INACTIVE**, then click **Activate**.
3. In the **Activate Plugin** dialog box, click **Yes** to confirm.
The plugin status changes to **ACTIVE** in the plugin list.

Review and download audit records for plugins

Role—System administrator.

The system administrator can review and download audit records for plugins.

The following user actions generate an audit record for a plugin.

- Import a plugin.
- Uninstall a plugin.
- Obsolete a plugin.
- Activate an inactive plugin.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **Audit Records**.
2. In the **Settings / Audit Records** screen, specify the filters, select **Plugin** in the last filter, then click **Search**.
3. Select a plugin audit record, then click **Audit**.
4. In the **Audit Details** screen, click a time stamp in the left of the screen to view details of the audited actions.
5. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

Manage audit records

Role—System administrator, organization administrator.

Administrators can use the options in the **Settings / Audit Records** screen to configure and manage audit records. Information about actions that are completed in the software is shown in the **Audit Records** screen, including the actions of users on samples, libraries, assays, planned runs, system setup, accounts, filter chains, gene lists, variants, software updates, user accounts, and organizations.

The **Audit Records** screen lists the available audit records for all actions that are performed on any data object, and the following information for each audited data object. Further details concerning the action on an object, including old values and new values, are available through the **Audit** link that accompanies each audit record.

The following information for an audit record is listed in the **Audit Records** screen.

Item	Description
User	The first and last name of the person who was signed in when the time-stamped action was performed.
Name	The name of the entity, such as the name of the sample or the gene list, for which an audit record is available.
Action Performed	The action that is performed on the run plan. • Create • Edit • Delete • Obsolete

(continued)

Item	Description
Data Object Name	The type of data object that is affected by the action. • All • Assay • Filter Chain • Gene List • Library Batches • Organization • Planned Runs • Plugin • Sample • Software Update • Software Setup • Thermofisher Account • User • Variants
Time Stamp	The time and date of the action.
Record	A link that opens the Audit Details screen for the selected object.

Audited actions can be searched and sorted, and the audit records can be downloaded and printed.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

Configure a reason for change requirement for audits

Role—System administrator, organization administrator.

Administrators can configure the audit function to specify that a reason for a change be required when a user performs an auditable action, such as change information or start a backup procedure. When a reason is required, the user must supply a reason for change when taking the action, and the reason is included in the audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, click  **Audit Configuration**.
3. In the **Audit Configuration** dialog box, select the **Require Reason** checkbox next to the **Data Object Name** for which a reason for change is required.
4. Click **Save**.

Users who make changes to the data object name for a which a reason is required, must include a **Reason for Change**. In the audit record that includes the change, the **Reason for Change** is shown in the list of time-stamped audit records in the **Audit Details** screen and in an exported PDF of the audit details.

To remove a reason for change requirement, click  **Audit Configuration**, in the **Audit Configuration** dialog box, deselect the **Require Reason** checkbox, then click **Save**.

Review and download audit records for a selected data object

Role—System administrator, organization administrator and manager, operator.

Users can search for, and then review and download the entire history of actions for a selected data object in Genexus™ Software for a user or date range in the **Settings / Audit Records** screen. A data object is a software component or data that is used in the software. An audit trail lists the records of actions made to the selected data object. Each time-stamped audit record shows the original values and any new values for the data object.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, you can narrow the list of audit records that are shown and then find the audit record.
 - a. To select a date range, click  **Calendar** next to **Start Date** and **End Date**.
If an end date is not selected, the search results include all records from the start date through the current date.

- b. To limit the results list to actions that are performed by a specific user, select the user from the **Select User** list.
 - c. To limit the results to specific actions, select **Edit**, **Create**, **Delete**, or **Obsolete** from the **Select Action** list. If you leave **Select Action** as the choice, all audited actions are shown in the search results.
 3. Select a data object to audit from the list of audit records.

The list is set to **Sample** by default and all items that are listed in the **Audit Records** screen have the data object of **Sample** in the **Data Object Name** column. You can change the type of objects that are shown in the table. For example, you can search for audit records of run plans when you select **Planned Runs** in the data object list.

If you select **All** in the data object list, you need to also select a value in the **Select User** dropdown list.

 - **All**
 - **Assay**
 - **Filter Chain**
 - **Gene List**
 - **Library Batches**
 - **Organization**
 - **Planned Run**
 - **Plugin**
 - **Sample**
 - **Software Update**
 - **System Setup**
 - **Thermofisher Account**
 - **Variants**
 4. Click **Search**.
 5. In the list of audit records shown, click **Audit** in the **Record** column.
 6. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for each audited action.
 7. In the **Audit Details** screen, you can do the following.

Option	Description
Review the audit record.	Each action that was performed for the assay is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attribute mappings Manage Assays screen.

Search audit records

Role—System administrator, organization administrator.

Administrators can search for an existing audit record with the search tools in the **Audit Records** screen.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, select the search parameters.
 - a. To select a date range, click  **Calendar** next to **Start Date** and **End Date**.

If an end date is not selected, the search results include all records from the start date through the current date.
 - b. To limit the results list to actions that are performed by a specific user, select the user from the **Select User** list.
 - c. To limit the results to specific actions, select from the **Select Action** list. If you leave **Select Action** as the selection, all audited actions are shown in the search results.
 - d. To list other items in the table, select from the data object list.

The list is set to **Sample** by default and all items that are listed in the **Audit Records** screen have the data object of **Sample** in the **Data Object Name** column. You can change the type of objects that are shown in the table. For example, you can search for audit records of run plans when you select **Planned Runs** in the data object list.

If you select **All** in the data object list, you must also select a value in the **Select User** dropdown list.
3. Click **Search**.
4. Click **Clear** to return to the default list of records, with the data object of **Sample** in the **Data Object Name** column.

Sort audit records

Role—System administrator, organization administrator.

Administrators can sort the columns in the **Settings / Audit Records** screen to display the list of audit records in a different order. The default order is the newest record on top.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, click the column heading of interest.
The list of records is reordered based on the heading that is selected. **User**, **Action Performed**, and **Data Object Name** sort alphabetically.
3. Click the column heading a second time to reverse the order of records displayed.
4. Click the **Time Stamp** heading to return to the default order, with the most recent record on top.

Export multiple audit records

Role—System administrator, organization administrator.

Administrators can export a print-ready PDF file of selected audit records from the **Audit Records** screen.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, select the record or records to be exported by clicking the checkbox next to the record of interest. Select the checkbox above the column to select all audit records.
3. Click  **Export**.
A PDF file is generated. Depending on your browser settings, the software downloads the file or prompts you to open or save the file.
4. Open the PDF file in an appropriate viewer to review and print the audit record or records.

Manage log files

Role—System administrator, organization administrator.

Administrators can view log files generated by the software and connected purification and sequencing instruments. Viewing log files is useful for troubleshooting activities.

Administrators can also manage the storage of log data by setting the retention period for the audit trail function and enabling auto-deletion rules for logs.

View log files

Role—System administrator, organization administrator.

Administrators can view software and instrument log files in Genexus™ Software.

1. Click  **(Settings)** ▶ **Logs**.
2. In the **Logs** screen, select an option in the software or connected instrument dropdown list.

Option	Description
View software log files.	Click Ion Torrent Genexus Software .
View log files for a connected purification or sequencing instrument.	Click Instrument .

3. Click the log file name of interest.

File name	Description
ApplicationLog_<date>.log	A log file with information related to some actions in the Genexus™ Software, including user sign-ins and sign-outs and failed attempts to sign-in or sign-out.
InstrumentGX5_<date>.log	A log of activity on the Genexus™ Integrated Sequencer.
InstrumentPurif_<date>.log	A log of activity on the Genexus™ Purification System.

The log file opens in a new tab in the browser.

Manage log settings

Role—System administrator.

The system administrator can manage the length of time that log files are available in Genexus™ Software and can configure the software to allow log files to be automatically deleted.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click  **(Settings)** ▶ **Logs**.
2. In the **Logs** screen, click  **Manage Logs**.
3. In the **Manage Logs** dialog box, in **Retention Period**, select the number of months that logs are to be retained.
4. Select **Enable Auto Deletion** to delete log files after the designated retention period.
5. Click **Save**.

Review and download audit records for log settings

Role—System administrator, organization administrator.

Administrators can review, export, or print the entire history of changes that are made to settings for the retention of log files in Genexus™ Software. An audit trail lists records of changes to log file settings. Each audit record shows the name of the setting attribute, the original setting, and the new setting.

1. Click  **(Settings)** ► **Logs**.
2. In the **Logs** screen, click  **Audit Trail**.
3. Review the list of audit records.
4. In the **Audit Details** screen, click a time stamp in the left of the screen to view details for an audited action.
5. (Optional) Click  **Download** to generate a PDF file of the records in the selected timestamp.

View system services

Role—System administrator, organization administrator and manager.

Administrators and managers can view information about system services on the **Settings / Services** screen. The information can be useful to troubleshoot error messages that are received during an active job, or to determine why a run failed.

Service	Description
Jobs Server	Shows a list of server information and active (running) software.
Services	Shows the service name, description, and status. The Services list can be used for troubleshooting. If a service has stopped during a run, an option to start the service is visible for administrators and managers. For more information, see “Start a stopped service” on page 404.
Active Jobs	• Shows a list of runs in progress that are active and queued on the Genexus™ Integrated Sequencer. • Allows administrators and managers to stop a job. For more information, see “Terminate an active job” on page 404.
ionCrawler Service Details	Provides information about the data transfer process from the sequencer. The information includes whether the service was started or stopped, which user completed the action, and a timestamp for the action.
Download Instrument Diagnostics	Contains the initialization logs of the sequencer, and allows administrators and managers to download the diagnostic archive file in ZIP format.
System Check Report	Administrators and managers can click Run System Check to view the system check report.

Terminate an active job

Role—System administrator, organization administrator.

If an active job is not going to be useful or if a different job is urgent, an administrator or manager can terminate an active job.

1. In the menu bar, click  **(Settings)** ▶ **Services**.
2. In the **Settings / Services** screen, in the **Active Jobs** section, find the active job that you want to stop, then click **Terminate**.
3. In the confirmation message, click **Terminate** to terminate the job.

Start a stopped service

Role—System administrator.

A system administrator can restart a stopped service if a service has stopped running for some reason. The two database services, **ntp** and **deeplaser**, and the **Ion Torrent API Service**, are the only services that can be restarted.

1. In the menu bar, click  **(Settings)** ▶ **Services**.
2. In the **Services** screen, in the **Services** section, find the service that you want to restart, then click **Start**.
3. In the confirmation message, click **Yes** to restart the service.

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You can expand the analysis capabilities of Genexus™ Software with plugins that are preinstalled with the software. The plugin results are shown in the software in a report summary and can be used for various purposes.

A system administrator can download and install more plugins from the Thermo Fisher™ Connect Platform.

coverageAnalysis plugin in Genexus™ Software

Use the coverageAnalysis plugin to view statistics and graphs that describe the level of sequence coverage produced for targeted genomic regions. The results for a run analyzed with the plugin vary based on the assay type that you select during assay creation. You can download output files with coverage statistics that are generated by the coverageAnalysis plugin.

Review coverageAnalysis plugin results

The coverageAnalysis plugin generates a **Coverage Analysis Report**. This report includes read statistics and several charts. The statistics and charts that are presented depend on the library type for the analysis.

The **Plugin Results** lists the barcode, the sample, the number of mapped reads, the percentage of on target reads, mean base coverage depth, and base coverage uniformity.

You can download coverageAnalysis plugin output files from the **Results** screen for a sample. For more information, see “Results files” on page 178 and “Output files generated by the coverageAnalysis plugin” on page 414.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.

3. Click the **Plugins** tab.

Plugin Results is selected and a summary table of the coverage analysis, by barcode, is shown in the **coverageAnalysis** summary pane.

4. In the **coverageAnalysis** summary pane, in the **Barcode Name** column, click the link in the row of the barcode of interest.

The detailed **Coverage Analysis Report** for the barcode opens in a separate window.

5. Click **⋮ (More) ▶ Download Files** to download coverageAnalysis plugin results files.

Note: Sometimes the file name can be too long to open in applications such as Excel™. To resolve this problem, right-click the file and click **Save As** to rename the downloaded file.

6. Use the options to view more reports, rerun the plugin, view the log for the plugin run, or delete the plugin output.

Option	Description
View more reports.	In Executed At , select an alternate timestamp, if available.
Configure the coverageAnalysis .	Select Select Plugins , then select the coverageAnalysis and click Configure .
Rerun the coverageAnalysis plugin.	Click  Select Plugins and select the coverageAnalysis plugin, then click Rerun Plugin .
View the coverageAnalysis log.	Click  View Log .
Delete the coverageAnalysis plugin output for the selected timestamp.	IMPORTANT! When you click Delete, a confirmation message is not shown and the delete is immediate. Ensure that you want to delete before you click Delete. If you accidentally delete the plugin results, you can rerun the plugin and then view the plugin results. Click  Delete.

coverageAnalysis plugin setup

The coverageAnalysis plugin uses the following settings.

Setting	Description
The following settings are available for all library types.	
Library Type	The default value is the assay type selected during assay creation.
Sample Tracking	The Ion AmpliSeq™ Sample ID Panel is a companion panel of 9 primer pairs that can be added to any Ion AmpliSeq™ human gDNA panel during target amplification to generate a unique identification label for research samples. Select this checkbox if you added the Ion AmpliSeq™ Sample ID Panel to the assay.

(continued)

Setting	Description
The following settings are advanced options.	
Uniquely Mapped Reads	Select this option to analyze only reads that are mapped to a unique location in the reference. Reads that are non-uniquely mapped can have equally well-aligned reads that are mapped to multiple locations and are typically mapped randomly to one.
Non-duplicate Reads	Select the checkbox to avoid copy reads. The analysis must have included alignments with Mark Duplicates active.
Minimum Aligned Length	Specify the minimum aligned length that is required to ensure that the read is included in an analysis. The default value 0 specifies that no reads are filtered out due to the aligned length for the reads.
Minimum Mapping Quality	Specify a minimum value that reads must exceed to be included in the analysis. The default value 0 specifies that no reads are filtered out due to the quality of the alignment of the reads.
Tier 1 Coverage Depth	Specify the first-tier coverage depth at which percentage of target coverage is reported. This value must be at least 2, because the coverage depth output is always specified at 1x read depth. The default value of 20 process that the percentage of targets, total base targets, and/or separate target bases with at least 20 reads is reported.
Tier 2 Coverage Depth	Specify the second-tier coverage depth at which percentage of target coverage is reported. This value must be greater than the value used for the first-tier coverage. The default value of 350 process that the percentage of targets, total target bases, and/or separate target bases with at least 350 reads is reported.
Tier 3 Coverage Depth	Specify the third-tier coverage depth at which percentage of target coverage is reported. This value must be greater that the value used for the second-tier coverage. The default value of 500 process that the percentage of targets, total target bases, and/or separate target bases with at least 500 reads is reported.

Example Coverage Analysis Report

The detailed **Coverage Analysis Report** that is available when you select a barcode in the coverageAnalysis plugin summary for a run includes a variety of reads statistics.

coverageAnalysis

Executed At: 2024-12-02 17:02 [View Log](#) [Delete](#)

Library type: AmpliSeq DNA
Target regions: OCAV3.20200626.GX5.designedUnmergedDetail

Barcode Name	Sample	Mapped Reads	On Target	Mean Depth	Uniformity
IonDual_0161-IonDual_0162	SRTestSample01	3,708,221	94.72%	1,049	59.90%

20 items per page 1 - 1 of 1 items

Reads statistics

The detailed **Coverage Analysis Report** that is available when you select a barcode in the coverageAnalysis plugin summary for a run includes a variety of reads statistics. The statistics that are displayed in your report depend on the type of library that is used in your sequencing experiment. Definitions are in tooltips. Place the pointer over a heading or description in the report to view the tooltip.

General statistics

Statistic	Description
Number of mapped reads	The total number of reads mapped to the reference genome.
Percent reads on target	The percentage of filtered reads mapped to any targeted region relative to all reads mapped to the reference. If no target regions file is specified, this value will be the percentage of reads passing uniquely mapped and/or nonduplicate filters, or 100% if no filters were specified. A read is considered on target if at least one aligned base overlaps at least one target region. A read that overlaps a targeted region but where only flanking sequence is aligned, for example, due to poor matching of 5' bases of the read, is not counted.
Average base coverage depth	The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, including any bases that have no coverage.
Uniformity of base coverage	The percentage of bases in all targeted regions (or in a whole genome) covered by at least 20% of the average base coverage depth reads. Cumulative coverage is linearly interpolated between nearest integer base read depths.

Amplicon read coverage statistics

The following statistics describe the reads that are assigned to specific amplicons. Each sequence read is assigned to exactly one of the amplicons specified by the targets file. If a read spans multiple amplicon targets, the target region that the reads covers most is assigned. In the event of a tie, the target that is the closest to the 3' end is assigned.

Statistic	Description
Number of amplicons	The number of amplicons that is specified in the target regions file.
Percent assigned amplicon reads	The percentage of reads that were assigned to individual amplicons relative to all reads mapped to the reference. A read is assigned to a particular (inner) amplicon region if any aligned bases overlap that region. If a read might be associated with multiple amplicons, the read is assigned to the either the amplicon with insert region that is best aligned to the ends of the read, or that has the greatest overlap of aligned sequence.
Average reads per amplicon	The average number of reads assigned to amplicons.

(continued)

Statistic	Description
Uniformity of amplicon coverage	The percentage of amplicons that had at least 20% of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts.
Amplicons with at least <i>N</i> reads	The percentage of all amplicons that had at least <i>N</i> reads.
Amplicons with no strand bias	The percentage of all amplicons that did not show a bias towards forward or reverse strand read alignments. An individual amplicon has read bias if it has ≥ 10 reads and the percentage of forward or reverse reads to total reads is $>70\%$. Amplicons with <10 reads are considered to have no strand bias.
Amplicons reading end-to-end	The percentage of all amplicons that were considered to have a sufficient proportion of assigned reads (70%) that covered the whole amplicon target from 'end-to-end'. To allow for error, the effective ends of the amplicon region for read alignment are within 2 bases of the actual ends of the region.
Amplicon base composition bias	A number that represents the proportion of amplicons showing low representation ($<0.2\times$ mean reads) in the lower and/or upper quartiles of amplicons ordered by increasing G/C base pair content of their insert sequences. The value is relative to that in the center 50th percentile of amplicons and weighted by the standard deviation of representation over all amplicons. An RMS (root mean square) value is used so that a bias greater in either upper or lower quartiles produces a larger value than a mean bias seen more equally in both outer quartiles. The value is 0 if the uniformity of amplicon coverage metric is 100%, however, the value is not necessarily high at lower amplicon uniformity.

Target base coverage statistics

The following statistics describe the targeted base reads of the reference. A base covered by multiple target regions is counted only once per sequencing read.

Statistic	Description
Bases in target regions	The total number of bases in all specified target regions of the reference.
Percent base reads on target	The percent of all bases covered by reads aligned to the reference that covered bases in target regions. Clipped bases, deletions, and insertions (relative to the reference) are not included in this percentage. If no specific target regions were specified, the whole genome is the targeted regions.
Average base coverage depth	The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage.
Uniformity of base coverage	The percentage of bases in all targeted regions (or whole genome) that is covered by at least 20% of the average base coverage depth reads. Cumulative coverage is linearly interpolated between nearest integer base read depths.

(continued)

Statistic	Description
Target base coverage at Nx	The percentage of target bases covered by at least <i>N</i> reads.
Target bases with no strand bias	The percentage of all target bases that did not show a bias toward forward or reverse strand read alignments. An individual target base is considered to have read bias if it has ≥ 10 reads and the percentage of forward or reverse reads to total reads is $>70\%$. Target bases with <10 reads are considered to have no strand bias.
Percent end-to-end reads	The percentage of on-target reads that fully cover their assigned amplicon (insert) from 'end-to-end'. To allow for error, the effective ends of the amplicon region for read alignment are within 2 bases of the actual ends of the region.
Bases in targeted reference	The total number of bases in all target regions of the reference. This statistic applies only to whole genome library analyses.
Bases covered (at least 1x)	The total number of target bases that had at least one read aligned over the proximal sequence. Only the aligned parts of each read are considered. For example, unaligned (soft-cut) bases at the 5' ends of mapped reads are not considered. Covered target reference bases can include sample DNA read base mismatches, but does not include read base deletions in the read, nor insertions between reference bases. This statistic applies only to whole genome library analyses.
Average base read depth	The average number of reads of all targeted reference bases that were read at least one time. This statistic applies only to whole genome library analyses.
Genome Base Coverage	Summary statistics for base reads of the reference genome. This statistic applies only to whole genome library analyses.
Genome base coverage at Nx	The percentage of reference genome bases covered by at least <i>N</i> reads. This statistic applies only to whole genome library analyses.

Example charts generated by the coverageAnalysis plugin

Many of the charts in the detailed **Coverage Analysis Report** include a **Plot** menu that allows you to change characteristics of the chart. For example, you can show both strands (see callout 3 in Figure 8).

The **Q** button (in the top right corner of a chart) opens the chart **Viewing Options** panel. The **?** button opens a description of the chart.

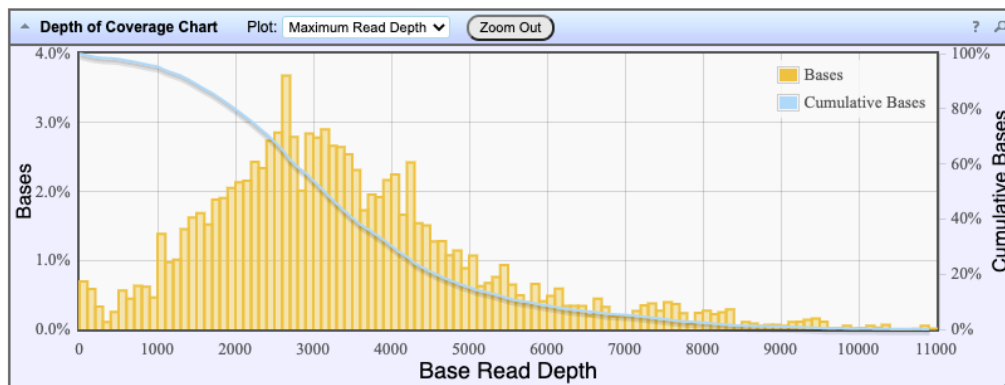


Figure 4 Representative Depth of Coverage Chart The Depth of Coverage Chart shows the distribution of targeted base coverage. The X-axis represents the base read depth. The left Y-axis represents the number of reads at a given base read depth or a range (bin) of base read depths, as a percentage of the total number of base reads. The right Y-axis represents the cumulative count of the number of reads at a given read depth or greater, as a percentage of the total number of reads. The individual orange bars represent the percentage of reads in the specific range of base read depths. The blue curve measures the cumulative reads at a given base read depth or greater. If your analysis includes a regions of interest file, this chart reflects only target regions (reads that fall within a region of interest).

In most charts, you can click a data point to open a detail pane for that data point. For example, in the Depth of Coverage Chart in Figure 4, click an individual orange bar to open the detail pane for bases within a specific range of base read depths. Click a point on the blue curve to open the detail pane (Figure 5) for cumulative bases at that base read depth or greater.

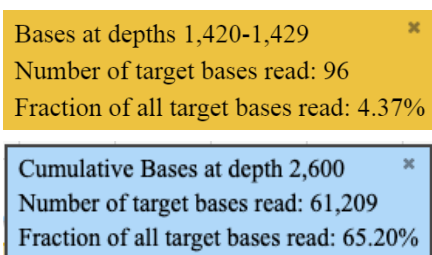


Figure 5 Example detail panes

Similarly, you can click any point or bar within the Amplicon Coverage chart to view details for each amplicon coverage bin (see Figure 7).

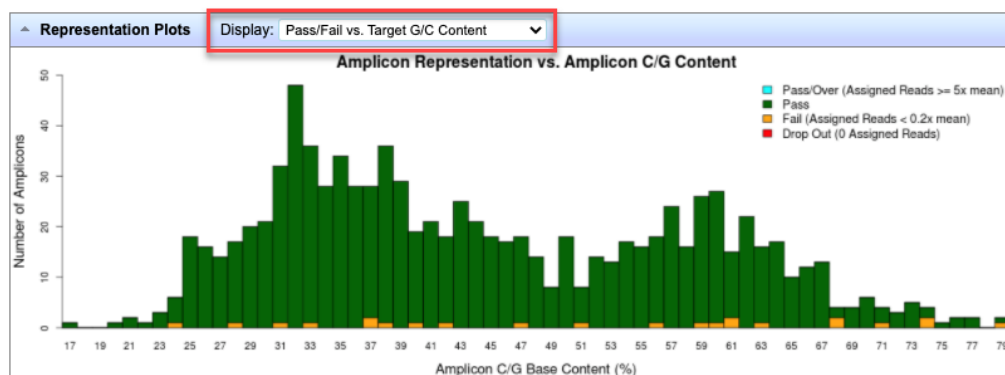


Figure 6 Representation Plots Use the **Display** list to view different amplicon representation plots. This figure shows an example Pass/Fail vs. Target G/C Content plot.

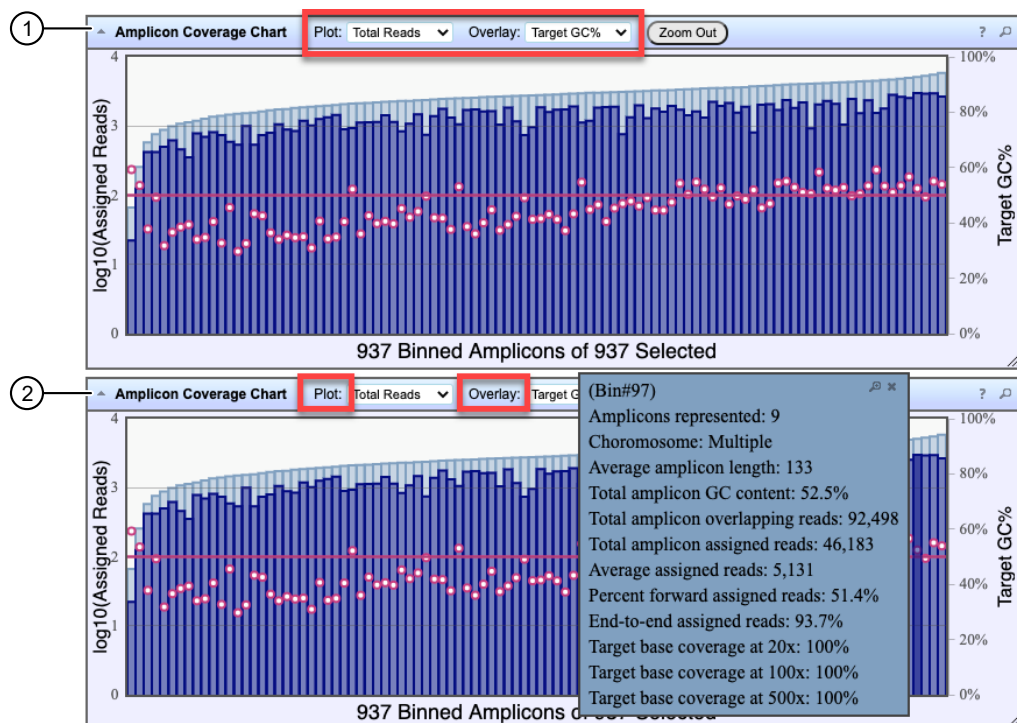


Figure 7 Example Amplicon Coverage Chart The Amplicon Coverage charts summarize the amplicon coverage results. Several plots views are available. The X-axis in all plots represents individual amplicons. The left Y-axis in all plots represents assigned reads (on a base 10 log scale). Use the **Plot** and **Overlay** lists to customize the chart view. The right Y-axis varies depending on the plot view that is selected from the **Overlay** dropdown list. Click any bar or any point on the chart to view the detail pane for that data.

- ① Example Total Reads plot with a Target GC percentage overlay
- ② Click a bar to open the detail pane for the specific amplicon. Click **View in Reference Coverage Chart** to zoom in on the amplicon in the **Reference Coverage Chart** (see callout 3 in Figure 8).

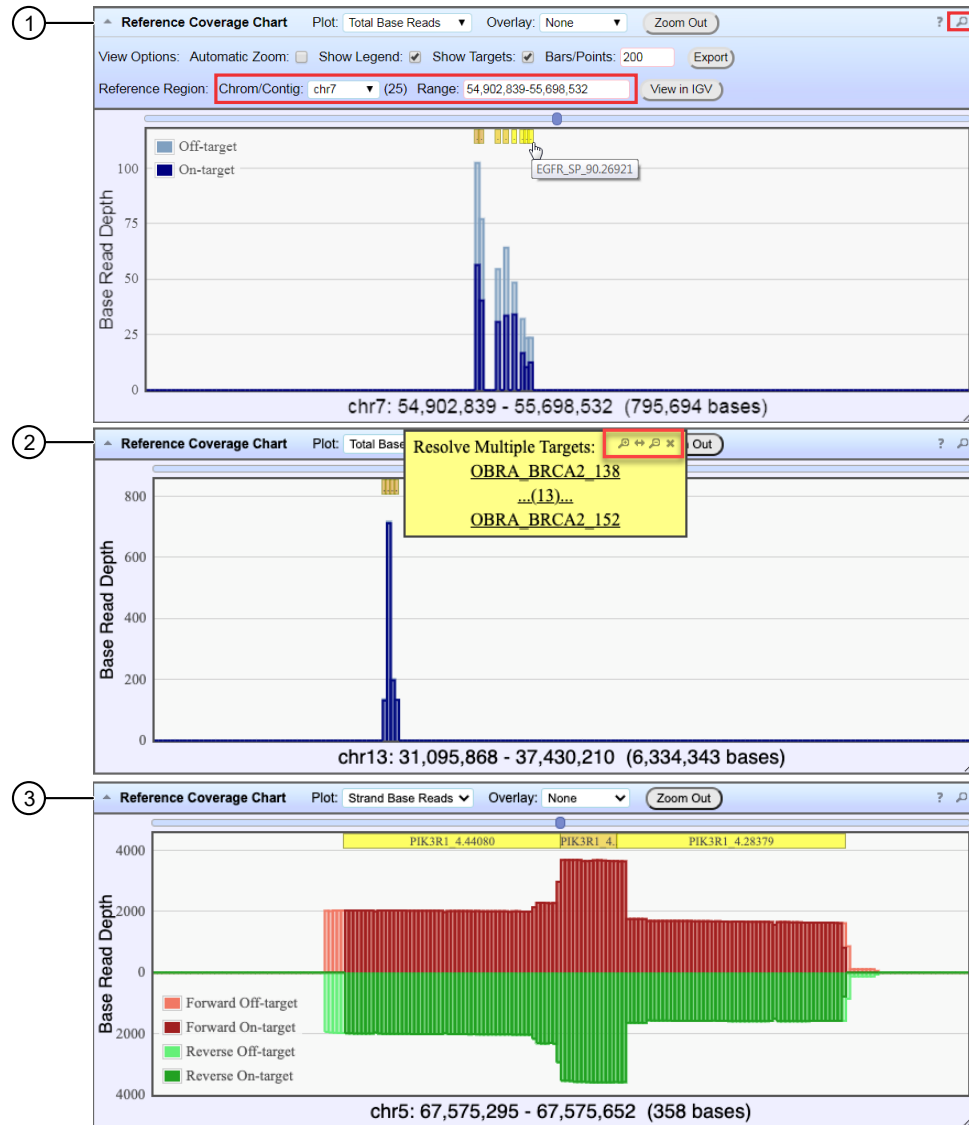


Figure 8 Example Reference Coverage Chart The Reference Coverage Chart is an overlay of where target regions are defined and overlap on the reference. The X-axis represents the target region chromosomal position. The Y-axis represents the Base Read Depth.

- ① Example Total Base Reads plot with Viewing Options panel expanded. The plot is zoomed in on a specific chromosomal region. Click a yellow bar (top of the chart) to open the detail pane for a specific amplicon in the Amplicon Coverage Chart (see callout 2 in Figure 7).
- ② Example Total Base Reads plot where a chromosomal region has multiple targets. In the Resolve Multiple Targets pane, click 🔍 to zoom in on the region, or click one of the links to open the detail pane for the specific amplicon in the Amplicon Coverage Chart.
- ③ Example Strand Base Reads plot that is zoomed in on one specific amplicon. Click the yellow box to open the detail pane for the specific amplicon in the Amplicon Coverage Chart.

Output files generated by the coverageAnalysis plugin

You can download coverageAnalysis plugin results files from the **Results** screen for a sample. For more information, see “Results files” on page 178.

Note: Sometimes the downloaded file name can be too long to open a file in applications such as Microsoft™ Excel™. To resolve this problem, right-click the file, click **Save As**, then rename the downloaded file with a shorter name.

The following tables describe the files that can be generated by the coverageAnalysis plugin. The list of files depends on the type of assay that was selected during assay creation.

File download selections in the Genexus™ Software

Selection	File Name
DNA Coverage Statistics	<DNA Barcode><ExpName>.stats.cov.txt
DNA Chromosome base coverage summary	<DNA Barcode><ExpName>.chr.cov.xls
DNA Base depth of coverage	<DNA Barcode><ExpName>.base.cov.xls
DNA Amplicon coverage summary	<DNA Barcode><ExpName>.amplicon.cov.xls
DNA Coverage Analysis Summary (.pdf)	<DNA Barcode><ExpName>.summary.pdf
RNA Coverage Statistics	<RNA Barcode><ExpName>.stats.cov.txt
RNA Amplicon coverage summary	<RNA Barcode><ExpName>.amplicon.cov.xls
RNA Coverage Analysis Summary (.pdf)	<RNA Barcode><ExpName>.summary.pdf
TNA Coverage Statistics	<TNA Barcode><ExpName>.stats.cov.txt
TNA Chromosome base coverage summary	<TNA Barcode><ExpName>.chr.cov.xls
TNA Base depth of coverage	<TNA Barcode><ExpName>.base.cov.xls
TNA Amplicon coverage summary	<TNA Barcode><ExpName>.amplicon.cov.xls
TNA Coverage Analysis Summary (.pdf)	<TNA Barcode><ExpName>.summary.pdf

File contents

File	Description
Coverage Statistics	A summary of the statistics presented in the tables at the top of the plugin report. The first line is the title. Each subsequent line is either blank or a particular statistic title followed by a colon (:) and its value.
Chromosome base coverage summary	Base reads per chromosome summary data that is used to create the default view of the Reference Coverage Chart. This file contains the following fields. <code>chrom</code>—The name of the chromosome or contig of the reference. <code>start</code>—The coordinate of the first base in this chromosome. This is always 1. <code>end</code>—The coordinate of the last base of this chromosome. Also, its length in bases. <code>fwd_basereads</code>—The total number of forward strand base reads for the chromosome. <code>rev_basereads</code>—The total number reverse strand base reads for the chromosome. <code>fwd_trg_basereads</code> (if present)—The total number of forward strand base reads that mapped over at least one target region. <code>rev_trg_basereads</code> (if present)—The total number of reverse strand base reads that mapped over at least one target region. <code>total_reads</code>—The total number of sequencing reads that are mapped to individual contigs.
Base depth of coverage	Coverage summary data used to create the Depth of Coverage Chart. This file contains the following fields. <code>read_depth</code>—The depth at which a (targeted) reference base has been read. <code>base_cov</code>—The number of times any base was read (covered) at this depth. Lines (read depths) for which <code>base_cov</code> is 0 are omitted to avoid excessively large files being produced in specific situations. <code>base_cum_cov</code>—The cumulative number of reads (coverage) at this read depth or greater. <code>norm_read_depth</code>—The normalized read depth (depth divided by average base read depth). <code>pc_base_cum_cov</code>—The same as <code>base_cum_cov</code> but represented as a percentage of the total base reads.

File contents (continued)

File	Description
Amplicon coverage summary	Coverage summary data used to create the Amplicon Coverage Chart. This file contains the following fields: <code>contig_id</code>—The name of the chromosome or contig of the reference for this amplicon. <code>contig_srt</code>—The start location of the amplicon target region. This coordinate is 1-based, unlike the corresponding 0-based coordinate in the original targets BED file. <code>contig_end</code>—The last base coordinate of this amplicon target region. The length of the amplicon target is given as $tlen = (contig\_end - contig\_srt + 1)$. <code>region_id</code>—The ID for this amplicon as given as the fourth column of the targets BED file. <code>gene_id</code> or <code>attributes</code>—The gene symbol or attributes field as provided in the targets BED file. <code>gc_count</code>—The number of G and C bases in the target region. The %GC that is uses this count divided by the amplicon (insert) length. <code>overlaps</code>—The number of times this target was overlapped by any read by at least one base. Individual reads might overlap multiple amplicons where the amplicon regions themselves overlap. <code>fwd_e2e</code>—The number of assigned forward strand reads that read from one end of the amplicon region to the other end. <code>rev_e2e</code>—The number of assigned reverse strand reads that read from one end of the amplicon region to the other end. <code>total_reads</code>—The total number of reads assigned to this amplicon. This value is the sum of <code>fwd_reads</code> and <code>rev_reads</code> and is the field that rows of this file are ordered by (then by <code>contig id</code>, <code>srt</code>, and <code>end</code>). <code>fwd_reads</code>—The number of forward strand reads that are assigned to this amplicon. <code>rev_reads</code>—The number of reverse strand reads that are assigned to this amplicon. <code>covNx</code>—The number of bases of the amplicon target that had at least N reads. There are 3 such columns for the specified coverage tiers, which by default are <code>cov100x</code>, <code>cov350x</code>, and <code>cov500x</code>.
Coverage Analysis Summary (.pdf)	A PDF file that contains the Coverage Analysis Report, including read statistics and charts that are generated by the coverageAnalysis plugin.

Customer Support Archive

A CustomerSupportArchive file is a downloadable archive that a support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Software. The archive contains log files and other technical data about the software and other files from sequencing runs.

The CustomerSupportArchive plugin generates a Customer Support Archive (CSA) file during sequencing runs. You can also rerun the CustomerSupportArchive plugin after a sequencing run. For more information, see “Run plugins after a sequencing run is complete” on page 184.

IMPORTANT! If you are copying and editing an assay, or running the plugin after a sequencing run, ensure that you select the latest CSA version. For more information about the latest version, see the Genexus™ Software release notes.

You can download the Customer Support Archive (CSA) file from the **Results / Sample Results** and the **Results / Run Results** screens.

For more information, see “Download a Customer Support Archive” on page 586.

generateGenome plugin in Genexus™ Software

Use the generateGenome plugin to generate a consensus genome for an individual sample that is based on a reference genome and variants called in a sequencing run. The full consensus genome for the sample is returned in a compressed FASTA file with regions that are not sufficiently covered, or otherwise of high uncertainty, denoted by 'N' characters for bases. It is therefore primarily suited to small genomes, such as viruses, but may also be used with larger targeted genomes that have multiple contigs or chromosomes.

If the **Major Allele Only** option is not selected in the plugin configuration, genome homozygous variants are substituted for reference bases and heterozygous variants may be denoted by International Union of Pure and Applied Chemistry (IUPAC) base codes, such as "Y" for C/T, in the FASTA file.

The plugin results also include a reference coverage report with information about the degree of reference coverage and how many variants of different types are annotated.

This plugin can be run for multiple samples when it is added to an assay that is used in a sequencing run, or can be run for each sample individually after the sequencing run. For more information, see “Assay creation overview” on page 233 and “Run plugins after a sequencing run is complete” on page 184.

Review generateGenome plugin results

The **generateGenome Report** in the generateGenome plugin results report includes some basic statistics about the genome reference file in a separate report that is available for download from the **generateGenome Report**.

The full consensus genome for the sample is returned in a compressed FASTA file with the following results.

- Regions that are not sufficiently covered, or otherwise of high uncertainty, are denoted by 'N' characters for bases.
- Reference bases are substituted with bases detected at homozygous variant positions.
- By default, in the FASTA file, the highest frequency allele is substituted into the reference where a variant is detected, or the International Union of Pure and Applied Chemistry (IUPAC) base codes, such as "Y" for C/T will be substituted, if the **Major Allele Only** option is not selected in the plugin configuration.

The **generateGenome Report** also includes a report named **Variant calls per contig summary**, which summarizes the number of variant calls made by the software for a contig, compared to the reference genome for that contig.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.
3. Click the **Plugins** tab.
Plugin Results is selected and a summary table of the coverage analysis, by barcode, is shown in the **generateGenome** summary pane.
4. In the **generateGenome** pane, in the **Barcode Name** column, click the link in the row of the barcode of interest.
The detailed **generateGenome Report** for the barcode opens in a separate window. The report provides tooltips with definitions. Place the pointer over a heading or description in the report to view the tooltip. For more information, see “Example generateGenome Report” on page 420.
5. Click the links at the bottom of the **generateGenome Report** to download a statistics file, a summary file, or an output file of the plugin results for the selected barcode.

Option	Description
Consensus sequence summary statistics	View a text summary of variant call summary statistics for the sample results. The statistics table can also be downloaded as a tab-delimited text file from the “Download Barcode Summary Report” link in the Plugins tab.
Variant calls per contig summary	Download the call summary for individual reference contigs in table format (TSV with XLS extensions).
Download consensus genome (FASTA)	Download the sample consensus FASTA file for this barcode. The file is in zip.gz format. You must use a tool such as 7-Zip in Microsoft™Windows™, or a command-line option in Linux™, to decompress and convert the file into FASTA format.

6. (Optional) Click the browser back button to return to the **Plugins** tab and use the options to view more reports, rerun the plugin, view the log for the plugin run, or delete the plugin output.

Option	Description
Rerun the generateGenome plugin.	In the Plugins tab, click  Select Plugins and select the generateGenome plugin, then click Rerun Plugin .
Click the link Download Barcode Summary Report .	View the consensus sequence summary statistics tab-delimited text file, which shows a summary of variant call summary statistics for the sample results.
View the generateGenome log.	Click  View Log . Review the generateGenome log for details about run parameters and the performance of the plugin.
Delete the generateGenome plugin output for the selected timestamp.	IMPORTANT! When you click Delete, a confirmation message is not displayed and the delete is immediate. Ensure that you want to delete before you click Delete. If you accidentally delete the plugin results, you can rerun the plugin and then view the plugin results. Click  Delete.

generateGenome plugin setup

The generateGenome plugin uses the following settings.

Setting	Description
The following settings are available for all library types.	
Library Type	The default value is the assay type selected during assay creation.
Sample Tracking	The Ion AmpliSeq™ Sample ID Panel is a companion panel of 9 primer pairs that can be added to any Ion AmpliSeq™ human gDNA panel during target amplification to generate a unique identification label for research samples. Select this checkbox if you added the Ion AmpliSeq™ Sample ID Panel to the assay.
The following settings are advanced options.	
Uniquely Mapped Reads	Select this option to analyze only reads that are mapped to a unique location in the reference. Reads that are non-uniquely mapped can have equally well-aligned reads that are mapped to multiple locations and are typically mapped randomly to one.
Non-duplicate Reads	Select the checkbox to avoid copy reads. The analysis must have included alignments with Mark Duplicates active.
Minimum Aligned Length	Specify the minimum aligned length that is required to ensure that the read is included in an analysis. The default value 0 specifies that no reads are filtered out due to the aligned length for the reads.
Minimum Mapping Quality	Specify a minimum value that reads must exceed to be included in the analysis. The default value 0 specifies that no reads are filtered out due to the quality of the alignment of the reads.

(continued)

Setting	Description
Tier 1 Coverage Depth	Specify the first-tier coverage depth at which percentage of target coverage is reported. This value must be at least 2, because the coverage depth output is always specified at 1x read depth. The default value of 20 process that the percentage of targets, total base targets, and/or separate target bases with at least 20 reads is reported.
Tier 2 Coverage Depth	Specify the second-tier coverage depth at which percentage of target coverage is reported. This value must be greater than the value used for the first-tier coverage. The default value of 350 process that the percentage of targets, total target bases, and/or separate target bases with at least 350 reads is reported.
Tier 3 Coverage Depth	Specify the third-tier coverage depth at which percentage of target coverage is reported. This value must be greater than the value used for the second-tier coverage. The default value of 500 process that the percentage of targets, total target bases, and/or separate target bases with at least 500 reads is reported.

Example generateGenome Report

The detailed **generateGenome Report** that is available when you select a barcode in the generateGenome plugin summary includes the following information about the sample.

The links at the bottom of the report includes a statistics file for consensus sequences, a report named **Variant calls per contig summary**, which summarizes the number of variant calls made by the software for a contig, compared to the reference genome for that contig, and a link to the consensus genome file in FASTA format.

In this example, there are only 9 variants in the sample genome compared to the genome reference. This reference had 11 contigs, including 10 control sequences. The **Percent N** reported largely reflects the targeted reference in this example, the 197KB viral contig itself had only 1.0375% 'N' padding (98.9625% sequenced), as shown in the **Variant calls per contig summary** file.

generateGenome Report

Sample Name: GenGenome_MPOX_1

GenGenome_MPOX_1_DNA144_1_Generate_genome_plugin_TC03_MPOX_Run02_Auto_R_2024_08_06_14_40_44_Gen

Reference: MonkeyPox_WG

Target regions: WG00616.20220817.designed.amplicon.overriding.withControl.finalUnmergedDetail

Reference Contigs	11
Total Variants	9
Homozygous SNPs	8
Homozygous InDels	1
Heterozygous SNPs	0
Heterozygous InDels	0
Other Variants	0
Output Contigs	11
Percent N	77.6814%

- [Consensus sequence summary statistics](#)
- [Variant calls per contig summary](#)
- [Download consensus genome \(FASTA\)](#)

Statistic	Description
Reference Contigs	Number of contigs (or chromosomes) in the reference (FASTA file).
Total Variants	The total number of variants that are detected by the Torrent Variant Caller module ^[1] The number of variants is used by the software to make edits to the input reference.
Homozygous SNPs	The number of homozygous SNPs. This information is used by the software to edit base changes to the input reference.
Homozygous InDels	The number of homozygous insertions and deletions. This information is used by the software to edit base changes to the input reference.
Heterozygous SNPs	The number of heterozygous SNPs. This information is used by the software to edit base changes to the input reference. These are represented by IUPAC multiallelic base codes in the consensus reference. For example, Y is the single base representation for a C/T allele.
Heterozygous InDels	The number of heterozygous insertions and deletions. This information is used by the software to edit base changes to the input reference. The output reference contains the allele that is represented by the majority of reads.
Other Variants	The number of other variants detected by the Torrent Variant Caller module ^[1] , typically a Multiple Nucleotide Polymorphism (MNP) and a Complex Structural Variant (CPX). That is, a general sequence substitution that is summarized as a deletion plus insertion.
Output contigs	The number of contigs output to the sample consensus genome (FASTA file).
Percent N	The percentage of the number of bases replaced by "N" in the output consensus sequence to its total length. These are sites that are not callable by the Torrent Variant Caller module ^[1] , typically due to the low depth of coverage.

^[1] The Torrent Variant Caller module evaluates the list of candidates and examines evidence for the variants in flow space.

Consensus sequence summary statistics

A summary of the statistics for the consensus file output (FASTA file) that is generated by the generateGenome plugin are shown in the screen when you click **Consensus sequence summary statistics** in the **generateGenome Report**.

Consensus sequence statistics

Statistic	Description
Sample name	The name of the sample in the sequencing run for which plugin results are shown.
Reference	The reference genome that is specified in the run plan, to which all reads are aligned.
Target Regions	The enriched regions of interest, if any.
Number contigs	The number of contigs (or chromosomes) in the reference (FASTA file).
Output contigs	The number of contigs output to the sample consensus genome (FASTA file). This number typically matches the number of reference contigs unless contig filtering options are specified, or because the sites were not targeted.

Consensus sequence statistics (*continued*)

Statistic	Description
Percent N	The percentage of the number of bases replaced by "N" in the output consensus sequence to its total length. These are sites that are not callable by Torrent Variant Caller module ^[1] , typically due to the low depth of coverage.
Sample Consensus	The name of the consensus sequence file that is available for download in the generateGenome plugin results.
Het indels	The number of heterozygous insertions and deletions detected by Torrent Variant Caller module ^[1] . The allele with most reads is used for the output consensus sequence.
Het snps	The number of heterozygous SNPs used to edit base changes to the input reference. These are represented by IUPAC multi-allelic base codes in the consensus reference. For example, Y is the single base representation for a C/T allele.
Homo indels	The number of homozygous insertions and deletions used to edit base changes to the input reference.
Homo snps	The number of homozygous SNPs used to edit base changes to the input reference.
Other Variants	MNP sequences are treated as adjacent SNPs in the consensus sequence output. Complex variants are treated like INDELs: The highest frequency allele detected is output to the consensus sequence
Contig Length	The length of the contig, in nucleotide bases.

^[1] The Torrent Variant Caller module evaluates the list of candidates and examines evidence for the variants in flow space.

Example variant calls per contig file generated by the generateGenome plugin

A table in XLS format is downloaded when you click **Variant calls per contig summary** in the **generateGenome Report** screen. The table is a summary of the number of variant calls made by the software for a contig, compared to the reference genome for that contig. For more information about the columns shown in the table, see “Consensus sequence summary statistics ” on page 421.

Example variant calls per contig table

Contig	Het indels	Het snps	Homo indels	Homo snps	Other	Variants	Contig Length	Percent N
ON563414.3	0	0	1	8	0	9	197201	1.0375
chrX_11314631_11315399	0	0	0	0	0	0	769	84.7854
chr7_137029407_137030178	0	0	0	0	0	0	772	84.3264
chr12_6945474_6946245	0	0	0	0	0	0	772	83.4197
chr5_178690302_178691059	0	0	0	0	0	0	758	84.8285
chr10_17192979_17193707	0	0	0	0	0	0	729	89.9863

Example variant calls per contig table (continued)

Contig	Het indels	Het snps	Homo indels	Homo snps	Other	Variants	Contig Length	Percent N
chr18_9749453_9750212	0	0	0	0	0	0	760	85.1316
chrY_6737588_6738362	0	0	0	0	0	0	775	84.129
chr3_193206956_193207719	0	0	0	0	0	0	764	84.8168
chr22_33559088_33559851	0	0	0	0	0	0	764	85.8639
chr4_169663235_169663986	0	0	0	0	0	0	752	86.1702

IonReporterUploader plugin

Sample results files that are generated in Genexus™ Software can be directly transferred to another Genexus™ Integrated Sequencer or to Ion Reporter™ Software. These transfers are done with the IonReporterUploader plugin.

Note: Ion Reporter™ Software is available under separate license and is not included with Genexus™ Software.

Ion Reporter™ Software performs analysis on unmapped BAM files that are output from Genexus™ Software. To transfer the BAM files to Ion Reporter™ Software, you must add one or more Ion Reporter™ Software accounts in Genexus™ Software. To configure an Ion Reporter™ Software account that you can use for data uploads, see “Register an Ion Reporter™ Server account” on page 356 and “Register an Ion Reporter™ Software on Connect account” on page 355.

After an account is configured, there are two ways that you can transfer files to Ion Reporter™ Software.

- When you plan a run, you can select an option to have Genexus™ Software directly transfer output files to Ion Reporter™ Software. For more information, see “Plan a Sample to Result run” on page 89, “Plan a Nucleic Acid to Result run” on page 97, and “Plan a Library to Result run” on page 103.
- You can upload data manually to Ion Reporter™ Software. Use this option if, for example, you want to upload data to multiple Ion Reporter™ Software servers. For more information, see “Upload results files to Ion Reporter™ Software” on page 196.

Before you can upload results files to another sequencer, you must register the account that you can use for data uploads. For more information, see “Register Genexus™ Software accounts” on page 354.

After an account is configured, you can transfer sample results files in BAM or CRAM format to another Genexus™ Integrated Sequencer in the following two ways.

- When you plan a run, select the **Upload BAM/CRAM files to Server** checkbox in the **Setup** step of when you plan a run to directly transfer sample results files to another sequencer.
- Upload sample results files manually to another sequencer. For more information, see “Upload results files to another sequencer” on page 195.

Review IonReporterUploader plugin results

After the sequencing run is complete, review the plugin results. You can view the information that is related to the data transfer.

If you are transferring sample results to Ion Reporter™ Server, the information includes the name of the Ion Reporter™ Server used, the version of Ion Reporter™ Software, the folders that contain the uploaded files, and the software organization and user account that was used.

If you are transferring sample results to another Genexus™ Integrated Sequencer, the information includes the name of the Genexus™ Integrated Sequencer used, the version of Genexus™ Software, the folders that contain the uploaded files, and the software organization and user account that was used.

You can also review details about barcoded samples that were uploaded with the IonReporterUploader plugin, or samples that failed to upload.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run of interest.
3. In **Select Assay**, select the assay of interest.
4. Click the **Plugins** tab.
A summary table of the IonReporterUploader plugin results is shown.
5. In the IonReporterUploader plugin results, view the information that is related to the data transfer.

To do this	Click
View error messages associated with the plugin run.	Errors
View warnings that contain details about the barcoded samples that are used in the plugin run.	Warnings
Show or hide a detailed status of the pre- and post-processing of the data transfer.	Show detailed status/Hide detailed status
Open the plugin log files in a separate browser tab.	 View Log
Download a CSV file that contains a list of samples that are uploaded.	Download CSV file that contains a list of samples
Download a compressed folder of the IonReporterUploader plugin log files and other plugin files.	Download IRU logs
Delete the plugin results.	 Delete

molecularCoverageAnalysis plugin in Genexus™ Software

Use the molecularCoverageAnalysis plugin to view statistics and graphs that describe the level of sequence molecular coverage produced for targeted genomic regions. The plugin generates statistics, downloadable data files, and interactive visualizations of molecular coverage over targeted regions of the reference genome.

The plugin is compatible only with Ion AmpliSeq™ HD library chemistry.

Review molecularCoverageAnalysis plugin results

The molecularCoverageAnalysis plugin generates a Molecular Coverage Analysis Report. This report includes molecule statistics and several charts. The report summary lists the barcodes, the samples, the median molecular coverage, the molecular uniformity, and the median reads per functional molecule, and the median percentage of functional reads. Details regarding molecular coverage are also provided on a per-barcode basis, along with a list of gene annotations for each sequenced region.

You can download statistics files from the file links at the bottom of the results screen when the **Plugins** tab is selected. After the sequencing run completes, review the plugin results in the report summary.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.
3. Click the **Plugins** tab.
Plugin Results is selected and a summary table of the molecular coverage analysis, by barcode, is included in the **molecularCoverageAnalysis** summary pane.
4. (Optional) Click the link next to **Parameters** to download a JSON file that shows the parameter settings for the plugin.
5. In the **molecularCoverageAnalysis** summary pane, click the barcode name of interest in the **Barcode Name** column to view a detailed **Molecular Coverage Analysis Report** for a sample.
6. Click the links at the bottom of the **Molecular Coverage Analysis Report** to download associated statistics and summary files for each barcoded sample in the run.

Option	Description
Download the coverage statistics summary file.	Text summary of the statistics presented in the tables at the top of the Molecular Coverage Analysis Report .
Download the amplicon molecular coverage summary file.	Amplicon molecular summary table that is used to create the Amplicon molecular coverage chart shown in the Molecular Coverage Analysis Report .
Links to targets (BED) file upload page.	Link to the target regions upload page where you can review and download the BED file that defined the regions of the reference analyzed in the Molecular Coverage Analysis Report .
Download the zip report.	ZIP file that contains a PDF of the current report page and primary coverage analysis files.

7. Use the options to view more reports, view the log for the plugin run, or delete the plugin output.

Option	Description
View more reports.	In Executed At , select an alternate timestamp, if available.
Rerun the molecular Coverage Analysis plugin.	Click  Select Plugins and select the coverageAnalysis plugin, then click Rerun Plugin .
View the molecular Coverage Analysis log.	Click  View Log .
Delete the molecular Coverage Analysis plugin output for the selected timestamp.	IMPORTANT! When you click Delete, a confirmation message is not shown and the delete is immediate. Ensure that you want to delete before you click Delete. If you accidentally delete the plugin results, you can rerun the plugin and then view the plugin results. Click  Delete.

Example Molecular Coverage Analysis Report

The following is an example of a **Molecular Coverage Analysis Report** that is available when you select a barcode in the molecularCoverageAnalysis plugin summary.

Molecular Coverage Analysis Report

Sample Name:

Library type: AmpliSeq HD - DNA
Reference: hg19 (DNA)
Target regions:

Number of amplicons	272
Median functional molecules per amplicon	2.521
Uniformity of amplicon molecular coverage	90.81%
Amplicons with LOD below 5%	99.26%
Amplicons with LOD below 1%	95.59%
Amplicons with LOD below 0.5%	85.66%
Amplicons with LOD around 0.1%	1.47%
Median number of reads per molecule	26.8
Median molecular loss due to strand bias	78.90%
Median reads to molecular conversion rate	80.56%

Molecule statistics

The detailed **Molecular Coverage Analysis Report** that is available when you select a barcode in the coverageAnalysis plugin summary for a run includes a variety of reads statistics. The statistics that are displayed in your report depend on the type of library that is used in your sequencing experiment. Definitions are in tooltips. Almost every statistic, plot, link, and functional widget in the report provides tooltips with definitions. Place the pointer over a heading or description in the report to view the tooltip.

Statistic	Description
Median Functional Molecular Coverage per Amplicon	A functional molecule is a collection of reads that covers the amplicon and satisfies all criteria associated with the parameters (for example, <code>min_tag_fam_size</code> and <code>min_fam_per_strand_cov</code>) in the parameter file. Molecular coverage is the number of functional molecules. The median is calculated across all amplicons.
Uniformity of Molecular Coverage for all Amplicons	The percentage of amplicons having molecular coverage between 0.5x and 2x of the median molecular coverage.
Percentage of Amplicons larger than 0.8x Median Functional Molecular Coverage	The percentage of amplicons having molecular coverage more than 0.8x of the median functional molecular coverage
Median Total Molecular Coverage per Amplicon	The number of molecules that satisfies size criteria in the parameters file. The median is calculated across all amplicons.
Percentage of Reads with Perfect Molecular Tags	The percentage of reads whose molecular tags are exactly the same with design.
Median Functional Molecular Loss due to Strand Bias per Amplicon	The metric is calculated by $1 - (\text{Functional Molecular Coverage}) / (\text{Molecular Coverage without Strand Constraint})$, which measures the loss of functional molecules due to strand constraint for molecular functionality. The median is calculated across all amplicons. This statistic is available in Ion AmpliSeq™ HD runs only.
Median Percentage of Functional Molecules out of Total Molecules per Amplicon	The percentage of functional molecules out of the number of molecules that satisfies size criteria. The median is calculated across all amplicons.
Median Reads per Functional Molecule	For each amplicon, the number of reads supporting each functional molecule is averaged across all functional molecules for that amplicon to determine the number of reads per functional molecule at the amplicon level. The median is calculated across all amplicons.
Median Reads Contributed to Functional Molecules per Amplicon	For each amplicon, the percentage of reads supporting functional molecules is $(\text{Number of Reads Supporting Functional Molecules}) / (\text{Number of Reads})$. The median is calculated across all amplicons.
Percentage of Amplicons below (around) n% LOD	LOD is calculated based on the number of functional molecules for each amplicon and variant calling parameters in the parameter file. Around x n% means in the range from 0.5x n% to 2x n%.

Example charts generated by the molecularCoverageAnalysis plugin

The charts in the detailed **Molecular Coverage Analysis Report** include **Plot** and **Overlay** menus that allow you to customize the data that is displayed in each chart.

Click **Q (Search)** (in the top right corner of a chart) to open the chart **Viewing Options** panel. Click **? (Help)** to open a description of the chart.

The Depth of Coverage Chart summarizes the amplicon depth of coverage results. The X-axis represents the amplicon molecular depth. The left Y-axis represents the number of amplicons at a given molecular depth or a range (bin) of molecular depths, as a percentage of the total number of amplicons. The right Y-axis represents the cumulative count of the number of amplicons, at a given molecular depth or greater, as a percentage of the total number of amplicons. The individual orange bars represent the percentage of amplicons in the specific range of molecular depths. The blue curve measures the cumulative amplicons at a given molecular depth or greater. Use the **Plot** dropdown list to switch between **Maximum Read Depth**, **99.9% of All Reads**, and **Normalized Coverage** plots.

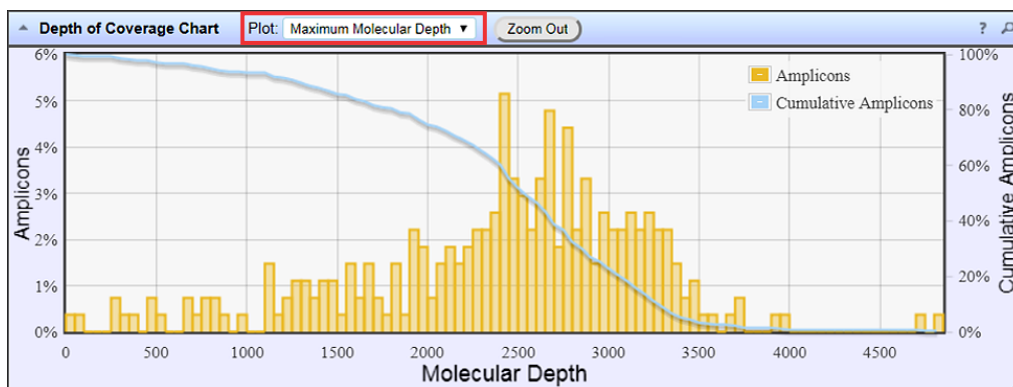


Figure 9 Representative Depth of Coverage Chart

The Amplicon Coverage charts summarize the amplicon molecular coverage results. The X-axis in all plots represents amplicons that are grouped into bins, where each bin contains amplicons that have the same molecular coverage. Based on the plot selection, the left Y-axis represents either the average number of molecules (on a base 10 log scale) or the percentage of functional molecules in a given bin. The right Y-axis represents the percentage of functional reads in a given bin. Use the **Plot** and **Overlay** dropdown lists to customize the chart view. To zoom in on a subset of amplicons, use the pointer to draw a rectangle that contains the region of interest. Use **Zoom Out** to revert to the initial view.

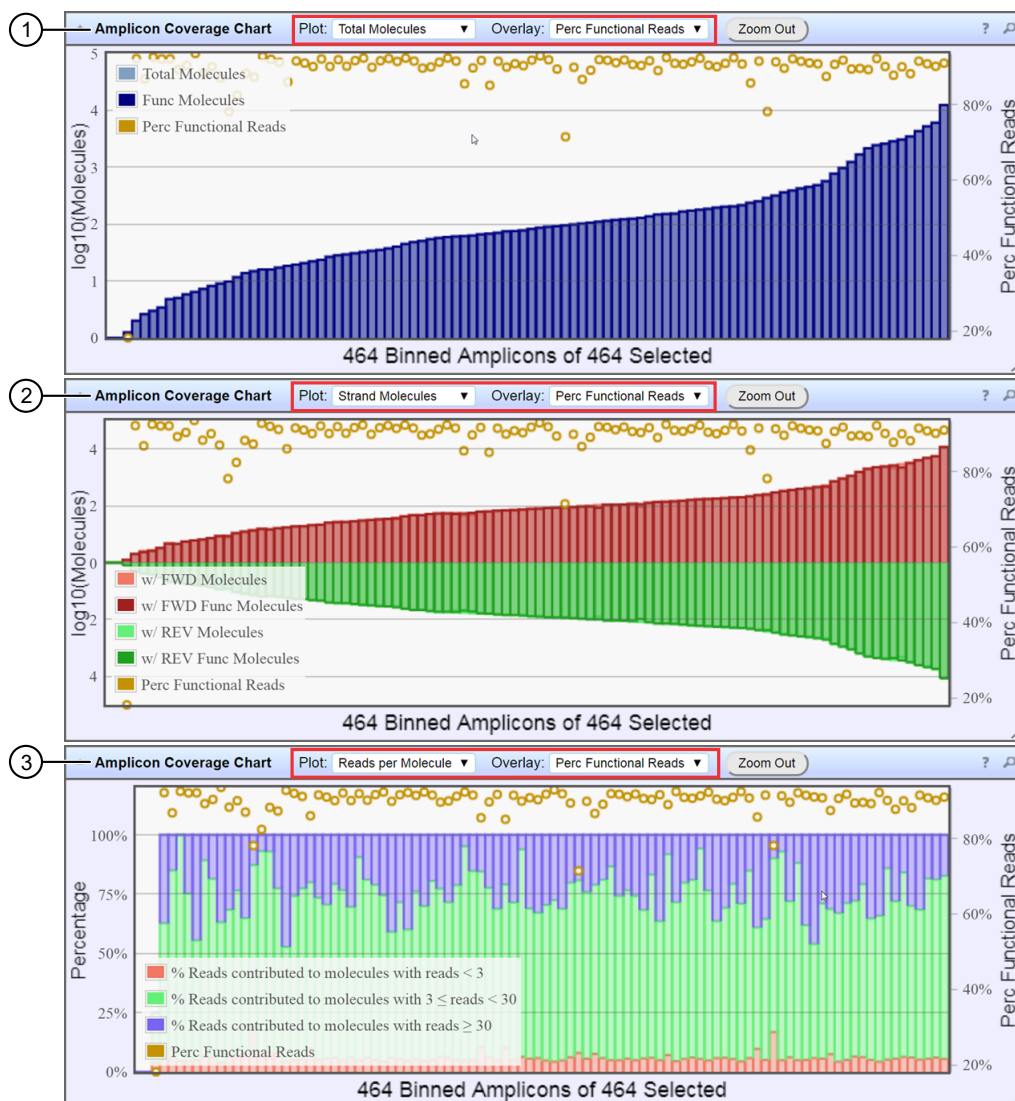


Figure 10 Representative Amplicon Coverage charts

- ① Representative **Total Molecules** plot with a **Perc Functional Reads** overlay.
- ② Representative **Strand Molecules** plot with a **Perc Functional Reads** overlay.
- ③ Representative **Reads per Molecule** plot with a **Perc Functional Reads** overlay.

In most plots, you can click a data point to open a detail pane for that data (see Figure 11). For example, in the Depth of Coverage Chart in Figure 9, click an individual orange bar to open the detail pane for amplicons in a specific range of molecular depths, or click a point on the blue curve to open the detail pane for cumulative amplicons at that molecular depth or greater.

Similarly, you can click any point or bar within each Amplicon Coverage chart to view details for each amplicon coverage bin.

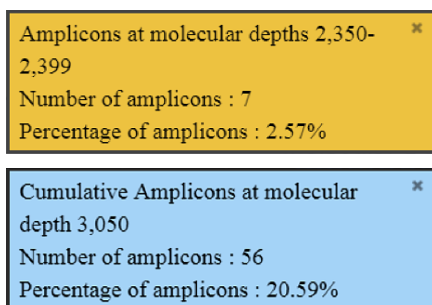


Figure 11 Example detail panes

Output files generated by the molecularCoverageAnalysis plugin

You can download plugin results files from the **Results** screen for a sample. For more information, see “Results files” on page 178.

Note: Sometimes the file name can be too long to open in applications such as Microsoft™ Excel™. To resolve this problem, right-click the file, then click **Save As** to rename the downloaded file.

The following tables describe the results files that can be generated by the molecularCoverageAnalysis plugin. The list of files depends on the type of assay template that was selected during assay creation.

File download selections in the Genexus™ Software

Selection	File name
TNA Molecular Coverage Statistics	<Barcode><ExpName>.cov.stats.txt
TNA Molecular depth of coverage	<Barcode><ExpName>.mol.cov.xls
TNA Molecular Coverage Analysis Summary	<Barcode><ExpName>.summary.pdf
TNA Molecular Amplicon coverage summary	<Barcode><ExpName>.amplicon.cov.xls

File contents

File	Description
Molecular Coverage Statistics	This file contains a summary of the statistics presented in the tables at the top of the Molecular Coverage Analysis Report plugin report. The first line is the title. Each subsequent line is either blank or a particular statistic title followed by a colon (:) and its value.
Molecular depth of coverage	Molecular coverage summary data used to create the Depth of Coverage Chart. This file contains the following fields: - mol_depth—The depth at which a (targeted) reference amplicon has been read. - amp_cov—The number of times any amplicon was read (covered) at this depth. - amp_cum_cov—The cumulative number of reads (coverage) at this read depth or greater. - norm_mol_depth—The normalized read depth (depth divided by average molecular read depth). - pc_amb_cum_cov—The same as amp_cum_cov but represented as a percentage of the total reads.

File contents (continued)

File	Description
Molecular Coverage Analysis Summary	A PDF file that contains the Molecular Coverage Analysis Report, including molecule statistics and charts that are generated by the molecularCoverageAnalysis plugin.
Molecular Amplicon coverage summary	Amplicon molecular coverage summary data used to create the Amplicon Coverage Chart. This file contains the following fields: - contig_id—The name of the chromosome or contig (from contiguous) of the reference for this amplicon. - contig_srt—The start location of the amplicon target region. Note that this coordinate is 1-based, unlike the corresponding 0-based coordinate in the original targets BED file. - contig_end—The last base coordinate of this amplicon target region. Note that the length of the amplicon target is given as (contig_end - contig_srt + 1). - region_id—The ID for this amplicon as given as the 4th column of the targets BED file. - gene_id or attributes—The gene symbol or attributes field as provided in the targets BED file. - func_mol_cov—Functional molecular coverage utilized for variant calling. - lod—LOD (limit of detection) calculated from the number of functional molecules. - strict_func_omt_rate—The percentage of functional molecules used with strict molecular tags. - func_mol_cov_loss_due_to_strand—The percentage of functional molecules not formed. - fwd_only_mol_cov—The number of molecules containing forward strand only. - rev_only_mol_cov—The number of molecules containing reverse strand only. - both_strands_mol_cov—The number of molecules containing both forward strand and reverse strand. - r2m_conv_rate_all—The percentage of reads contributed to functional molecules. - reads_per_func_mol—The average reads per functional molecules. - perc_to_mol_(<3_reads) —The percentage of reads contributed to small size molecules (size <3). - perc_to_mol_(≥3&<30_reads) —The percentage of reads contributed to median size molecules (size ≥3 && size <30). - perc_to_mol_(≥30_reads) —The percentage of reads contributed to large size molecules (size ≥30).

sampleID plugin

Use the sampleID plugin to track samples or possibly identify misassignment or mix up between samples and barcodes in a sequencing run. The sampleID plugin produces a unique identification code (**SampleID**) for each barcode in a sample.

The sampleID plugin can be run on sequencing results from any panel that includes the 9 primer pairs of the Ion AmpliSeq™ Sample ID Panel.

For example, the Ion AmpliSeq™ Sample ID Panel contains 9 primer pairs that can be combined with any Ion AmpliSeq™ Ready-to-Use Panel or Custom Panel. The sampleID plugin can be used with the Ion AmpliSeq™ Sample ID Panel, which is a human SNP genotyping panel, to help ensure sample accuracy and increase confidence in sample data management. The Ion AmpliSeq™ Sample ID Panel is composed of the identified human sample gender and International Union of Pure and Applied Chemistry (IUPAC) base letters for eight high-frequency noncoding SNPs.

For the samples to work with this plugin, the Ion AmpliSeq™ library must have been prepared with Ion AmpliSeq™ sample tracking amplicons.

The sampleID plugin is preconfigured and does not require user input.

Review sampleID plugin results in Genexus™ Software

After the sequencing run completes, review the plugin results.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.
3. Click the **Plugins** tab.
A summary table of the **sampleID** results is shown.
4. Click a barcode name to open the **Sample ID Report**.
5. To return to Genexus™ Software, click back in the browser.

Sample ID Report

The **Sample ID Report** that is available when you select a barcode in the **SampleID** plugin summary for a run includes a variety of read statistics.

Definitions are in tooltips. Almost every statistic, link, and functional widget in the report provides a tooltip with definitions. Place the pointer over a heading or description in the report to view the tooltip.

Statistic	Description
Number of mapped reads	The total number of reads mapped to the reference genome.
Number of reads in sample ID regions	The total number of reads mapped to Sample ID regions.
Percent reads in sample ID regions	The percent of all reads aligned to the reference that covered bases in Sample ID regions.
Total base reads in sample ID regions	The total base reads aligned to the reference that covered bases in Sample ID regions.
Percent base reads in sample ID regions	The percent of all bases covered by reads aligned to the reference that covered bases in Sample ID regions.
Male sample ID region reads	Number of reads mapped to the specific male (Y) identification target or targets.
Female sample ID region reads	Number of reads mapped to the specific female (X) identification target or targets.
Sample ID Regions and Sample ID SNPs	
Bases in target regions	The total number of bases in all specified target regions of the reference.
Average base coverage depth	The average number of DNA reads of all targeted reference bases.
Uniformity of coverage	The percentage of target bases covered by at least 0.2x of the average base coverage depth.
Coverage at 1x	The percentage of target bases covered by at least 1 read.
Coverage at 20x	The percentage of target bases covered by at least 20 reads.
Coverage at 100x	The percentage of target bases covered by at least 100 reads.

Output files generated by the sampleID plugin

You can download sampleID plugin results files from the **Results** screen for a sample. For more information, see “Results files” on page 178.

Note: Sometimes the file name can be too long to open in applications such as Microsoft™ Excel™. To resolve this problem, right-click the file and click **Save As** to rename the downloaded file.

page 434 describes the files that can be generated by the sampleID plugin. The list of files depends on the application type that was selected during assay creation.

File download selections in the Genexus™ Software

Selection	File Name
Tracking Loci Regions (SNPs) File	tracking_loci.bed
Aligned Tracking Reads Index (BAI) File	<DNA Barcode><ExpName>.bam.bai
Hard-copy A4 PDF Image Of This Report Page	<DNA Barcode><ExpName>.sampleID.pdf
Tracking Target Regions File	tracking_regions.bed
Aligned Tracking Reads (BAM) File	<DNA Barcode><ExpName>.bam
All Variant Calls As a Table File	allele_counts.xls



Genexus™ Software API

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Genexus™ Software has a web services API that allows you to automate the retrieval of key information from the system.

This section provides examples to show you how to leverage the API.

Genexus™ Software API introduction

Genexus™ Software integrates with laboratory information management systems (LIMS) to help with sample, library, and run plan creation. The interaction is through the LIMS calling a set of RESTful APIs provided by the software. The LIMS connects to the software by making an HTTP call to the URL of an API, and sending a user name and a password in the HTTP header. The user name and password must be for a registered active user account of the software. The LIMS passes the input information in JSON format within the HTTP body, while the response from the software is returned in JSON format as well. Users must configure their LIMS to create JSON files containing the required attributes for each API request file.

Outlined in the following table are the API names, functions, and URLs of the RESTful APIs that are provided in the Genexus™ Software API.

API	Function	API URL
Create a library batch For more information, see “Create a library batch” on page 439.	Create a library batch from existing samples.	<code>https://<host>/genexus/api/lims/v2/library-batches</code>
Create a library run For more information, see “Create a library run” on page 440.	Create a library run from one or more library batches and an assay.	<code>https://<host>/genexus/api/lims/v2/plan-by-library</code>
Create a Nucleic Acid to Result run For more information, see “Create a Nucleic Acid to Result run” on page 440.	Create a Nucleic Acid to Result run from one or more samples and an assay.	<code>https://<host>/genexus/api/lims/v2/plan-by-samples</code>
Create a Sample to Result (purification) run For more information, see “Create a Sample to Result (purification) run” on page 441.	Create a Sample to Result run from one or more samples and an assay.	<code>https://<host>/genexus/api/lims/v2/plan-by-purification</code>
Create samples For more information, see “Create samples” on page 442.	Create samples.	<code>https://<host>/genexus/api/lims/v2/samples</code>
Download files for completed runs For more information, see “Download files for completed runs” on page 443.	Returns a compressed file in ZIP format that contains the requested files for the sample.	<code>https://<host>/genexus/api/lims/v2/download</code>
Get a list of Ion Reporter™ Software accounts For more information, see “Get list of Ion Reporter™ Software accounts” on page 444.	Returns a list and the details of active Ion Reporter™ Software accounts.	<code>https://<host>/genexus/api/lims/v2/ir-accounts</code>
Get report templates For more information, see “Get report templates” on page 445.	Returns report templates.	<code>https://<host>/genexus/api/lims/v2/report-templates</code>

(continued)

API	Function	API URL
Get a list of assays For more information, see “Get assay list” on page 446.	Returns a list of the assays in the system. PQ assays are excluded.	https:// <host>/genexus/api/lims/v2/assays
Get a list of samples For more information, see “Get samples” on page 447.	Returns a list of the samples in the system.	https:// <host>/genexus/api/lims/v2/samples
Get a list of library batches For more information, see “Get library batches” on page 448.	Returns a list of the library batches in the system.	https:// <host>/genexus/api/lims/v2/library-batches
Get a list of run plans For more information, see “Get run plans” on page 450.	Returns a list of available run plans in the system.	https://<host>/genexus/api/lims/v2/plans
Get results For more information, see “Get results” on page 451.	Returns results, including plugins and plugin results. Returns a JSON object for each plugin. All plugins for the run and the sample are returned.	https:// <host>/genexus/api/lims/v2/results
Get a list of signed off samples For more information, see “Get signed off samples” on page 454.	Returns a list of the samples that are designated with (Signed Off).	https:// <host>/genexus/api/lims/v2/signedOffSamples

API common parameters

If the JSON download process converts @ symbols to %40, do not change the text. Instead, copy the JSON files as is into the browser.

Parameter	Entry
BASE_URL	https://<host>/genexus/api/lims/v2
Request	Authorization:Basic Accept:application/json
Response	Content-Type:application/json

Response fields

Code	Status
200	Successful response
400	Bad request Error response: <pre>{ "errors": [{ "field": "validation error" }] }</pre>
401	Unauthorized
500	Internal Server Error

Get requests

Type of request	Entry
Get list of resources	BASE_URL/resource_url/
Get single object	BASE_URL/resource_url?id=object_id

Common request parameters

Parameter	Entry	Value
Starting page number	pageNumber	The default value is "1".
Number of items to fetch ^[1]	pageSize	The default value is "20".
A field based on which, the result set needs to be ordered	orderBy	Only specific values are allowed. If no value is provided, the result set is ordered by ID.
Order direction of the result set	sortOrder	Valid values are "asc" or "desc". The default value is "asc".

^[1] You can limit the number of items that are fetched when you add parameters to the request. For example, add "status" to the get assay list request.

Create a library batch

library-batches	
URL	BASE_URL/library-batches
Description	Create a library batch from one or more existing samples. If any requested sample does not exist in the software, the creation of the entire library batch fails.
Method	POST
Header	curl -k --header 'username:<user name>' --header 'password:<user password>' --header 'Content-Type: application/json'
Example URL	curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/library-batches'
Example request	<pre>{ "objects": [{ "libraryBatchId": "LibBatch_DNA_NTC_01", "assayName": "Oncomine Comprehensive v3 - GX5 - DNA - w5.0.2", "libraryPrepType": "DNA", "libraryKitBarcode": "01ABC0012300000017221123102-321AB234240A40252", "panelKitBarcode": "01ABC0012300000017221123102-321AB234240A44351", "libraries": [{ "barcodeID": "IonDual_0101", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "DNA_Lib1_99", "sampleId": "Test_Sample1_127", "extractionKitBarcode": "9110009055910123456717211019" }, { "barcodeID": "IonDual_0102", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "DNA_Lib2_99", "sampleId": "Test_Sample1_127", "extractionKitBarcode": "9110009055910123456717211019" }, { "barcodeID": "IonDual_0103", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "", "sampleId": "NTC", "controlKitBarcode": "9110008037510224548317211230" }, { "barcodeID": "IonDual_0104", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "", "sampleId": "NTC", "controlKitBarcode": "9110008037510224548317211230" }] }] }</pre>



Create a library run

plan-by-library	
URL	BASE_URL/plan-by-library
Description	Create a library run from one or more library batches and an assay. The library batch or batches and the assay must exist in the system.
Method	POST
Header	curl -k --header 'username:<user name>' --header 'password:<user password>' --header 'Content-Type: application/json'
Example URL	curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/plan-by-library'
Example request	<pre>{ "irAccountName": "iraccount", "samples": [{ "assayName": "Generic Sequencing Ampliseq Assay", "libraryBatchId": "LibraryBatch1" }, { "assayName": "Custom DNA Assay", "libraryBatchId": "LibraryBatch2" }, { "assayName": "Custom DNA Assay", "libraryBatchId": "LibraryBatch3" }], "notes": "my notes", "planName": "plan_library_run" }</pre>

Create a Nucleic Acid to Result run

plan-by-samples	
URL	BASE_URL/plan-by-samples
Description	Create a Nucleic Acid to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system.
Method	POST
Header	curl -k --header 'username:<user name>' --header 'password:<user password>' --header 'Content-Type: application/json'

(continued)

plan-by-samples	
Example URL	<code>curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/plan-by-samples'</code>
Example request	<pre>{ "irAccountName": "iraccount", "reportTemplateName": "mytemplate", "samples":[{ "assayName": "Generic Sequencing Ampliseq Assay", "sampleId":"Sample1" }, { "assayName": "Custom DNA Assay", "sampleId":"Sample2" }, { "assayName": "Custom DNA Assay", "sampleId":"Sample3" },], "notes":"my notes", "planName":"plan_sample_run" }</pre>

Create a Sample to Result (purification) run

plan-by-purification	
URL	<code>BASE_URL/plan-by-purification</code>
Description	Create a Sample to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system.
Method	POST
Header	<code>curl -k --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json'</code>



(continued)

plan-by-purification	
Example URL	<code>curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/plan-by-purification'</code>
Example request	<pre>{ "irAccountName": "iraccount", "reportTemplateName": "mytemplate", "samples": [{ "assayName": "Purification FFPE DNA and Fusion Assay", "sampleId": "Sample1", "purificationProtocol": "FFPE DNA + RNA", "elutionVolumeDNA": 50, "elutionVolumeRNA": 50, "pauseForReview": false }, { "assayName": "Purification DNA Assay", "sampleId": "Sample2", "purificationProtocol": "Blood 50-100 µl", "elutionVolumeDNA": 40, "pauseForReview": false }, { "assayName": "Purification DNA Assay", "sampleId": "Sample3", "purificationProtocol": "Blood 50-100 µl", "elutionVolumeDNA": 40, "pauseForReview": false }, { "assayName": "Purification RNA Assay", "sampleId": "Sample4", "purificationProtocol": "Biofluid", "elutionVolumeDNA": 90, "pauseForReview": false }, { "assayName": "Purification RNA Assay", "sampleId": "Sample5", "purificationProtocol": "Cells and tissue", "elutionVolumeDNA": 90, "pauseForReview": false }], "notes": "my notes", "planName": "plan_purification_run" }</pre>

Create samples

samples	
URL	BASE_URL/samples
Description	Create 1 to <i>n</i> samples. "All" or "None" is followed for all samples in a request. If the creation of any one sample fails, creation of the entire batch fails.
Method	POST
Header	<code>curl -k --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json'</code>

(continued)

samples	
Example URL	<code>curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/samples'</code>
Attribute definitions	• <code>sampleId</code>. String. Required. Sample name. • <code>sampleType</code>. String. Required. Examples of valid values are "DNA" or "RNA". • <code>collectionDate</code>. String. Required. The date that the sample was collected. • <code>gender</code>. String. Required. The biological sex of the sample: Female, Male, or Unknown. • <code>diseaseCategory</code>. String. Required. The sample application category. • <code>cancerStage</code>. String. Required. Cancer stage. • <code>origin</code>. String. Required. Cancer type. • <code>notes</code>. String. Required. An open-entry field for more sample information. • <code>necrosis</code>. String. Optional. The percentage of cellular necrosis in the sample. • <code>tumorCellularity</code>. String. Optional. The percentage of tumor cells over normal cells in the sample. • <code>condition</code>. String. Optional. The condition of the sample.
Example request	<pre>{ "objects":[{ "sampleId":"Sample_NSCLC_Tag_{{\$randomInt}}", "diseaseCategory":"Solid Tumor", "sampleType":"DNA & RNA", "origin":"Non-Small Cell Lung Cancer", "cancerStage":"Unknown", "necrosis":100, "tumorCellularity":31, "gender":"Unknown", "collectionDate":"2018-10-02", "condition":"good", "tagDisplayName":["NSCLC"], "notes":"LIMS API created Cancer sample and existing Tag NSCLC" }] }</pre>

Download files for completed runs

download	
URL	<code>BASE_URL/download</code>
Description	Returns a compressed file in ZIP format that contains the requested files for the sample.
Method	GET
Header	<code>curl -k --header 'username:<user name>' --header 'password: <user password>'</code>

(continued)

download	
Example URL	<code>curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/download'</code>
Request query parameters	<code>file_list</code> String. <i>Required.</i> Provide the file name in comma separated format. <code>path</code> String, <i>Required.</i> Provide the LIMS folder path of the analyzed sample. You can request the folder path when you get signed off samples. The folder path is listed as the "base_URL". For more information, see "Get signed off samples" on page 454.
Example request	- Perform a checksum function to query for the list of files for a sample that has been sequenced and analyzed. <code>https://BASE_URL/download?file_list=CHECKSUM&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q</code> - Parse the checksum to get the file name, then request that file. <code>https://BASE_URL/download?file_list=Info.csv,BaseCaller.json,analysis.log&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q</code> - Ensure that the name of the downloaded file matches the checksum requested in step 1.

Get list of Ion Reporter™ Software accounts

accounts	
URL	<code>BASE_URL/ir-accounts</code>
Description	Returns a list and the details of active Ion Reporter™ Software accounts.
Method	GET
Header	<code>curl -k --header 'username:<user name>' --header 'password: <user password>'</code>
Example request	<code>curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://server/genexus/api/lims/v2/ir-accounts'</code>

(continued)

accounts	
Request query parameters	irversion String. <i>Mandatory</i>. Ion Reporter™ Software version.
Example response	<pre>{ "total": 2, "objects": [{ "id": "2", "displayName": "TFAccount_Local_516 (Default)", "serverType": "HTTPS", "port": "443", "state": "Active", "userName": "firstname.lastname@tfc.com", "org": "IR Org", "status": "ENABLED", "server": "<servername>", "irVersion": "IR516", "defaultAccount": true, "accountType": "Ion Reporter Local", "usedAccount": false, "irAccountListError": false, "irAccount": true, "createdBy": "AutolAdmin" }] }</pre>

Get report templates

report-templates	
URL for a list of resources	BASE_URL/report-templates
URL for a single object	BASE_URL/report-templates
Description	Returns report templates. API supports multiple filters.
Method	GET
Header	curl -k --header 'username:<user name>' --header 'password: <user password>'
Example URL	curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lms/v2/report-templates'



(continued)

report-templates	
Request query parameters	assayName String. <i>Optional</i>. Get the report template for the given assay. Use the Assay Name. Do not use the Assay Full Name.
Example response	<pre>{ "mode": "AssayDev", "reportingTemplate": [{ "name": "default", "description": "Default", "locale": "en" }, { "name": "oca3_dr_oncominepositive", "assayName": "Oncomine Comprehensive v3 - GX5 - DNA and Fusions - w5.0.2", "assayId": 50, "description": "OCA3_DR_OncominePositive", "locale": "en" }], "total": 1 }</pre>

Get assay list

assays	
URL	BASE_URL/assays
Description	Returns a list of the assays in the system. PQ assays are excluded.
Method	GET
Header	curl -k --header 'username:<user name>' --header 'password:<user password>'
Example URL	curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/assays'

(continued)

assays	
Supported parameters	- assayName String. <i>Optional</i>. Assay name. - status String, <i>Optional</i>. Valid values are "draft", "obsolete", or "locked".
Example response	<pre>{ "total": 1, "objects": [{ "id": 122, "assayName": "copy_of_OCAv3_GX5_DNA_RNA_w5.0.2", "assayShortName": "OCAv3 DNA Fus w5.0.2", "assayDisplayedName": "Oncomine Comprehensive v3 - GX5 - DNA and Fusions - w5.0.2", "application": "DNA+RNA", "createdBy": "System Installed", "createdDate": "2021-06-22T16:50:15.000+0000", "isReanalysisAssay": false, "genomeReference": "hg19", "targetRegions": "OCAv3_w5.0.2_dna_fusions_20200626.GX5.designed.bed", "hotspots": "OCAv3_w5.0.2_dna_fusions_20201117.GX5.hotspots.bed", "fusionReference": "OCAv3_w5.0.2_designs_072720_Reference.fasta", "fusionTargetRegions": "OCAv3_w5.0.2_dna_fusions_designs_072720_Reference.bed", "chipType": "Ion Torrent™ GX5™ Chip", "panelKit": "Oncomine™ Comprehensive Assay v3 DNA and RNA, GX", "controlKit": "Other", "extractionKit": "Ion Torrent™ Genexus™ FFPE DNA/RNA Purification Combo Kit", "libraryKit": "Ion Torrent™ Genexus™ Library Strips 1 and 2-AS", "templatingKit": "Ion Torrent™ Genexus™ Templating Strips 3-GX5 and 4", "barcodeKit": "Ion Torrent™ Genexus™ Barcodes 1-96 AS", "sequencingKit": "Ion Torrent™ Genexus™ Sequencing Kit", "flows": 400, "templatingSize": 200, "status": "Locked", "tumorCellularity": "1", "reanalysisAssay": false }] }</pre>

Get samples

samples	
URL	BASE_URL/samples
Description	Returns a list of the samples in the system.
Method	GET
Header	curl -k --header 'username:<user name>' --header 'password: <user password>'
Example URL	curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/samples'



(continued)

samples	
Request query parameters	sampleId String. <i>Optional</i>. Sample name.
Example response	<pre>{ "total": 1, "objects": [{ "otherAttributes": {}, "id": 554, "sampleId": "Test_Sample1_127", "collectionDate": "2018-10-02", "gender": "Male", "sampleType": "DNA", "tumorCellularity": "70", "origin": "Non-Small Cell Lung Cancer", "cancerStage": "Stage 0", "diseaseCategory": "Solid Tumor" }] }</pre>

Get library batches

library-batches	
URL	BASE_URL/library-batches
Description	Returns a list of the library batches in the system.
Method	GET
Header	curl -k --header 'username:<user name>' --header 'password:<user password>'
Example URL	curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/library-batches'

(continued)

library-batches	
Request query parameters	id String. <i>Required</i>. Library batch ID.
Example response	<pre>{ "total": 1, "objects": [{ "libraries": [{ "libraryType": "DNA", "barcodeID": "IonHDDual_0102", "libraryName": "NTC_DNAOPA_DNA_Fusion_Library_Bacth_013", "inputQuantity": 3.0, "sampleId": "NTC", "tumorCellularity": null, "controlKitBarcode": "9110008037510224548317211230", "extractionKitBarcode": null }, { "libraryType": "RNA", "barcodeID": "IonHDDual_0104", "libraryName": "NTC_RNAOPA_DNA_Fusion_Library_Bacth_014", "inputQuantity": 3.0, "sampleId": "NTC", "tumorCellularity": null, "controlKitBarcode": "9110008037510224548317211230", "extractionKitBarcode": null }, { "libraryType": "DNA", "barcodeID": "IonHDDual_0101", "libraryName": "DNA_01", "inputQuantity": 3.0, "sampleId": "Test_Sample11_218", "tumorCellularity": null, "controlKitBarcode": null, "extractionKitBarcode": "9110009055910123456717211019" }, { "libraryType": "RNA", "barcodeID": "IonHDDual_0103", "libraryName": "RNA_01", "inputQuantity": 3.0, "sampleId": "Test_Sample11_218", "tumorCellularity": null, "controlKitBarcode": null, "extractionKitBarcode": "9110009056010123456717211231" }], "id": 233, "assayName": "Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0", "libraryBatchId": "OPA_DNA_Fusion_Library_Bacth_01", "libraryKitBarcode": "", "panelKitBarcode": "", "libraryPrepType": "DNA+RNA", "ampliSeqHd": false }] }</pre>



Get run plans

plans	
URL	BASE_URL/plans
Description	Returns a list of available run plans in the system.
Method	GET
Header	curl -k --header 'username:<user name>' --header 'password:<user password>'
Example URL	curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/plans'
Request query parameters	- planName String. <i>Optional</i>. Name of the run plan. - viewType String. <i>Optional</i>. Valid values are "all" or "toBeProcessed".
Example response	<pre>{ "objects": [{ "id": 184, "planName": "OPA_DNA_Fusion_PlanRun_01", "planShortId": "4RSKR", "type": "sample", "createdBy": "System Installed", "updatedOn": "2021-07-22T06:08:03.000+0000", "noOfSamples": 2, "noOfLibraries": 4, "samples": [{ "id": 973, "sampleId": "NTC", "assayName": "Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0", "dnaBarcodes": [], "rnaBarcodes": [], "libraryBatchId": "234", "libraryPrepType": "DNA+RNA", "pauseForReview": false }, { "id": 972, "sampleId": "Test_Sample11_278", "assayName": "Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0", "dnaBarcodes": [], "rnaBarcodes": [], "libraryBatchId": "234", "libraryPrepType": "DNA+RNA", "pauseForReview": false }], "runAssays": ["Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0"] }, { "total": 1 }] }</pre>

Get results

results	
URL	BASE_URL/results
Description	Returns results, including plugins and plugin results. Returns a JSON object for each plugin. All plugins for the run and the sample are returned.
Method	GET
Header	<code>curl -k --header 'username:<user name>' --header 'password: <user password>'</code>
Example URL	<code>curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/results'</code>

(continued)

results	
Request query parameters	id String. <i>Optional</i>. Results for the selected result id. Example: 123 assay_id String. <i>Optional</i>. Results for the assay id. Example: 123 plan_id String. <i>Optional</i>. Results for selected run plan id. Example: 123 sampleId String. <i>Optional</i>. Results for the selected sample name. Example: MySample planName String. <i>Optional</i>. Results for selected run plan name. Example: MyPlan analysisComplete String. <i>Optional</i>. Results for completed runs. Results included for failed and successfully completed runs. Example: true updatedBefore String. <i>Optional</i>. Returns results updated before the selected date. Use date format: <code>yyyy-MM-dd HH:MM</code>. Example: 2019-01-15 10:30 updatedAfter String. <i>Optional</i>. Returns results updated after the selected date. Use date format: <code>yyyy-MM-dd</code> Example: 2019-01-15 orderBy String. <i>Optional</i>. Parameter to sort results by selected field. The default sort is by <code>updatedBefore</code> or <code>updatedAfter</code>. Results are listed in descending order, with the most recent results listed first.



(continued)

	results
Example response	<pre> { "objects": [{ "id": 328, "planName": "EK_FFPE_D5_0.3.0_6.5.26-NA-T2.0.0-2", "sampleId": "SRTestSample61", "assay": { "id": 18, "value": "Generic Assay - DNA and Fusions - EU v0.3.0", "accountType": null, "isGreaterVersion": false, "runType": null, "greaterVersion": false }, "planRun": { "id": 59, "value": "EK_FFPE_D5_0.3.0_6.5.26-NA-T2.0.0-2", "accountType": null, "isGreaterVersion": false, "runType": null, "greaterVersion": false }, "sample": { "id": 45, "value": "SRTestSample61", "accountType": null, "isGreaterVersion": false, "runType": null, "greaterVersion": false }, "planShortCode": "YYRU2", "resultName": "Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_328", "libraryNames": "SRTestSample61_RNA831, SRTestSample61_DNA831", "runStartDate": "2021-07-23T00:00-0400", "runCompleteDate": "2021-07-24T00:45-0400", "updatedAt": "2021-07-24T00:44-0400", "libraryprepId": 428, "analysisComplete": true, "analysisVersion": "6.5.26", "isThumbnail": false, "isArchived": false, "isReanalysis": false, "runStatus": { "id": null, "name": "Analysis", "value": "Completed" }, "qcStatus": "Passed", "instrumentName": "ValkyrieTestRig", "outputsLink": "/analysis_output/Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_20210723215902373/ChipLane1_27SRTestSample61_LibPrep428/Outputs", "outputsDir": "<resultsoutputdirectory>", "loadingMetrics": [{ "name": "Total Addressable Wells", "count": 64847855 }, { "name": "Wells With ISPs", "count": 60145720, "percentage": 92.75 }, ...], "filteringMetrics": [{ "name": "Filtered: Polyclonal", "count": 22931697, "percentage": 38.27 }, { "name": "Filtered: Primer-Dimer", "count": 9955, "percentage": 0.02 }, ...], "runQCMetrics": [{ "data": [{ "metricName": "Key Signal", "value": "75", "referenceRange": "Not Set", "qcStatus": "Not Calculated", "percentValue": false, "iscosmic": false, "isPQCControl": false, </pre>



Get signed off samples

signedOffSamples	
URL	BASE_URL/signedOffSamples
Description	Returns a list of the samples that are designated with <i>(Signed Off)</i> .
Method	GET
Header	curl -k --header 'username:<user name>' --header 'password: <user password>'
Example URL	curl -k -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/signedOffSamples'
Request query parameters	- start_date String, <i>Optional</i>. Provide the start date as yyyy-mm-dd. - end_date String, <i>Optional</i>. Provide the end date as yyyy-mm-dd. - signedOff String, <i>Required</i> A Boolean data type. Valid values are "true" or a "false". The default value is "true".
Example response	<pre>{ "meta": { "pageSize": 20, "next": null, "pageNumber": 0, "previous": null, "total_count": 76 }, "objects": [{ "sample": { "assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0", "base_url": "AssayDev_PQSample1_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS", "checksum_file": "CHECKSUM", "created_date": "2021-06-23", "sample_file": "Info.csv", "sample_id": "92", "sample_name": "PQSample1", "planName": "IQ_Lib" } }, { "sample": { "assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0", "base_url": "AssayDev_PQSample2_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS", "checksum_file": "CHECKSUM", "created_date": "2021-06-23", "sample_file": "Info.csv", "sample_id": "93", "sample_name": "PQSample2", "planName": "IQ_Lib" } }] }</pre>



Genexus™ Software Service API

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Genexus™ Software has a web services API that allows you to automate the retrieval of key information from the system.

This section provides examples to show you how to leverage the API.

Genexus™ Software Service API introduction

Genexus™ Software Service API integrates with laboratory information management systems (LIMS) to help with sample, library, and run plan creation, and to track samples, monitor run progress and retrieve sample results. This service can be installed using the Software Update feature and is a new enhancement provided with Genexus™ Software 7.0.

The Genexus™ Software Service API is intended to fully replace the Genexus™ Software API in a future release. If you are currently using the Genexus™ Software API, which is documented in Appendix A, “Genexus™ Software API”, you are encouraged to transition to this new Genexus™ Software Service API.

The following table shows the API names and functions of the RESTful APIs that are provided in the Genexus™ Software Service API.

API	Function
createLibraryBatch “Create a library batch” on page 460.	Creates a library batch from one or more existing samples. Creates a library batch and one or more new samples in a single request. All or None Library batches are created in case of success or failure.
createSamples “Create samples” on page 461.	Creates one to <i>n</i> samples. All or None is followed for all samples in a request. If any one of sample creation fails, none of the samples in the input payload is created in system.
createBamSamples “Create BAM samples” on page 463	Creates one to <i>n</i> samples. All or None is followed for all samples in a request. If any one of sample creation fails, none of the samples in the input payload is created in system.
createTagByDisplayName “Create a sample tag by display name” on page 465.	Creates a sample tag based on display name.
downloadResultFiles “Download result files” on page 466.	Downloads results files such as VCF, reports, bam files, and so on. Returns a compressed file in ZIP format that contains the requested files for the sample.
getAccounts “Get a list of Ion Reporter™ Software accounts” on page 467.	Returns a list and the details of active Ion Reporter™ Software accounts.
getAllVariants “Get a list of variants” on page 469.	Returns filtered variants based on the filter chain associated with the run.
getAssays “Get a list of assays” on page 471.	Returns a list of the assays in the system. PQ (performance qualification) assays are excluded.
getAuthToken “Get the authenticator token” on page 459.	Returns the authenticator token for the LIMS api.
getInstrumentStatus “Get instrument status” on page 473.	Returns the status of the Genexus™ Integrated Sequencer.

(continued)

API	Function
getLibraryBatch See “Get library batches” on page 477.	Returns a list of the library batches in the system.
getPlans “Get a list of run plans” on page 479.	Returns a lists of plans that are available in the system. The API supports multiple filters to filter the plans to retrieve.
getReportTemplates “Get a list of report templates” on page 481.	Returns a list of report templates that are available in the system. The API supports multiple filters to filter the templates to retrieve.
getReproSeqVariants “Get a list of variants in Ion ReproSeq™ assays” on page 482.	Returns Ion ReproSeq™ variant data.
getResultFiles “Get a list of result files” on page 497.	Returns files that are available to download for a result.
getResults “Get analysis results” on page 498.	Returns analysis results, including plugins and plugin results. Returns a JSON object for each plugin. All plugins for the run and the sample are returned.
getSamples “Get a list of samples” on page 501.	Returns a list of the samples that are in the system.
getSampleStatus “Get sample status” on page 500.	Returns the current status of a sample.
getSignedOffSamples “Get a list of signed-off samples” on page 503.	Returns a list of the samples that are designated with (Signed Off) .
updateSamples “Update samples” on page 504.	Updates one to n samples. All or None is followed for all samples in a request.

API common parameters

If the JSON download process converts @ symbols to %40, do not change the text. Instead, copy the JSON files, as is, into the browser.

Parameter	Entry
BASE_URL	https://<host>/service/api/
Request	Authorization:Basic Accept:application/json
Response	Content-Type:application/json

Response codes

Status code	Status
200—	Successful response. This is the default response.
400—Bad Request	Bad input parameter. The error message should indicate which one and why. Error response: <pre>{ "errors": [{ "field": "validation error" }] }</pre>
401—Authentication failure	The client passed in the invalid Auth token. The client should refresh the token and then try again.
404—Not found	The requested resource was not found.
483—Forbidden	Customer does not exist. Application is not registered. Application tries to access properties that do not belong to an application. Application tries to trash/purge the root node. Application tries to update content properties. Operaton is blocked (for third-party apps). Customer account is over quota.
500—Internal Server Error	Internal Server Error

Common request parameters

Parameter	Entry	Value
Starting page number	pageNumber	The default value is 1.
Number of items to fetch. You can limit the number of items that are fetched when you add parameters to the request. For example, add "status" to the get assay list request.	pageSize	The default value is 20.
A filed based on which, the result set needs to be ordered	orderBy	Only specific values are allowed. If no value is provided, the result set is ordered by ID.
Order direction of the result set	sortOrder	Valid values are asc or desc. The default value is asc.

Get the authenticator token

The `getAuthToken` API returns the authenticator token required for use with the Genexus™ Software Service API. The authenticator token identifies the authorized user and password, mode, and organization.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is `/auth/getAuthToken`.

CURL command syntax

```
curl -X GET
"https://<host>/service/auth/getAuthToken"
-H "accept: */*"
-H "mode: AssayDev"
-H "orgName: GNX Org"
-H "Authorization: Basic [[base 64 has of username:password]]"
```

where `basicHash` = Base64Encoded value of `username:password`

Request URL

`https://<host>/service/auth/getAuthToken`

Header parameters

Item	Description
mode	The application mode. String. <i>Required</i> Allowed value and default is "AssayDev". Leave the default value as "AssayDev".
orgName	The organization name. String. <i>Optional</i> If passed as a null token, the response is based on the default organization of the API requesting user.

Example response

```
Token <ACCESS_TOKEN>
```

Create a library batch

The `createLibraryBatch` API creates a library batch from one or more existing samples.

This API also creates a library batch and one or more new samples in a single request. All or None Library batches are created in case of success or failure.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is POST `/api/library-batches`.

CURL command syntax

```
curl -X POST
"<https://<host>/service/api/library-batches"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
-H "Content-Type: application/json" --data '<input payload>'
```

Body parameters

```
{ []
total: integer (int32)
objects: [ []
  { []
    Required: libraryBatchId,libraryPrepType
    libraries: [ []
      { []
        Required: barcodeID,libraryType
        libraryType: string
        barcodeID: string
        libraryName: string
        inputQuantity: number (float)
        sampleId: string
        tumorCellularity: string control
        KitBarcode: string
        extractionKitBarcode: string
      }
    ]
    assayName: string
    assayShortName: string
    libraryBatchId: string
    libraryKitBarcode: string
    panelKitBarcode: string
    libraryPrepType: string
  }
]
```

Response

```
{
total: integer (int32)
objects: [
```



```

{
  Required: libraryBatchId,libraryPrepType
  libraries: [
    {
      Required: barcodeID,libraryType
      libraryType: string
      barcodeID: string
      libraryName: string
      inputQuantity: number (float)
      sampleId: string
      tumorCellularity: string
      controlKitBarcode: string
      extractionKitBarcode: string
    }
  ]
  id: integer (int32)
  assayName: string
  assayShortName: string
  libraryBatchId: string
  libraryKitBarcode: string
  panelKitBarcode: string
  libraryPrepType: string
}
]
}

```

Create samples

The `createSamples` API creates one to n samples. All or None is followed for all samples in a request. If any one of sample creation fails, none of samples in the input payload is created in system.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is POST `/api/samples`.

CURL command syntax

```

curl -X POST
"https://<host>/service/api/samples"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
-H "Content-Type: application/json" --data '<input payload>'

```

Body parameters

```

{
  objects: [
    {
      otherAttributes: {
        Attribute1: string
        Attribute2: string
      }
    }
  ]
  tags: [

```

```

        string
    ]
    sampleId: string maxLength:50
    collectionDate: string // year-month-day
    gender: string // Male, Female, Unknown
    notes: string
    sampleType: string // FFPE, Fresh Frozen Tissue, Core Needle Biopsy
(CNB), Fine Needle Aspiration (FNA), Bone marrow, Blood (Whole), Blood
(Plasma), Blood (Serum), Blood (Buffy Coat), Blood (Umbilical/Placental
Cord), Cerebrospinal Fluid (CSF), Saliva, Swab (Nasal), Swab (Buccal),
Swab (Vaginal), Urine, Unknown, Embryo Biopsy, Spent Media, Product Of
Conception, Cell Line, Other
    tumorCellularity: string
    necrosis: string
    origin: string
    customOrigin: string
    extractionKit: string
    cancerStage: string
    diseaseCategory: string
    customDiseaseCategory: string
    reason: string
    patientId: string
    orderingPhysician: string
    receiveDate: string // year-month-day
    dateOfBirth: string // year-month-day
    diagnosis: string
    customCancerType: string
    cancerTypeDisplay: string
    sixteensrRNA: string
    targetSpecies: string
    libraryChemistry: string
    caseId: string
    category: string
    }
    ]
}

```

Response

```

{
  List of Sample Lims DTO
  Required: objects
  total: integer (int64)
    total number of samples
  objects: [ (1..2147483647)
    List of sample DTO's
    {
      Sample Lims DTO
      Required: collectionDate, sampleId
      otherAttributes: {
      }
      tags: [
        string
      ]
      id: integer (int32)
      sampleId: string maxLength:50
      collectionDate: string
      gender: string
      notes: string
      sampleType: string
    }
  ]
}

```

```

        tumorCellularity:      string
        necrosis:              string
        origin:                 string
        customOrigin:          string
        extractionKit:          string
        cancerStage:            string
        diseaseCategory:        string
        customDiseaseCategory:  string
        reason:                 string
        patientId:              string
        orderingPhysician:      string
        receiveDate:            string
        dateOfBirth:            string
        diagnosis:              string
        sampleOrigin:           string
        editAndAmend:           boolean
        customCancerType:       string
        cancerTypeDisplay:      string
        sixteensrRNA:           string
        targetSpecies:          string
        libraryChemistry:       string
        caseId:                 string
        metaData:                {
                                }
        category:               string
        dnaBamFile:              string
        rnaBamFile:              string
        tnaBamFile:              string
    }
}

```

Create BAM samples

The `createBamSamples` API creates one to n samples. All or None is followed for all samples in a request. If any one of sample creation fails, none of samples in the input payload is created in system.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is POST `/api/BAM-samples`.

CURL command syntax

```

curl -X POST
"https://<host>/service/api/bam-samples"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
-H "Content-Type: application/json" --data '<input payload>'

```

Body parameters

```

{
  objects: [

```

```

{
  otherAttributes: {
    Attribute1: string,
    Attribute2: string
  }
  tags: [
    string
  ]
  sampleId: string maxLength:50
  collectionDate: string // year-month-day
  gender: string // Male, Female, Unknown
  notes: string
  sampleType: string // FFPE, Fresh Frozen Tissue, Core Needle Biopsy
  (CNB), Fine Needle Aspiration (FNA), Bone marrow, Blood (Whole), Blood
  (Plasma), Blood (Serum), Blood (Buffy Coat), Blood (Umbilical/Placental
  Cord), Cerebrospinal Fluid (CSF), Saliva, Swab (Nasal), Swab (Buccal),
  Swab (Vaginal), Urine, Unknown, Embryo Biopsy, Spent Media, Product Of
  Conception, Cell Line, Other
  tumorCellularity: string
  necrosis: string
  sampleOrigin: string
  customOrigin: string
  extractionKit: string
  cancerStage: string
  diseaseCategory: string
  customDiseaseCategory: string
  reason: string
  patientId: string
  orderingPhysician: string
  receiveDate: string // year-month-day
  dateOfBirth: string // year-month-day
  diagnosis: string
  customCancerType: string
  cancerTypeDisplay: string
  sixteensrRNA: string
  targetSpecies: string
  libraryChemistry: string
  caseId: string
  category: string
  dnaBamFile: string
  rnaBamFile: string
  tnaBamFile: string
}
]
}

```

Response

```

{
  total: integer (int64)
  objects: [
    {
      id: integer (int32)
      otherAttributes: {
        Attribute1: string
        Attribute2: string
      }
      tags: [
        string
      ]
    }
  ]
}

```

```

sampleId: string maxLength:50
collectionDate: string
gender: string
notes: string
sampleType: string
tumorCellularity: string
necrosis: string
sampleOrigin: string
customOrigin: string
extractionKit: string
cancerStage: string
diseaseCategory: string
customDiseaseCategory: string
reason: string
patientId: string
orderingPhysician: string
receiveDate: string
dateOfBirth: string
diagnosis: string
customCancerType: string
cancerTypeDisplay: string
sixteensrRNA: string
targetSpecies: string
libraryChemistry: string
caseId: string
category: string
dnaBamFile: string
rnaBamFile: string
tnaBamFile: string
    }
  ]
}

```

Create a sample tag by display name

The `createTagByDisplayName` API creates a sample tag based on display name.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is POST `/api/createTag`.

CURL command syntax

```

curl -X POST
"https://<host>/service/api/createTag?tagName=<tag_display_name>"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: */*"

```

Query parameters

Item	Description
tagName	The name of the tag to create. String. <i>Required</i> .

Example response

Status: 200— Tag Id

```
integer
```

Download result files

The `downloadResultFiles` API downloads results files, such as VCF, reports, bam files, and so on, for completed runs. The API returns a compressed file in ZIP format that contains the requested files for the sample. The `getResultFiles` API returns the list of result files and the `downloadResultFiles` API downloads the result files.

If the user selects BAM files and/or Lab Reports, the API may return an HTTP 206 status, along with a message indicating that the file generation is still in progress.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is `/api/results/{resultId}/files/download`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/results/{resultId}/files/download?
files=file1&files=file2"
-H "Authorization: [[Auth token which
starts as Token, for example, Token xyz...]]"
-H "mode: AssayDev"
```

Path parameters

Item	Description
resultId	The ID of the result for which to download the files. The ID of the result is provided by first using the <code>getResultFiles</code> API. Integer (int32). <i>Required</i> .

Query parameters

Item	Description
files	A list of the file names. Array (String). <i>Required</i>

Response

If CRAM-to-BAM conversion is required, the API returns a **206** status with the message:

File generation is in progress. Please check back later.

When the process completes successfully, the API returns a **200** status along with the ZIP file.

Get a list of Ion Reporter™ Software accounts

The `getAccounts` API returns a list and the details about active Ion Reporter™ Software accounts.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/ir-accounts`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/ir-accounts?
irversion=&pageNumber=&pageSize=&orderBy=&sortOrder="
-H "Authorization: [[Auth
token which starts as Token, for example, Token xyz...]]"
-H "mode: AssayDev"
```

Query parameters

Item	Description
irversion	Ion Reporter™ Software account version <i>String. Optional.</i>
pageNumber	Starting page number. The default is 1. <i>Integer (int32). Optional</i>
pageSize	Number of items to fetch. The default value is 20. <i>String. Optional.</i>
orderBy	The field to be sorted on. The default sorting is based on display name. <i>String. Optional.</i> Valid values are "ID", "displayName", "server", "serverType", "userName", and "accountType".
sortOrder	Sort order, ascending or descending. The default is ascending. <i>String. Optional.</i> Valid values are "asc" and "desc".

Example response

```
{
  "total": 0,
  "objects": [
    {
      "id": "string",
      "displayName": "string",
      "serverType": "string",
      "port": "string",
      "state": "string",
      "version": "string",
      "userName": "string",
      "password": "string",
      "org": "string",
      "status": "string",
      "server": "string",
      "irVersion": "string",
      "irVersions": [
        "string"
      ],
      "actionMap": {
        "additionalProp1": [
          {
            "actionType": "string",
            "actionLocalizeKey": "string",
            "mandatory": true,
            "permission": "string",
            "allowedRoles": "string"
          }
        ],
        "additionalProp2": [
```



```

        {
          "actionType": "string",
          "actionLocalizeKey": "string",
          "mandatory": true,
          "permission": "string",
          "allowedRoles": "string"
        }
      ],
      "additionalProp2": [
        {
          "actionType": "string",
          "actionLocalizeKey": "string",
          "mandatory": true,
          "permission": "string",
          "allowedRoles": "string"
        }
      ],
      "additionalProp3": [
        {
          "actionType": "string",
          "actionLocalizeKey": "string",
          "mandatory": true,
          "permission": "string",
          "allowedRoles": "string"
        }
      ]
    },
    "defaultAccount": true,
    "accountType": "string",
    "usedAccount": true,
    "irAccountListError": true,
    "irAccount": true,
    "createdBy": "string",
    "reason": "string"
  }
]
}

```

Get a list of variants

The `getAllVariants` API returns a list of filtered variants in the results of a run based on the filter chain associated with the run.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/variants`.

CURL command syntax

```

curl -X GET
"https://<host>/service/api/variants?resultId="
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"

```

Query parameters

Item	Description
resultId	The ID of the result for which to get the list of variants. Integer (int32). <i>Required.</i>

Response

Status: 200—List of variants, where column contains key as column name same as UI label

```
{
  total: integer (int32)
  variants: [
    {
      columns: {
      }
      metas: {
        hasNotes: boolean
        defaultFilterGene: string
        chromosome: string
        id: string
        position: integer (int64)
        category: string
      }
      oncomineInfo: {
        variantName: string
        tier: string
        therapiesInIndication: {
          breakthrough: [
            string
          ]
          contraindicated: [
            string
          ]
          fastTrack: [
            string
          ]
          indicated: [
            string
          ]
          notRecommended: [
            string
          ]
          resistance: [
            string
          ]
        }
        therapiesInOtherIndication: {
          breakthrough: [
            string
          ]
          contraindicated: [
            string
          ]
          fastTrack: [
            string
          ]
          indicated: [

```

```
    string
    ]
    notRecommended: [
        string
    ]
    resistance: [
        string
    ]
    }
    trialCount: integer (int32)
}
]
```

Get a list of assays

The `getAssays` API returns a list of the assays in the system for the API requesting user. Performance qualification (PQ) assays are excluded.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/assays`.

CURL command syntax

```
curl -X GET "https://<host>/service/api/assays?
assayName=&assayShortName=&assayDisplayName=&status=&pageNumber=&pageSize=&or
derBy=&sortOrder=" -H "Authorization: [[Auth token which starts as Token,
for example, Token xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
```

Query parameters

Item	Description
assayName	Assay name. String. <i>Optional</i> .
assayShortName	Assay short name. String. <i>Optional</i> .
assayDisplayName	Assay display name. String. <i>Optional</i> .
status	Status. String. <i>Optional</i> . Valid statuses are "OBSOLETE", "LOCKED", "DRAFT", or "ALL". If any other value is entered, the API call is made using "ALL".

(continued)

Item	Description
pageNumber	Starting page number. The default is 1. Integer (int32). <i>Optional</i> .
pageSize	Number of items to fetch. The default value is 20. Integer (int32). <i>Optional</i> .
orderBy	The field to be sorted on. The default sorting order is based on id. String. <i>Optional</i> . Valid values are "id", "assayName", "assayShortName", "assayDisplayedName", and "application".
sortOrder	Sort order, ascending or descending. The default is ascending (the oldest assays first). String. <i>Optional</i> . Valid values are "asc" and "desc".

Response

```

{
  total:    integer (int64)
  objects: [
    {
      id:                integer (int32)
      assayName:         string
      assayShortName:    string
      assayDisplayedName: string
      application:       string
      createdBy:         string
      createdAt:         string (date-time)
      genomeReference:   string
      targetRegions:     string
      hotspots:          string
      fusionReference:    string
      fusionTargetRegions: string
      chipType:          string
      panelKit:          string
      controlKit:        string
      extractionKit:     string
      libraryKit:        string
      templatingKit:     string
      barcodeKit:        string
      sequencingKit:     string
      flows:             integer (int32)
      templatingSize:    integer (int32)
      status:            string
      tumorCellularity:  string
      reanalysisAssay:   boolean
    }
  ]
}

```

Get instrument status

The `getInstrumentStatus` API returns the status of the Genexus™ Integrated Sequencer.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/instrument-status`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/instrument-status"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
```

Response

Status: 200—Instrument status, "runData" returns if user has the role permission to view instrument data.

```
{
  runData: [
    {
      id: integer (int32)
      name: string
      user: string
      runStatus: string
      startTime: string
      completionTime: string
      estimatedCompletionTime: string
      elapsedTime: string
      purificationKit: string
      purificationBatchIdStatus: string
      percentCompleted: integer (int32)
      analysisStatus: string
      outOfSync: boolean
      displayStatus: string
      outOfSyncMessage: string
      stageIndex: integer (int32)
      stageStatus: string
      pqStatus: boolean
      pqDueDate: string
      rigStatus: string
      rigState: string
      instrumentType: string
      serial: string
      instrumentName: string
      instrumentAlarms: [
        string
      ]
      sequencingStage: boolean
      runStages: [
        {
          index: integer (int32)
          shortName: string
        }
      ]
    }
  ]
}
```

```

        lastInitDate:      string
        deckStatus:        string
        chipBarcode:       string
        noShowRunDetails:  boolean
        noShowStageDetails: boolean
        pluginStatus:      string
    }
]
planSamples: [
    {
        sequencingStage: boolean
        id:               integer (int32)
        planRun:          {
            id:            integer (int32)
            value:         string
            accountType:   string
            runType:       string
            greaterVersion: boolean
        }
        runType:          string
        resultInfo:       [
            {
                expLibraryprepId: integer (int32)
                planRun: {
                    id:            integer (int32)
                    value:         string
                    accountType:   string
                    runType:       string
                    greaterVersion: boolean
                }
                libraryNames:      string
                assay: {
                    id:            integer (int32)
                    value:         string
                    accountType:   string
                    runType:       string
                    shortName:     string
                    assayAnnotations: string
                    name:          string
                    useLatest:     boolean
                    applicationType: string
                    greaterVersion: boolean
                }
            }
        ]
        runStatus: {
            id:      string
            name:    string
            value:   string
        }
        qcStatus:      string
        hasNotes:       boolean
        resultId:       integer (int32)
        analysisComplete: boolean
        fusion:         boolean
        analysisStatus: boolean
        genericWorkflow: boolean
        enableReanalysis: boolean
        summaryReportRequired: boolean
        archived:       boolean
        viewResult:     boolean
        shortCode:      string
    }
]

```

```

hideLabReport:      boolean
sample:             {
                    id:          integer(int32)
                    value:       string
                    accountType: string
                    runType:     string
                    greaterVersion: boolean
                    }
runType:            string
otStatus:           string
pgmStatus:          string
actionMap:          {
                    }
signedOff:          boolean
startedOn:          string
relaunchPlugins:    boolean
updatedAt:          string
id:                 integer (int32)
sampleDetails: {
    specimenAttributes: [
        {
            id:          integer (int32)
            attributeId:  integer (int32)
            value:       string
            attributeName: string
            defaultAttrName: string
            mandatory:    boolean
        }
    ]
    id:                 integer (int32)
    specimenId:         string
    patientId:          string
    orderingPhysician:  string
    collectionDate:     string
    receiveDate:        string
    gender:             string
    notes:              string
    hasNotes:           boolean
    dateOfBirth:        string
    sampleType:         string
    tumorCellularity:   string
    necrosis:           string
    diagnosis:          string
    sampleOrigin:       string
    editAndAmend:        boolean
    customSampleOrigin:  string
    customCancerType:    string
    cancerTypeDisplay:   string
    cancerStage:        string
    diseaseCategory:     string
    customDiseaseCategory: string
    dnaBamFile:         string
    rnaBamFile:         string
    tnaBamFile:         string
    sixteensrRNA:       string
    targetSpecies:      string
    vcfFile:            string
    libraryChemistry:   string
    caseId:             string
    filePaths:          {
                    }
}

```

```

        metaData: {
        }
        category: string
        inlineError: boolean
        customAttributes: {
        }
        bulkCreate: boolean
        reason: string
        extractionKit: string
        edit: boolean
        actionMap: {
        }
        disabledAttributes: {
        }
        tags: [
            string
        ]
        hasTag: boolean
        badgeText: string
    }
    researchApplication: string
    applicationType: string
    customAttributes: {
    }
    hideVariantAudit: boolean
    tags: [
        string
    ]
    hasTag: boolean
    rigAlarms: [
        string
    ]
    lockedSignoff: boolean
    variantReportSignOffs: [
        {
            value: string
            icon: string
            id: integer (int32)
        }
    ]
    okrReportSignOffs: [
        {
            value: string
            icon: string
            id: integer (int32)
        }
    ]
    userEsignature: string
    usedAnnotationsSources: [
        string
    ]
    amended: boolean
    okrReportAmended: boolean
    reanalysis: boolean
    verification: boolean
    reanalysisAssay: boolean
    }
    ]
    actionMap: {
    }
    updatedAt: string

```



```

    startedOn:      string
    runInfo:        {
    }
    viewResult:     boolean
    assays:         [
        {
            id:              integer (int32)
            value:            string
            accountType:     string
            runType:         string
            shortName:       string
            assayAnnotations: string
            name:            string
            useLatest:       boolean
            applicationType: string
            greaterVersion:  boolean
        }
    ]
    numberOfSamples: integer (int32)
    resultId:         integer (int32)
    dataRetention:    boolean
    rawDataDeleted:   boolean
    intermediateFilesDeleted: boolean
    runStages:        [
        {
            index:           integer (int32)
            shortName:       string
        }
    ]
    overallQC:        {
    }
    stageIndex:       integer (int32)
    stageStatus:      string
    pluginStatus:     string
    instrumentAlarms: [
        string
    ]
    backupStatus:     [
        string
    ]
    purifInfo:        {
    }
    reanalysis:       boolean
    }
}

```

Get library batches

The `getLibraryBatch` API returns a list of the library batches that are available in the system for the API requesting user.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/library-batches`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/library-batches?
id=&pageNumber=&pageSize=&orderBy=&sortOrder="
-H "Authorization: [[Auth token
which starts as Token, for example, Token xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
```

Query parameters

Item	Description
id	The Library Batch ID that you want to fetch. String. <i>Optional</i> .
pageNumber	Starting page number. The default is 1. Integer (int32). <i>Optional</i> .
pageSize	Number of items to fetch. The default value is 20. Integer (int32). <i>Optional</i> .
orderBy	The field to be sorted on. The default sorting order is based on id. String. <i>Optional</i> . Valid values are "id", "panelKitBarcode", and "libraryKitBarcode".
sortOrder	Sort order, ascending or descending. The default is ascending (the oldest library batch first). String. <i>Optional</i> . Valid values are "asc" and "desc".

Example response

Status: 200—List of Library batches

```
{
  total: integer (int32)
  objects: [
    {
      Required: libraryBatchId, libraryPrepType
      libraries: [
        {
          Required: barcodeID, libraryType
          libraryType: string
          barcodeID: string
          libraryName: string
          inputQuantity: number (float)
          sampleId: string
          tumorCellularity: string
          controlKitBarcode: string
          extractionKitBarcode: string
        }
      ]
      id: integer (int32)
```

```
        assayName:      string
        assayShortName: string
        libraryBatchId: string
        libraryKitBarcode: string
        panelKitBarcode: string
        libraryPrepType: string
    }
}
```

Get a list of run plans

The `getPlans` API returns a list of run plans that are available in the system for the API requesting user. The API supports multiple filters to filter the plans to retrieve.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/plans`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/plans?
planName=&viewType=&pageNumber=&pageSize=&orderBy=&sortOrder="
-H "Authorization: [[Auth
token which starts as Token, for example, Token xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
```

Query parameters

Item	Description
planName	Specific plan name for which to get the details. String. <i>Optional</i> .
viewType	Specify whether to view all plans or only those to be processed. If not passed, the default is "all". String. <i>Optional</i> . Valid values are "all" or "toBeProcessed".
pageNumber	A positive integer to display results from a given offset (also known as page number). Integer (int32). <i>Optional</i> . The default value is 1.
pageSize	The number of pages to fetch. Integer (int32). <i>Optional</i> . The default number is 20.

(continued)

Item	Description
orderBy	The field to be sorted on. The default sorting is based on ID. String. <i>Optional</i> . Valid values are "id", "planName", and "planShortId".
sortOrder	Sort order, ascending or descending. The default is descending (latest plans first). String. <i>Optional</i> . Valid values are "asc" and "desc".

Example response

Status: 200—default response

```
{
  objects: [
    {
      id:
integer (int32)
      planName:      string maxLength:50
                     Pattern: [A-Za-z0-9\.\_\-]*
      planShortId:   string
      type:          string
      status:        string
      createdBy:     string
      createdOn:     string (date-time)
      updatedOn:     string (date-time)
      noOfSamples:   integer (int32)
      noOfLibraries: integer (int32)
      notes:         string
      samples:       [
        {
          id:                integer (int32)
          sampleId:          string
          assayName:         string
          purificationProtocol: string
          dnaBarcodes:       [
            string
          ]
          rnaBarcodes:       [
            string
          ]
          libraryBatchId:    string
          libraryPrepType:   string
          elutionVolumeDNA: integer (int32)
          elutionVolumeRNA: integer (int32)
          elutionVolumeTNA: integer (int32)
          concentration:     number (double)
          kitBarcode:        string
          pauseForReview:    boolean
        }
      ]
      runAssays: [ (0..∞)
                  string
                ]
    }
  ]
}
```

```
        instrument:      string
        reportTemplateName: string
        irAccountName:   string
        irAccountVersion: string
    }
]
total: integer (int64)
}
```

Get a list of report templates

The `getReportTemplates` API returns a list of report templates that are available in the system for the API requesting user. The API supports multiple filters to filter the templates to retrieve.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/report-templates`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/report-templates?assayName=&assayShortName="
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
```

Query parameters

Item	Description
assayName	The assay full name is used if <code>assayShortName</code> is not set. If both are not set, then the API fetches all report templates for the API requesting user. <i>String. Optional.</i>
assayShortName	If <code>assayShortName</code> is set, then <code>assayName</code> is ignored and report templates are fetched based on <code>assayShortName</code> . <i>String. Optional.</i>

Response

Status: 200—List of report templates

```
{
  Required: mode
  mode:      string
  reportingTemplate: [
    {
      Required: assayName, name
      name:      string
      id:         integer (int32)
      assayName:  string
    }
  ]
}
```

```

        assayId:      integer (int32)
        description:   string
        locale:        string
        localeDisplayName: string
        category:      string
        status:        string
        active:        boolean
        locked:        boolean
    }
]
total: integer (int64)
}

```

Get a list of variants in Ion ReproSeq™ assays

The `getReproSeqVariants` API returns a list of variant data from Ion ReproSeq™ preimplantation genetic screening (PGS) assays.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/repro-seq-variants`.

CURL command syntax

```

curl -X GET
"https://<host>/service/api/repro-seq-variants?resultId=11&cnvCategory="
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"

```

Query parameters

Item	Description
resultId	The ID of the result for which to get the list of files. The ID of the result is provided by first using the <code>getResults</code> API. Integer (int32). <i>Required</i> .
cnvCategory	Copy number variations category. If a value is not specified, the API returns full ploidy variants as the default. String. <i>Optional</i> . Allowed values are "fullploidy" and "mosaic".

Example response

Status: 200—ReproSeq variants data

```

{
  "cnvPlotResultDataDtoList": {
    "resultId": [
      {
        "chr": "string",

```

```

        "begin": 0,
        "end": 0,
        "ploidy": 0,
        "isAbove": true,
        "conf": 0,
        "precision": 0,
        "cytoband": "string",
        "percentContig": 0,
        "beginWithoutOffset": 0,
        "endWithoutOffset": 0,
        "chrNo": 0
    }
}
],
},
"cnvSampleInfoDtoResultIdMap": {
    "resultId": {
        "coupleId": "string",
        "embryoId": "string",
        "detectedGender": "string",
        "amplificationDate": "string",
        "technician": "string",
        "resultId": 0,
        "sampleName": "string",
        "collectionDate": "string",
        "runName": "string",
        "primary": true,
        "callingMethod": "string",
        "canChangePrimary": true,
        "morphologyGrade": "string",
        "biopsyDays": "string",
        "mapd": "string"
    }
},
"cnvMultiSampleDto": [
    {
        "resultId": 0,
        "sampleName": "string",
        "embryoId": "string",
        "gender": "string",
        "mapd": "string",
        "anPlus": "string",
        "anMinus": "string",
        "mtDna": "string",
        "summary": "string",
        "triploidy": "string",
        "edit": true,
        "signedOff": true
    }
],
"multiCnvSampleInfoDtoResultIdMap": {
    "additionalProp1": {
        "additionalProp1": {
            "coupleId": "string",
            "embryoId": "string",
            "detectedGender": "string",
            "amplificationDate": "string",
            "technician": "string",
            "resultId": 0,
            "sampleName": "string",
            "collectionDate": "string",
            "runName": "string",

```

```

        "primary": true,
        "callingMethod": "string",
        "canChangePrimary": true,
        "morphologyGrade": "string",
        "biopsyDays": "string",
        "mapd": "string"
    }
}
}, "multiCnvPlotResultDataDtoList": {
    "additionalProp1": {
        "additionalProp1": [
            {
                "chr": "string",
                "begin": 0,
                "end": 0,
                "ploidy": 0,
                "isAbove": true,
                "conf": 0,
                "precision": 0,
                "cytoband": "string",
                "percentContig": 0,
                "beginWithoutOffset": 0,
                "endWithoutOffset": 0,
                "chrNo": 0
            }
        ]
    }
},
"multiCnvMultiSampleDto": {
    "additionalProp1": {
        "additionalProp1": {
            "resultId": 0,
            "sampleName": "string",
            "embryoId": "string",
            "gender": "string",
            "mapd": "string",
            "anPlus": "string",
            "anMinus": "string",
            "mtDna": "string",
            "summary": "string",
            "triploidy": "string",
            "edit": true,
            "signedOff": true
        }
    }
},
"applicationCategory": "string"
}

```

Get result details for a sample

The `result-details` API retrieves detailed result data for a specified result ID, returning all associated plugins and key variant metrics.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is `/api/result-details`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/result-details?resultId="
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
```

Query parameters

Item	Description
resultId	The ID of the sample that is specified by the result ID for which to get details. The ID of the result is provided by first using the <code>getResults</code> API. Integer (int32). <i>Required</i> .

Response

Status: 200—default response

```
{
  "id": 0,
  "planName": "string",
  "sampleId": "string",
  "assay": {
    "id": 0,
    "value": "string",
    "accountType": "string",
    "runType": "string",
    "greaterVersion": true
  },
  "planRun": {
    "id": 0,
    "value": "string",
    "accountType": "string",
    "runType": "string",
    "greaterVersion": true
  },
  "sample": {
    "id": 0,
    "value": "string",
    "accountType": "string",
    "runType": "string",
    "greaterVersion": true
  },
  "planShortCode": "string",
  "resultName": "string",
  "libraryNames": "string",
  "runStartDate": "string",
  "runCompleteDate": "string",
  "updatedAt": "string",
  "libraryprepId": 0,
  "analysisComplete": true,
  "analysisVersion": "string",
  "isThumbnail": true,
  "isArchived": true,
  "isReanalysis": true,
  "runStatus": {
```

```

    "id": "string",
    "name": "string",
    "value": "string"
  },
  "qcStatus": "string",
  "instrumentName": "string",
  "outputsDir": "string",
  "updatedAnnotation": "string",
  "downloadFilesPath": "string",
  "baseFolderName": "string",
  "outputsLink": "string",
  "plugins": [
    {
      "id": 0,
      "pluginName": "string",
      "pluginVersion": "string",
      "updatedOn": "2025-07-15T22:49:38.497Z",
      "status": "string",
      "path": "string",
      "runType": "string"
    }
  ],
  "loadingMetrics": {
    "seq": [
      {
        "name": "string",
        "count": 0,
        "percentage": 0
      }
    ],
    "reseq": [
      {
        "name": "string",
        "count": 0,
        "percentage": 0
      }
    ]
  },
  "filteringMetrics": {
    "seq": [
      {
        "name": "string",
        "count": 0,
        "percentage": 0
      }
    ],
    "reseq": [
      {
        "name": "string",
        "count": 0,
        "percentage": 0
      }
    ]
  },
  "runQCMetrics": [
    {
      "title": "string",
      "data": [
        {
          "metricName": "string",
          "value": "string",

```

```

"referenceRange": "string",
"getQCStatus": "string",
"message": "string",
"percentValue": true,
"strValue": "string",
"strThreshold": "string",
"displayRange": "string",
"iscosmic": true,
"samples": {
  "additionalProp1": {
    "value": "string",
    "getQCStatus": "string",
    "strValue": "string"
  },
  "additionalProp2": {
    "value": "string",
    "getQCStatus": "string",
    "strValue": "string"
  },
  "additionalProp3": {
    "value": "string",
    "getQCStatus": "string",
    "strValue": "string"
  }
},
"ratio": "string",
"uniqueName": "string",
"sampleList": [
  "string"
],
"sampleA1": "string",
"sampleB1": "string",
"sampleC1": "string",
"sampleD1": "string",
"libraryType": "string",
"relationTypeDto": {
  "id": 0,
  "name": "string",
  "uniqueName": "string"
},
"qcStatusPdfReport": [
  "string"
],
"dataType": "string",
"sortOrder": 0,
"pqcontrol": true,
"displayMetric": true,
"displayQc": true
}
],
"sequence": 0,
"localizationKey": "string",
"inlineControlQc": true,
"customApp": "string",
"libraryType": "string",
"relationTypeDto": {
  "id": 0,
  "name": "string",
  "uniqueName": "string"
},
"images": [

```

```

        {
            "title": "string",
            "filePath": "string",
            "status": true,
            "type": "string"
        }
    ]
},
"purificationQCMetrics": [
    {
        "title": "string",
        "data": [
            {
                "metricName": "string",
                "value": "string",
                "referenceRange": "string",
                "getqCStatus": "string",
                "message": "string",
                "percentValue": true,
                "strValue": "string",
                "strThreshold": "string",
                "displayRange": "string",
                "iscosmic": true,
                "samples": {
                    "additionalProp1": {
                        "value": "string",
                        "getqCStatus": "string",
                        "strValue": "string"
                    },
                    "additionalProp2": {
                        "value": "string",
                        "getqCStatus": "string",
                        "strValue": "string"
                    },
                    "additionalProp3": {
                        "value": "string",
                        "getqCStatus": "string",
                        "strValue": "string"
                    }
                },
                "ratio": "string",
                "uniqueName": "string",
                "sampleList": [
                    "string"
                ],
                "sampleA1": "string",
                "sampleB1": "string",
                "sampleC1": "string",
                "sampleD1": "string",
                "libraryType": "string",
                "relationTypeDto": {
                    "id": 0,
                    "name": "string",
                    "uniqueName": "string"
                },
                "qcStatusPdfReport": [
                    "string"
                ],
                "dataType": "string",
                "sortOrder": 0,
            }
        ]
    }
]

```

```

        "pqcontrol": true,
        "displayMetric": true,
        "displayQc": true
    }
],
"sequence": 0,
"localizationKey": "string",
"inlineControlQc": true,
"customApp": "string",
"libraryType": "string",
"relationTypeDto": {
    "id": 0,
    "name": "string",
    "uniqueName": "string"
},
"images": [
    {
        "title": "string",
        "filePath": "string",
        "status": true,
        "type": "string"
    }
]
},
],
"controlQCMetrics": [
    {
        "title": "string",
        "data": [
            {
                "metricName": "string",
                "value": "string",
                "referenceRange": "string",
                "getqCStatus": "string",
                "message": "string",
                "percentValue": true,
                "strValue": "string",
                "strThreshold": "string",
                "displayRange": "string",
                "iscosmic": true,
                "samples": {
                    "additionalProp1": {
                        "value": "string",
                        "getqCStatus": "string",
                        "strValue": "string"
                    },
                    "additionalProp2": {
                        "value": "string",
                        "getqCStatus": "string",
                        "strValue": "string"
                    },
                    "additionalProp3": {
                        "value": "string",
                        "getqCStatus": "string",
                        "strValue": "string"
                    }
                }
            },
            "ratio": "string",
            "uniqueName": "string",
            "sampleList": [
                "string"
            ]
        ]
    }
]

```

```

    ],
    "sampleA1": "string",
    "sampleB1": "string",
    "sampleC1": "string",
    "sampleD1": "string",
    "libraryType": "string",
    "relationTypeDto": {
      "id": 0,
      "name": "string",
      "uniqueName": "string"
    },
    "qcStatusPdfReport": [
      "string"
    ],
    "dataType": "string",
    "sortOrder": 0,
    "pqcontrol": true,
    "displayMetric": true,
    "displayQc": true
  }
],
"sequence": 0,
"localizationKey": "string",
"inlineControlQc": true,
"customApp": "string",
"libraryType": "string",
"relationTypeDto": {
  "id": 0,
  "name": "string",
  "uniqueName": "string"
},
"images": [
  {
    "title": "string",
    "filePath": "string",
    "status": true,
    "type": "string"
  }
]
}
],
"sampleQCMetrics": [
  {
    "title": "string",
    "data": [
      {
        "metricName": "string",
        "value": "string",
        "referenceRange": "string",
        "getQCStatus": "string",
        "message": "string",
        "percentValue": true,
        "strValue": "string",
        "strThreshold": "string",
        "displayRange": "string",
        "iscosmic": true,
        "samples": {
          "additionalProp1": {
            "value": "string",
            "getQCStatus": "string",
            "strValue": "string"
          }
        }
      }
    ]
  }
]

```

```

        },
        "additionalProp2": {
            "value": "string",
            "getQCStatus": "string",
            "strValue": "string"
        },
        "additionalProp3": {
            "value": "string",
            "getQCStatus": "string",
            "strValue": "string"
        }
    },
    "ratio": "string",
    "uniqueName": "string",
    "sampleList": [
        "string"
    ],
    "sampleA1": "string",
    "sampleB1": "string",
    "sampleC1": "string",
    "sampleD1": "string",
    "libraryType": "string",
    "relationTypeDto": {
        "id": 0,
        "name": "string",
        "uniqueName": "string"
    },
    "qcStatusPdfReport": [
        "string"
    ],
    "dataType": "string",
    "sortOrder": 0,
    "pqcontrol": true,
    "displayMetric": true,
    "displayQc": true
    }
],
"sequence": 0,
"localizationKey": "string",
"inlineControlQc": true,
"customApp": "string",
"libraryType": "string",
"relationTypeDto": {
    "id": 0,
    "name": "string",
    "uniqueName": "string"
},
"images": [
    {
        "title": "string",
        "filePath": "string",
        "status": true,
        "type": "string"
    }
]
}
],
"sampleInlineControlQCMetrics": [
    {
        "title": "string",
        "data": [

```

```

{
  "metricName": "string",
  "value": "string",
  "referenceRange": "string",
  "getQCStatus": "string",
  "message": "string",
  "percentValue": true,
  "strValue": "string",
  "strThreshold": "string",
  "displayRange": "string",
  "iscosmic": true,
  "samples": {
    "additionalProp1": {
      "value": "string",
      "getQCStatus": "string",
      "strValue": "string"
    },
    "additionalProp2": {
      "value": "string",
      "getQCStatus": "string",
      "strValue": "string"
    },
    "additionalProp3": {
      "value": "string",
      "getQCStatus": "string",
      "strValue": "string"
    }
  },
  "ratio": "string",
  "uniqueName": "string",
  "sampleList": [
    "string"
  ],
  "sampleA1": "string",
  "sampleB1": "string",
  "sampleC1": "string",
  "sampleD1": "string",
  "libraryType": "string",
  "relationTypeDto": {
    "id": 0,
    "name": "string",
    "uniqueName": "string"
  },
  "qcStatusPdfReport": [
    "string"
  ],
  "dataType": "string",
  "sortOrder": 0,
  "pqcontrol": true,
  "displayMetric": true,
  "displayQc": true
},
"sequence": 0,
"localizationKey": "string",
"inlineControlQc": true,
"customApp": "string",
"libraryType": "string",
"relationTypeDto": {
  "id": 0,
  "name": "string",

```



```

        "uniqueName": "string"
    },
    "images": [
        {
            "title": "string",
            "filePath": "string",
            "status": true,
            "type": "string"
        }
    ]
},
"ntcQCMetrics": [
    {
        "title": "string",
        "data": [
            {
                "metricName": "string",
                "value": "string",
                "referenceRange": "string",
                "getQCStatus": "string",
                "message": "string",
                "percentValue": true,
                "strValue": "string",
                "strThreshold": "string",
                "displayRange": "string",
                "iscosmic": true,
                "samples": {
                    "additionalProp1": {
                        "value": "string",
                        "getQCStatus": "string",
                        "strValue": "string"
                    },
                    "additionalProp2": {
                        "value": "string",
                        "getQCStatus": "string",
                        "strValue": "string"
                    },
                    "additionalProp3": {
                        "value": "string",
                        "getQCStatus": "string",
                        "strValue": "string"
                    }
                },
                "ratio": "string",
                "uniqueName": "string",
                "sampleList": [
                    "string"
                ],
                "sampleA1": "string",
                "sampleB1": "string",
                "sampleC1": "string",
                "sampleD1": "string",
                "libraryType": "string",
                "relationTypeDto": {
                    "id": 0,
                    "name": "string",
                    "uniqueName": "string"
                },
                "qcStatusPdfReport": [
                    "string"
                ]
            }
        ]
    }
]

```

```

        ],
        "dataType": "string",
        "sortOrder": 0,
        "pgcontrol": true,
        "displayMetric": true,
        "displayQc": true
    }
],
"sequence": 0,
"localizationKey": "string",
"inlineControlQc": true,
"customApp": "string",
"libraryType": "string",
"relationTypeDto": {
    "id": 0,
    "name": "string",
    "uniqueName": "string"
},
"images": [
    {
        "title": "string",
        "filePath": "string",
        "status": true,
        "type": "string"
    }
]
},
"keyFindingsResponse": {
    "geneSummaryList": [
        {
            "geneName": "string",
            "geneDetectionStatus": "string",
            "variantDetectionStatus": "string",
            "variantDisplayID": "string",
            "aminoAcid": "string",
            "alleleFraction": "string",
            "variantId": "string",
            "variantCategory": "string",
            "moleFrequency": "string"
        }
    ],
    "filterChainName": "string",
    "geneList": [
        {
            "id": 0,
            "displayName": "string",
            "selected": true,
            "factoryProvided": true,
            "genes": [
                "string"
            ]
        }
    ],
    "selectedGeneList": {
        "id": 0,
        "displayName": "string",
        "selected": true,
        "factoryProvided": true,
        "genes": [
            "string"
        ]
    }
}

```

```

    ]
  },
  "selectedFilterPresetId": 0,
  "selectedFilterChainId": "string",
  "keyMetrics": [
    {
      "metricSectionTitle": "string",
      "metricDetails": [
        {
          "key": "string",
          "value": {},
          "locus": {}
        }
      ],
      "sectionType": "string",
      "columnHeaders": [
        "string"
      ]
    }
  ],
  "bioMarkerResultsMetrics": [
    {
      "metricSectionTitle": "string",
      "metricDetails": [
        {
          "key": "string",
          "value": {},
          "locus": {}
        }
      ],
      "sectionType": "string",
      "columnHeaders": [
        "string"
      ]
    }
  ],
  "geneCoverageGraph": [
    {}
  ],
  "specimenDetails": [
    {
      "key": "string",
      "value": "string"
    }
  ],
  "coverageMetrics": {
    "additionalProp1": {},
    "additionalProp2": {},
    "additionalProp3": {}
  },
  "oncomineReporterEnabled": true,
  "biomarkerResults": [
    {
      "localeKey": "string",
      "level": "string",
      "columnNames": [
        "string"
      ],
      "variants": [
        {
          "additionalProp1": {},

```

```

        "additionalProp2": {},
        "additionalProp3": {}
    }
  ],
  "qcMessages": [
    "string"
  ],
  "qcDetailMessages": [
    "string"
  ],
  "hrrData": {
    "additionalProp1": {},
    "additionalProp2": {},
    "additionalProp3": {}
  },
  "categoryQcMessages": {
    "additionalProp1": [
      "string"
    ],
    "additionalProp2": [
      "string"
    ],
    "additionalProp3": [
      "string"
    ]
  },
  "biomarkerIndicator": "string",
  "categoryToImportantMessagesMap": {
    "additionalProp1": [
      {
        "message": "string",
        "type": "string",
        "order": 0,
        "variantCategories": [
          "string"
        ]
      }
    ],
    "additionalProp2": [
      {
        "message": "string",
        "type": "string",
        "order": 0,
        "variantCategories": [
          "string"
        ]
      }
    ],
    "additionalProp3": [
      {
        "message": "string",
        "type": "string",
        "order": 0,
        "variantCategories": [
          "string"
        ]
      }
    ]
  },
  },

```

```
"variantsCountsByType": {
  "additionalProp1": {
    "additionalProp1": 0,
    "additionalProp2": 0,
    "additionalProp3": 0
  },
  "additionalProp2": {
    "additionalProp1": 0,
    "additionalProp2": 0,
    "additionalProp3": 0
  },
  "additionalProp3": {
    "additionalProp1": 0,
    "additionalProp2": 0,
    "additionalProp3": 0
  }
},
"noCallReported": true
}
```

Get a list of result files

The `getResultFiles` API returns a list of files that are available for a result.

The result ID and filenames provided by this API can then be used by the `downloadResultFiles` API.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/results/{resultId}/files`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/results/{resultId}/files"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
```

Path parameters

Item	Description
resultId	The ID of the result for which to get the list of files. Integer (int32). <i>Required</i> .

Example response

```
[
  {
    displayName: string
    fileNameToDownload: string
```

```
}
]
```

Get analysis results

The `getResults` API returns a high-level list of sample results. To retrieve detailed result information, the `result-details` API must be called.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/results`.

CURL command syntax

```
curl -X GET "https://<host>/service/api/results?
plan_id=&assay_id=&id&sampleId&planName&analysisComplete&updatedBefore&update
dAfter"
-H 'mode: AssayDev'
-H 'Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]'
```

Query parameters

Item	Description
<code>id</code>	Return results for the selected result ID, for example, "123".
<code>assay_id</code>	Return results for the selected assay ID, for example, "123".
<code>plan_id</code>	Return results for the selected plan ID, for example, "123".
<code>sampleId</code>	Return results for the selected sample name, for example, "MySample".
<code>planName</code>	Return results for the selected plan, for example, "MyPlan".
<code>analysisComplete</code>	Return results that completed processing (either Completed, Failed, Terminated, or Stalled), for example, "true".
<code>updatedBefore</code>	Return results that updated before a specific date, for example, "2019-01-15".
<code>updatedAfter</code>	Return results that updated after a specific date, for example, "2019-01-15"
<code>pageNumber</code>	Starting page number. The default is 1. Integer (int32). <i>Optional</i>
<code>pageSize</code>	Number of items to fetch. The default value is 10. String. <i>Optional</i> .

(continued)

Item	Description
orderBy	The field to be sorted on. The default sorting order is based on "resultUpdatedOn". <i>String. Optional.</i> Valid values are "id", "resultUpdatedOn", and "resultsName".
sortOrder	Sort order, ascending or descending. The default is descending (latest results first). <i>String. Optional.</i> Valid values are "asc" and "desc".

Response

```
{
  objects: [
    {
      id: integer (int32)
      planName: string
      sampleId: string
      assay: {
        id: integer (int32)
        value: string
        accountType: string
        runType: string
        greaterVersion: boolean
      }
      planRun: {
        id: integer (int32)
        value: string
        accountType: string
        runType: string
        greaterVersion: boolean
      }
      sample: {
        id: integer (int32)
        value: string
        accountType: string
        runType: string
        greaterVersion: boolean
      }
      planShortCode: string
      resultName: string
      libraryNames: string
      runStartDate: string // 2020-08-10T10:10+0000
      runCompleteDate: string // 2020-08-10T14:43+0000
      updatedDate: string // 2020-08-10T14:43+0000
      libraryprepId: integer (int32)
      analysisComplete: boolean
      analysisVersion: string
      isThumbnail: boolean
      isArchived: boolean
      isReanalysis: boolean
      runStatus: {
        id: string
        name: string
        value: string
      }
    }
  ]
}
```

```
}
qcStatus: string
instrumentName: string
updatedAnnotation: string
}
]
total: integer (int64)
}
```

Get sample status

The `getSampleStatus` API returns the current status of a sample.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/sampleStatus`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/sampleStatus?specimenId="
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
```

Query parameters

Item	Description
specimenId	The ID of the specimen. String. <i>Required</i> .

Example response

Status: 200 - Sample current status/stage, possible values are:

- **Sample Created**—This status indicates that a sample has been successfully generated or registered in the system.
- **Library Batch Created**—This status suggests that a library batch has been created using the previously generated sample.
- **Plan Run Created**—This status indicates that a plan for running experiments or tests using the samples or library batches has been created.
- Any purification -related or sequencing-related statuses are displayed as is.
- Sample Analysis Pipeline Status will be rolled up to following:
 - Analysis Pending—Prefix "Analysis" to **Pending** status
 - Analysis Stalled—Prefix "Analysis" to **Stalled** status
 - Analysis Terminated—Prefix "Analysis" to **Terminated** status
 - Analysis Completed—Prefix "Analysis" to **Completed** status

- Analysis Failed—Prefix "Analysis" to **Failed** status
- Analysis In Progress—For all the intermediate analysis statuses when the sample is being analyzed (: RUNNING) displays this status.

```
string
```

Get a list of samples

The `getSamples` API returns a list of the samples in the system for the API requesting user.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/samples`.

CURL command syntax

```
curl -X GET
"https://<host>/service/api/samples?
sampleId=&viewType=&pageNumber=&pageSize=&orderBy=&sortOrder="
-H "Authorization: [[Auth
token which starts as Token, for example, Token xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json" -H "Content-Type: application/json"
```

Query parameters

Item	Description
sampleId	The name of the sample for which to get the details. String. <i>Optional</i> .
viewType	The view type of the sample. If not specified, the default value is "all". String. <i>Optional</i> . Valid values are "all", "bamSamples", and "toBePrepared".
pageNumber	Starting page number. The default is 1. Integer (int32). <i>Optional</i>
pageSize	Number of items to fetch. The default value is 20. String. <i>Optional</i> .

(continued)

Item	Description
orderBy	The field to be sorted on. The default sorting order is based on "sampleId". String. <i>Optional</i>. Valid values are "id", "sampleId", "gender", "collectionDate", "diseaseCategory", "cancerTypeDisplay", and "sampleType".
sortOrder	Sort order, ascending or descending. The default is descending (latest samples first). String. <i>Optional</i>. Valid values are "asc" and "desc".

Response

```
{
List of Sample Lims DTO
Required: objects
total:          integer (int64)
                total number of samples
objects:        [ (1..2147483647)
List of sample DTO's
  {
    Sample Lims DTO
    Required: collectionDate, sampleId
    otherAttributes: {
      tags: [
        string
      ]
      id: integer (int32)
      sampleId: string maxLength:50
      collectionDate: string
      gender: string
      notes: string
      sampleType: string
      tumorCellularity: string
      necrosis: string
      origin: string
      customOrigin: string
      extractionKit: string
      cancerStage: string
      diseaseCategory: string
      customDiseaseCategory: string
      reason: string
      patientId: string
      orderingPhysician: string
      receiveDate: string
      dateOfBirth: string
      diagnosis: string
      sampleOrigin: string
      editAndAmend: boolean
      customCancerType: string
      cancerTypeDisplay: string
      sixteensrRNA: string
      targetSpecies: string
      libraryChemistry: string
    }
  }
]
```

```

        caseId:                string
        metaData:              {
                                }
        category:              string
        dnaBamFile:            string
        rnaBamFile:            string
        tnaBamFile:            string
    }
}
}

```

Get a list of signed-off samples

The `getSignedOffSamples` API returns a list of the samples that are designated with **(Signed Off)**.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is GET `/api/signedOffSamples`.

CURL command syntax

```

curl -X GET "https://<host>/service/api/signedOffSamples?
signedOff=&start_date=&end_date=&locked=&pageNumber=&pageSize="
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"

```

Query parameters

Item	Description
<code>signedOff</code>	Flag to filter signed off results. The default is true. Boolean. <i>Required</i> .
<code>start_date</code>	Samples to fetch from date. The default is null. String. <i>Optional</i> .
<code>end_date</code>	Samples to fetch up to date. The default is null. String. <i>Optional</i> .
<code>locked</code>	Flag to filter samples that are signed with a locked signature. The default is "true". Boolean. <i>Optional</i> .
<code>pageNumber</code>	Starting page number. The default value is page 1. Integer (int32). <i>Optional</i> .
<code>pageSize</code>	Number of items to fetch. The default value is 10. String. <i>Optional</i> .

Response

```
{
  total:      integer (int64)
              total number of signed off samples
  objects:    [
                {
                  assayName:      string
                  baseUrl:         string
                  checksumFile:    string
                  createDate:      string
                  sampleFile:      string
                  sampleId:        string
                  sampleName:      string
                  planName:        string
                  resultId:        integer (int32)
                  limsDirectory:   string
                }
              ]
}
```

Update samples

The `updateSamples` API updates one or more samples. All or None is followed for all samples in a request. If there are multiple records to update, any issue with one record causes the remaining records to be skipped.

The authenticator token is required for use in all API requests, and limits the responses based on the name of the organization of the API requesting user.

The method to use is PUT `/api/samples`.

CURL command syntax

```
curl -X PUT "https://<host>/service/api/samples"
-H "Authorization: [[Auth token which starts as Token, for example, Token
xyz...]]"
-H "mode: AssayDev"
-H "Accept: application/json"
-H "Content-Type: application/json" --data '<input payload>'
```

Body parameters

```
▼{
  List of Sample Lims DTO
  Required: objects
  total:    [] integer (int64)
            total number of samples
  objects:  [ (1..2147483647)
              List of sample DTO's
              { []
                Sample Lims DTO
                Required: collectionDate, sampleId
                otherAttributes: {
                                }
                tags:            [

```

```

        string
    ]
    id: integer (int32)
    sampleId: string maxLength:50
    collectionDate: string
    gender: string
    notes: string
    sampleType: string
    tumorCellularity: string
    necrosis: string
    origin: string
    customOrigin: string
    extractionKit: string
    cancerStage: string
    diseaseCategory: string
    customDiseaseCategory: string
    reason: string
    patientId: string
    orderingPhysician: string
    receiveDate: string
    dateOfBirth: string
    diagnosis: string
    sampleOrigin: string
    editAndAmend: boolean
    customCancerType: string
    cancerTypeDisplay: string
    sixteensrRNA: string
    targetSpecies: string
    libraryChemistry: string
    caseId: string
    metaData: {
    }
    category: string
    dnaBamFile: string
    rnaBamFile: string
    tnaBamFile: string
  }
]
}

```

Response

```

▼{
List of Sample Lims DTO
Required: objects
total: [] integer (int64)
      total number of samples
objects: [ (1..2147483647)
          List of sample DTO's
          { []
            Sample Lims DTO
            Required: collectionDate, sampleId
            otherAttributes: {
            }
            tags: [
                    string
                  ]
            id: integer (int32)
            sampleId: string maxLength:50
            collectionDate: string
          }
        ]
}

```

```

        gender:                string
        notes:                 string
        sampleType:            string
        tumorCellularity:      string
        necrosis:              string
        origin:                string
        customOrigin:          string
        extractionKit:         string
        cancerStage:           string
        diseaseCategory:       string
        customDiseaseCategory: string
        reason:                string
        patientId:             string
        orderingPhysician:     string
        receiveDate:           string
        dateOfBirth:           string
        diagnosis:             string
        sampleOrigin:          string
        editAndAmend:          boolean
        customCancerType:      string
        cancerTypeDisplay:     string
        sixteensrRNA:          string
        targetSpecies:         string
        libraryChemistry:      string
        caseId:                string
        metaData:              {
                                }
        category:              string
        dnaBamFile:            string
        rnaBamFile:            string
        tnaBamFile:            string
    }
}

```



Programming guidelines for Genexus™ Software plugins

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This document explains design specifications for creating a plugin that can be executed on the Genexus™ Software 7.0 platform. This document specifies the mandatory files and folder structure for the plugin. The installation, validation, and execution steps are also specified, along with example screenshots to help develop the plugin.

Steps to develop a plugin

Plugin package content

The plugin is packaged in the form of a ZIP file. The asset ZIP file can include one or more plugin folders.

Asset ZIP file

The `ConfigurableDemoPluginAsset.zip` file content must be structured as shown.

```
|-- ConfigurableDemoPluginAsset
|-- ConfigurableDemoPluginAsset/ConfigurableDemoPlugin.zip
|-- ConfigurableDemoPluginAsset/Plugin_Example_EULA_PDF.pdf
|-- ConfigurableDemoPluginAsset/asset-metadata.json
|-- ConfigurableDemoPluginAsset/README.md
```



File name	Description	Mandatory
ConfigurableDemoPlugin.zip	The plugin ZIP file that is installed in Genexus™ Software. At least one plugin ZIP file should be present inside of the asset ZIP file.	Yes
asset-metadata.json	The JSON file that contains metadata for the plugin ZIP file that needs to be installed. Do not change the file name.	Yes
Plugin_Example_EULA_PDF.pdf	The EULA PDF file for the asset (can have all plugin EULAs combined).	No
README.md	Read me file. Do not change the file name.	No

Plugin ZIP file

The ConfigurableDemoPlugin.zip file content must be structured as shown.

```
|-- appmeta.json
|-- build
|   |-- launcher.sh
|   |-- ConfigurableDemoPlugin_env.tar
|-- executePlugin.py
|-- gx_plan.html
|-- css
|   |-- style.css
|-- js
|   |-- jquery-1.6.1.min.js
|   |-- jquery-ui-1.8.20.custom.min.js
|   |-- jquery.rightClick.js
|   |-- kendo.custom.min.js
```

File name	Description	Mandatory
appmeta.json	The JSON file that contains metadata for the plugin. Do not change the file name.	Yes
build/launcher.sh	The script that gets executed while running a plugin on Genexus™ Software. This acts as an initial entry point. Do not change the file name.	Yes
gx_plan.html	This file can be used to supply parameter values for plugin execution. Do not change the file name.	No
js (folder)	The JavaScripts, if needed, for the HTML files to function, for example, jquery, and so on.	No
css (folder)	The CSS files, if needed, to apply styles to HTML files.	No



Files and format

asset-metadata.json **file**

The following is an example of an asset metadata ZIP file that contains two plugin ZIP files, for example, Plugin_Example_1.zip and Plugin_Example_2.zip.

Note: The version and the name of the plugin list in the asset-metadata.json file should exactly match each plugin in the appmeta.json file.

```
{
  "category": "Apps",
  "asset_version": "0.0.1",
  "instrument": "Genexus",
  "eulaUri": "Plugin_Example_0.0.1/Plugin_Example_EULA_PDF.pdf",
  "plugin_list": [
    {
      "name": "Plugin_Example_1",
      "version": "1.0.0",
      "format": "zip"
    },
    {
      "name": "Plugin_Example_2",
      "version": "1.0.0",
      "format": "zip"
    }
  ]
}
```

Key	Description	Example Value	Is mandatory
category	Category of the asset.	"Apps" – DO NOT CHANGE	Yes
asset_version	Version for the asset.	"0.0.1"	Yes
eulaUrl	EULA document url.	<asset-zip_version>/<eula-document>	No
plugin_list	JSON format for a list of plugins that the asset has. Must follow the example format.	[{ "name": "<plugin-name>", "version": "<version>", "format": "zip" }]	Yes



appmeta.json file

Each plugin ZIP file should have its own appmeta.json file as follows.

```
{
  "name": "ConfigurableDemoPlugin",
  "version": "0.0.1",
  "description": "ConfigurableDemoPlugin using algorithm Jammy for
reference.",
  "runAllowed": "SAMPLE",
  "pluginLauncher": "build/launcher.sh",
  "dependsOn": [
    {
      "pluginName": "coverageAnalysis",
      "versionRange": "5.18.0.0 - 5.22.0.0"
    }
  ],
  "image_name_with_tag": "",
  "configurable": true,
  "lowest_supported_sw_version": "7.0",
  "validWorkflowAppTypes": [
    "DNA",
    "RNA",
    "DNA_RNA"
  ],
  "inputs": [
    "analysisDir",
    "pluginOutputDir"
  ],
  "embeddedOutput": false,
  "executeDxAdapter": true
}
```

Attribute name	Description	Possible values	Mandatory
name	The name of the plugin.		Yes
version	This attribute contains the plugin version and should be kept updated for every update in the plugin code.		Yes
description	A description for the plugin.		No
runAllowed	- SAMPLE—If the value is set as Sample, then the plugin is executed for each sample within the run. The plugin output is shown only at the sample level view. - ASSAY—If the value is set as Assay, then the plugin is executed one time for each assay within the run. The plugin output is shown at the assay level results view.	SAMPLE or ASSAY	Yes



(continued)

Attribute name	Description	Possible values	Mandatory
pluginLauncher (System Required Attribute)	This attribute shows the entry point of the launcher script for the plugin. The folder and file must be the same as the example.	"build/launcher.sh"	Yes
dependsOn	This attribute accepts an array of plugin names that must be executed before this plugin can be executed.		No
	Use case	Example	
	Plugin is dependent on the latest active version of another plugin.	<pre>"dependsOn": [{ "pluginName": "coverageAnalysis" }]</pre>	
	Plugin is dependent on the version of the plugin that is indicated by the version number that appends the plugin name.	<pre>"dependsOn": [{ "pluginName": "coverageAnaly- sis_0.0.1" }]</pre>	
	Plugin is dependent on a range of versions of another plugin. Boundary values are included.	<pre>"dependsOn": [{ "pluginName": "coverageAnalysis", "versionRange": "0.0.1-0.0.3" }]</pre>	



(continued)

Attribute name	Description	Possible values	Mandatory
image_name_with_tag	• If the plugin is using the already installed Docker™ program, then use this attribute to specify the image name and tag. • Attribute is not found, or value is empty, – If the Docker™ TAR file is found in the plugin bundle, the same Docker™ file is installed and used for plugin execution. – If the Docker™ TAR file is missing, the assay image is used during plugin execution.	Example format: <image-name>:<tag>	No
configurable	This option marks the plugin as configurable. The user interface shows the configure link that can take configurable parameters and can be used in plugin execution.	true or false	No
lowest_supported_sw_version	The plugin is supported on the earliest version of software.	7.0/6.8	No
validWorkflowAppTypes	• DNA—The plugin is listed and executed only for DNA assays. • RNA—The plugin is listed and executed only for RNA assays. • DNA_RNA—The plugin is listed and executed for DNA and Fusions assays. • Blank—If no value is specified, the plugin is listed for all assay types.	DNA / RNA / DNA_RNA / blank	No
inputs	Keys specified in this attribute are set as an environment variable that can be read by the plugins.	Example : <pre>"inputs": ["analysisDir", "pluginOutputDir"]</pre>	No



(continued)

Attribute name	Description	Possible values	Mandatory
embeddedOutput	- If this attribute is set to true, the output of the plugins, that is, HTML pages, is embedded on the user interface. The page cannot be opened in a new browser tab. - If this attribute is set to false, the output of the plugins, that is, HTML pages, is shown as a link. to open the page in a new browser tab.	true or false	No
executeDxAdapter (System required attribute)	This attribute should always be set to true if the plugin code is dependent on the startplugin.json and the barcodes.json file for the inputs.	true or false	No
amplificationType	Amplification type that the plugin supports.	ampliseqhd or ampliseq or blank	No
researchApplication	The research application that the plugin supports.	DNA /RNA / DNA+RNA / blank	No
applicationCategory	The application category that the plugin supports.	DNA+RNA /DNA /RNA / Microbiome_Analysis / ANEUPLOIDY / blank	No

launcher.sh file

```
#!/bin/bash
ANALYSIS_SAMPLE_DIR=$1
DIRNAME=$2
SAMPLE_PLUGIN_DIR=$3
export DIRNAME=${DIRNAME}
export CHIP_LEVEL_ANALYSIS_PATH=${ANALYSIS_SAMPLE_DIR}
VERSION=`grep -ir 'lscope-setenv.sh' ${ANALYSIS_SAMPLE_DIR}/run.sh | awk '{print $3}'`
bash /share/apps/IR/ionreporter${VERSION}/bin/lscope-setenv.sh ${VERSION}
bash /share/apps/IR/ionreporter${VERSION}/bin/app-env.sh ${VERSION}
cd ${SAMPLE_PLUGIN_DIR}
python ${DIRNAME}/executePlugin.py
```

The last line is the launcher script for the plugin to be executed.

Examples:

1. python \${DIRNAME}/executePlugin.py
2. bash \${DIRNAME}/executePlugin.sh



Runtime environment

- For plugins that use an installed Docker™ image as the runtime environment, change `appmeta.json` to add key “image_name_with_tag”: `<available_image:tag>`.
- For plugins that use your own Docker™ image as the runtime environment:
 - Create a Docker™ image and export it as a TAR file. See “Docker™ image creation” on page 523
 - Add a TAR image, for example, `ConfigurableDemo_env.tar` that was created in the previous step, inside the build folder. Only a TAR file is accepted.

```
|-- build
|   |-- launcher.sh
|   |-- ConfigurableDemoPlugin_env.tar
```

Docker™ TAR is an execution environment. Supporting files and executables must be part of it.

- For plugins that use an assay Docker™ image as the runtime environment:
 - Keep the value of “image_name_with_tag” as blank, or exclude the key from the `appmeta` file.
 - Do not include any Docker™ TAR file in the build folder.

How to accept user input

If a plugin needs user input values to be used while running, add a configuration file named `gx_plan.html` in the plugin package. Ensure to have “configurable”:True in the `appmeta.json` for the plugin.

Create a `gx_plan.html` file

- Optional css to match Genexus styling.
`<link rel="stylesheet" href="/genexus/styles/*.css" media="all" onload="this.media='all'">`
- Mandatory script in the body.

```
<script type="text/javascript">
  var TB_result=parent.TB_result;
  var TB_plugin=parent.TB_plugin;
  var GX_server=parent.GX_server;
  var GX_CSS= parent.GX_CSS;
  var TB_result_json=parent.TB_result_json;
  var TB_plan_json=parent.TB_plan_json;
  var assayId=parent.assayId;
  var expId=parent.expId;
  var token=parent.token;
</script>
```

- Add a control that captures user inputs (text box, checkbox, select box, and so on).
- Any details related to the run or plugin to be shown on this configuration user interface can be done by using the APIs exposed.
See “Genexus™ Software APIs to be used in configuration” on page 517.



- Must contain a **Submit** button that triggers data submission to the POST API.
See “savePluginConfig API” on page 517.
- The data submitted to the API should be in the required JSON format.
See “savePluginConfig API” on page 517.

Configure the plugin

1. Use one of these methods to configure the plugin.
 - Configure when you create or edit an assay.
 - a. Go to the **Plugin** step and select the plugin that is configurable.
 - b. Click the configuration link.
 - c. On the `gx_plan.html` configuration page, update the parameter values. These values are valid only for the assay and do not override the default configuration values set for the assay.
 - Configure when you create or edit a run plan.
 - a. Go to the **Plugin** step that is based on the assay selected that has a configurable plugin.
 - b. Click the configuration link on the plugin.
 - c. On the `gx_plan.html` configuration page, update the parameter values. These values are valid only for the run plan and do not override the default configuration values set for the assay.
 - Configure when you rerun the plugin in the **Results** page.
 - a. Go to **Results ▶ Run Results**.
 - b. Select an assay, click the **Plugins** tab, then click **Select Plugins**.
 - c. Select a plugin that is configurable.
 - d. On the `gx_plan.html` configuration page, update the parameter values. These values are valid only for execution of that plugin.
2. After you provide values for the parameters, click **Save**.

A JSON file is created with the configuration details and submitted to the POST API, that saves it to the database for the assay, plugin, and experiment. See “savePluginConfig API” on page 517.

- The POST API to save the configuration from the user interface:
`/genexus/secure/api/plugins/savePluginConfig?expld=expld`
- Example usage:
`pluginURL = "/genexus/secure/api/plugins/savePluginConfig?" + (expld ? "expld=" + expld : "");`

The GET API can be used to prepopulate the values on the form based on the configuration that is available. See “getPluginConfig API” on page 519.

- `/genexus/secure/api/plugins/getPluginConfig?assayId=assayId&pluginId=pluginId&expld=expld`
- Example usage:
`url: '/genexus/secure/api/plugins/getPluginConfig?assayId=' + assayId + '&pluginId=' + pluginId + (ex-pld ? '&expld=' + expld : ''),`

After the plugin is configured, a run can be initiated, or the plugin can be rerun.



After the run is initiated to execute the plugin, the software creates 2 files in the plugin output folder.

- `startplugin.json`—A JSON file that contains all data related to the run. The file also contains the configuration that was set while configuring the plugin, under “plan”-> “pluginconfig”.
- `barcodes.json`—A JSON file that contains barcode-related information that is used in the run.

While the plugin is executing, the plugin reads these files in the current output folder using the file names `startplugin.json` and `barcodes.json`.

Expected output files after plugin execution

After plugin execution is complete, two files are generated in the plugin output folder.

- `<plugin-name_version>_block.html`
 - `<plugin-name_version>_block.html` is considered the output of plugin execution and is displayed on the user interface in the **Results** page in the **Plugins** tab.
 - This file can include the CSS file from Genexus™ Software to comply with the software user interface appearance.
For example: `<link rel="stylesheet" href="/genexus/styles.*.css" media="all" onload="this.media='all'">`.
 - Additional CSS files can be added on top of the default Genexus™ Software CSS file.
- `<plugin-name_version>.log`
 - This log file contains plugin execution-related logs.
 - This file is available on the user interface on the **Results** page in the **Plugins** tab. Click **View Log**.

In addition to the above files, the required css/js files referenced in the `<plugin-name_version>_block.html` file must also be present in the plugin output folder.

Do not use soft link js /css files. Instead, copy the required files in the plugin output folder. Users cannot view plugin results on the user interface in the proper format if the plugin is uninstalled.

The following is an example of the contents of the plugin output folder.

```
ConfigurableDemoPlugin_0.0.1/
|-- 1720512568857
|   |-- ConfigurableDemoPlugin _0.0.1.log
|   |-- ConfigurableDemoPlugin _0.0.1_block.html
|   |-- barcodes.json
|   |-- context.json
|   |-- docker.env
|   |-- in.json
|   |-- log.err
|   |-- log.out
|   |-- params.json
|   |-- js/
|   |-- css/
|-- startplugin.json
```




Genexus™ Software APIs to be used in configuration

savePluginConfig API

Description—Save the configuration from the configuration page.

Method—POST

URL— /genexus/secure/api/plugins/savePluginConfig?expId=expId

URL parameter

Parameter	Mandatory	Example
expId	No	expId DO NOT CHANGE

Request payload

Parameter	Mandatory	Value
assayId	Yes	assayId DO NOT CHANGE
pluginId	Yes	TB_plugin.pk DO NOT CHANGE
Config	Yes	JSON format

Example output JSON

```
{
  "assayId": assayId,
  "pluginId": TB_plugin.pk,
  "config": {
    "ext_config": { },
    "minFamilySize": "3",
    "SRTestSample02": {
      "input-1": "SRTS2_1",
      "input-2": "SRTS2_2"
    },
    "minFunctionalCov": "5",
    "runBidirectional": false,
    "minVariantFamilies": "4113"
  }
}
```



Plugins

Configure ConfigurableDemoPlugin_0.0.1

Configure Demo Plugin Parameters

Min Functional Family Size

Min Number of Variant Families

Min Functional Coverage

Run Bi-directional sequencing Analysis ☐

External Configure File for:

Samplewise Configurations:

Sample Name: SRTestSample02

Param1

Param2

Figure 12 Example screenshot

getSamplesForRun API

Description—Get the list of samples used in a plan run while creating or rerunning a plugin.

Method—GET

URL—/genexus/secure/api/specimen/getSamplesForRun?
expId=expId&assayId=assayId

URL parameters

Parameter	Mandatory	Example
expId	No	expId DO NOT CHANGE
assayId	Yes	assayId DO NOT CHANGE

Example output JSON

```
{
  "sampleId": "Sample1",
  "nucleicAcidType": "DNA"
}
```



Figure 13 Example screenshot

getPluginConfig API

Description—Get saved plugin config data based on experiment ID.

Method—GET

URL—/genexus/secure/api/plugins/getPluginConfig?assayId=assayId&pluginId=<TB_plugin.pk>&expId=expId

URL parameters

Parameter	Mandatory	Example
expId	No	expId DO NOT CHANGE
assayId	Yes	assayId DO NOT CHANGE
pluginId	Yes	pluginId DO NOT CHANGE

Example output JSON

```
{
  "ext_config": {},
  "minFamilySize": "3",
  "SRTestSample02": {
    "input-1": "SRTS2_1",
    "input-2": "SRTS2_2"
  },
  "minFunctionalCov": "5",
  "runBidirectional": true,
  "minVariantFamilies": "4",
  "message": "Plugin configuration is retrieved from last save."
}
```



Configure Demo Plugin Parameters

Plugin configuration is retrieved from last save.

Min Functional Family Size: 3

Min Number of Variant Families: 4

Min Functional Coverage: 5

Run Bi-directional sequencing Analysis: ☒

External Configure File for bio: [Load external parameter file](#)

Samplewise Configurations:

Sample Name: SRTestSample02

Param1: SRTS2_1

Param2: SRTS2_2

[Save & Close](#)

Figure 14 Example screenshot with values

getPluginConfigFromAssay API

Description—Get plugin configuration from assay, if any.

Method—GET

URL—/genexus/secure/api/plugins/getPluginConfigFromAssay?
assayId=assayId&pluginId=<TB_plugin.pk>

URL parameters

Parameter	Mandatory	Example
assayId	Yes	assayId DO NOT CHANGE
pluginId	Yes	TB_plugin.pk DO NOT CHANGE

Example output JSON

```
{
  "minFamilySize": "3",
  "SRTestSample02": {
    "input-1": "SRTS2_1",
    "input-2": "SRTS2_2"
  },
  "minFunctionalCov": "5",
  "runBidirectional": true,
  "minVariantFamilies": "4",
}
```



pluginResults API

Description—Get result data, for example, experiment status, runId, name, and so on.

Method—GET

URL—/genexus/secure/api/plugins/results/{resultId}/pluginResults

URL parameter

Parameter	Mandatory	Example
resultId	Yes	resultId DO NOT CHANGE

Example output JSON

```
{
  "id": 130,
  "analysisVersion": "6.99.60.0",
  "bamLink": null,
  "pluginState": {
    "ConfigurableDemoPlugin": "COMPLETED",
    "generateGenome": "COMPLETED",
    "sampleID": "COMPLETED",
    "coverageAnalysis": "COMPLETED",
    "molecularCoverageAnalysis": "COMPLETED"
  },
  "planShortID": "5FNUC",
  "pluginStore": {
    "generateGenome": {},
    "sampleID": {},
    "coverageAnalysis": {},
    "molecularCoverageAnalysis": {}
  },
  "reference": "hg19",
  "resultsName":
  "Auto_R_2021_09_24_14_44_33_ionadmin_GNXS-0207-5-24SEP2021_0207_1_EEM_VHL_VIQ_
  _cftNA_2_15ng_L2_74_130",
  "runid": "",
  "status": "Completed",
  "reportLink": "/results/ValkyrieTestRig/cropped_auto_hawkeyeHD_reseq_01/",
  "filesystempath": null,
  "pluginresults": [ ],
  "resource_uri": "/rundb/api/v1/results/130"
}
```



getRunType API

Description—Get all predefined run types for plugins.

Method—GET

URL— /genexus/secure/api/plugins/getRunType

Example output JSON

```
{
  "objects": [
    {
      "description": "AmpliSeq HD - DNA and Fusions (Single Library)",
      "nucleotideType": "dna",
      "runType": "AMPS_HD_DNA_RNA_1",
      "alternate_name": "AmpliSeq HD - DNA and Fusions (Single Library)"
    }
  ]
}
```

getAssayRunType API

Description—Get assay run type for the given assayId

Method—

URL— /genexus/secure/api/assay/getAssayRunType?id=assayId

URL parameter

Parameter	Mandatory	Example
assayId	Yes	assayId DO NOT CHANGE

Example output JSON

```
"AMPS_HD_DNA_RNA_1"
```



User interface for plugin output

After all plugins are executed, you can see the output of the plugins on the **Sample Results** page or **Run Results** page under the **Plugins** tab. Each plugin output generates all output files including mandatory `<plugin-name>_block.html` and `<plugin-name>.log`. The files are shown on the user interface.

This `<pluginname_version>_block.html` file is shown as output on the **Plugins** tab.

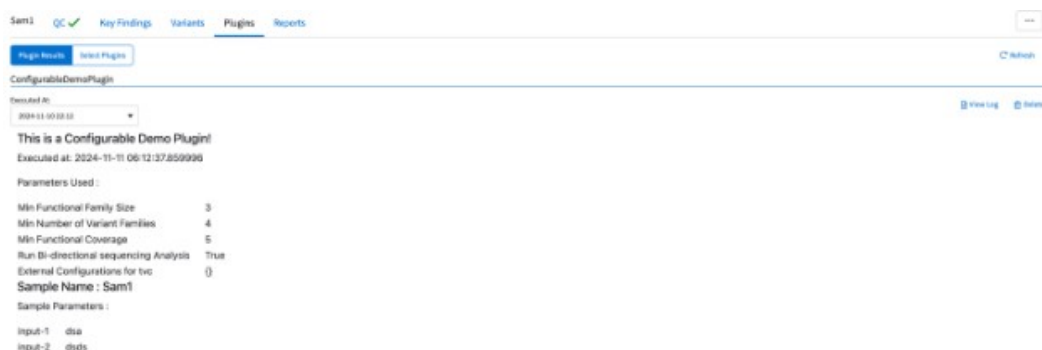


Figure 15 Example Plugin Results screen

The `<plugin-name_version>.log` file shows relevant logs for the executed plugin. Click **View Log** on the **Plugins** tab.

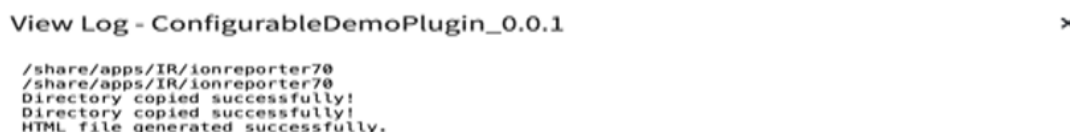


Figure 16 Example log file screenshot

Docker™ image creation

Docker™ is a platform designed to help developers build, share, and run container applications.

The container is executed as user `iruser`. The end user needs to add user `iruser` and group `irgrp` when creating an image. Add the following lines in the Docker™ file text document that contains the commands that a user could call on the command line to assemble an image.

The following syntax is compatible with the distribution of python:3.11.5-slim-bookworm.

```
# create runtime user: iruser and group irgrp
RUN groupadd -g 1002 irgrp && useradd -u 1001 -m iruser -g irgrp -d /home/
iruser -s /bin/bash && \
chown -R iruser:irgrp /home/iruser && \
sed -i 's@^iruser:.*:@iruser:!!:15854:0:99999:7:::@g' /etc/shadow
```

Note: The syntax for adding user and group may be different for different distributions. You need to use the syntax for the distribution that you selected for your image.



Use the following command to create a Docker™ image TAR file.

```
docker save -o ConfigurableDemoPlugin_env.tar ConfigurableDemoPlugin:latest
```




Reference information

■ Rules for variant names in Genexus™ Software	525
■ Annotation criteria for the OncoPrint™ Variant Annotator	527
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■ System-installed barcode sets	550
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Rules for variant names in Genexus™ Software

Genexus™ Software provides standard conventions for the names of genomic variants. Standard variant names are derived from sample and results data and properties that are available in the software for each variant, such as variant, gene name, and amino acid change. The conventions are used for both de novo variants and variants that are included in a hotspots file. Variant names are shown in the **Variant Name** column of variant tables, in the gene tiles in the **Key Findings** tab, and in the reports generated by the software.

Summary of variant types and examples of standardized variant names

Genexus™ Software applies a set of rules to a variant, then assigns or returns a name for the variant that best describes variant details. The following table describes the rules that are used to name variants.

Variant type	Description	Variant name examples
Variants with protein change	Variants that provide an amino acid change syntax.	<i>BRAF</i> V600E <i>BRAF</i> p.(V600E) c.1799A>C
Variants with coding change but no protein change	Variants that provide a coding change, but for which an amino acid is missing or is p . ?.	<i>MET</i> c.2942-29_2942-28delinsGG
Oncomine class-based variants	Any variant with a variant type of <i>SNV</i> or <i>INDEL</i> at the locus based on Oncomine™ annotations. These changes are typically complex and are frequently referenced in scientific literature.	<i>EGFR</i> exon 19 deletion <i>MET</i> exon 14 skipping
<i>EGFR</i> exon 19 insertion variants	Variant with <code>Function="nonframeshiftInsertion"</code> in exon 19 of <i>EGFR</i> gene.	exon 19 insertion
Copy number variants	Copy number gain or loss, signified by a variant type of <i>CNV</i> .	<i>ALK</i> amplification <i>PTEN</i> deletion
RNA exon variants	Specific RNA exon variants, identified by variant ID.	<i>MET</i> exon 14 skipping <i>EGFRvIII</i>
Long deletion Variants	Oncomine Variant Class equals <i>LongDel</i> and length attribute is a positive integer.	<i>BRCA1</i> 40 base deletion
Exon deletions	A special type of <i>CNV</i> that impacts <i>BRCA</i> genes only. These variants are identified by a type of <i>CNV</i> , a subtype of <i>BigDel</i> , and call details that specify the exon range affected.	<i>BRCA1</i> exon 1 deletion
Exon duplications	A special type of <i>CNV</i> that impacts <i>BRCA</i> genes only. These variants are identified by a type of <i>CNV</i> , a subtype of <i>BigDup</i> , and call details that specify the exon range affected.	<i>BRCA1</i> exon 1 duplication
Chromosome arm-level alterations	Copy-number alterations occurring at the length of chromosome arms. These variants are identified by a type of <i>CNV</i> and a subtype of <i>ARM</i> .	9p amplification Xq deletion
Targeted fusions	Fusions detected by targeting a specific breakpoint.	<i>SLC34A2-ROS1</i> fusion
Non-targeted fusions	Non-targeted fusions	<i>SLC45A3-ETV1</i> fusion

(continued)

Variant type	Description	Variant name examples
Expression imbalance variants	Variants for which the OncoPrint Variant Class is "ExpressionImbalance"; . This is measured by the partner-agnostic detection of fusions in driver genes using imbalance in exonic expression.	<i>MET</i> fusion
All other mutations	Mutation variants that do not match the criteria of other variants in this table.	<i>ADAMTSL4-AS1</i> mutation
Sample MSI	Sample-level microsatellite stability/instability. This is a measure of the entire sample instead of an individual variant.	Microsatellite instability-High Microsatellite stable
Expression control variants	Variants with SVTYPE=ExprControl.	<i>LMNA</i> expression control
Gene expression variants	Variants with SVTYPE=GeneExpression.	<i>MET</i> expression
Sample TMB	Sample-level tumor mutational burden. This is a measure of the entire sample instead of an individual variant.	Tumor Mutational Burden
Sample LOH	Sample-level loss of heterozygosity. This is a measure of the entire sample instead of an individual variant.	Loss of Heterozygosity
Sample TAI	Sample-level telomeric allelic imbalance. This is a measure of the entire sample instead of an individual variant.	Telomeric Allelic Imbalance
Sample LST	Sample-level large-scale state transition. This is a measure of the entire sample instead of an individual variant.	Large-Scale State Transition
Gene-level LOH Variants	OncoPrint Variant Class equals "LOH".	<i>BARD1</i> LOH

Annotation criteria for the OncoPrint™ Variant Annotator

In Genexus™ Software, analysis results are the final results generated by a run analysis. The analysis results include run details, all BAM files, filtered results, graphs, run reports, metrics, plugin results, annotated variant data, and VCF files.

The OncoPrint™ Variant Annotator is a software module that annotates variant data relevant to cancer, based on analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types.

When you create or copy and edit an assay in Genexus™ Software, you have the option to apply an **OncoPrint™ Variant Annotations Ruleset**. You can select a version of the ruleset that matches the version of the analysis pipeline that is used for the assay. The OncoPrint™ Variant Annotator then uses

the selected ruleset as an annotation reference. For information about how to apply an OncoPrint™ variant annotation ruleset to an assay, or remove the ruleset, see “Panel step assay options” on page 240.

This section summarizes the requirements that must be met for the OncoPrint™ Variant Annotator to annotate variants in results for each OncoPrint™ Research Assay and Ion AmpliSeq™ HD Panel.

For each variant type in these tables, the OncoPrint™ Variant Annotator annotates a variant only if all conditions in the corresponding Annotation Criteria column are satisfied. The annotations are shown in results in the **OncoPrint™ Gene Class** and **OncoPrint™ Variant Class** columns.

You can find all relevant annotation criteria in VCF files.

OncoPrint™ BRCA Assay GX

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number deletion	Loss-of-Function	Deletion	- SVTYPE = "CNV" - FILTER = "LOSS" - SUBTYPE = "GeneCNV"
Copy number exon deletion	Loss-of-Function	ExonDeletion	- SVTYPE = "CNV" - FILTER = "LOSS" - SUBTYPE = "BigDel"
Copy number exon duplication	Loss-of-Function	ExonDuplication	- SVTYPE = "CNV" - FILTER = "GAIN" - SUBTYPE = "BigDup"
Long deletion	Loss-of-Function	Truncating	- Positive mutation call - SVTYPE = "LongDel"
Loss-of-function truncating de novo mutation	Loss-of-Function	Truncating	- Positive mutation call - Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense - Occurs in a designated loss-of-function gene
Missense hotspot mutation	Loss-of-Function	Hotspot	- Positive mutation call - Functional impact is missense - Transcript and codon position occur in predefined missense hotspot list
Splice site hotspot mutation	Loss-of-Function	Hotspot	- Positive mutation call - Transcript, location, and exon occur in predefined splice site hotspot list

OncoPrint™ Comprehensive Assay Plus GX

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number amplification	Gain-of-Function	Amplification	- SVTYPE = "CNV" - FILTER = "GAIN" - Occurs in a designated copy-gain gene
Copy number deletion	Loss-of-Function	Deletion	- SVTYPE = "CNV" - FILTER = "LOSS" - Occurs in a designated copy-loss gene
Copy number exon deletion	Loss-of-Function	ExonDeletion	- SVTYPE = "CNV" - FILTER = "LOSS" - SUBTYPE = "BigDel"
Copy number exon duplication	Loss-of-Function	ExonDuplication	- SVTYPE = "CNV" - FILTER = "GAIN" - SUBTYPE = "BigDup"
Gene fusion	Gain-of-Function	Fusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Is a targeted fusion isoform
Non-targeted fusion	Gain-of-Function	NonTargetedFusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Non-targeted field is present - NT_FUSION_IN_FRAME is not FALSE - NT_FUSION_DRIVER_INVOLVED is not FALSE - NT_FUSION_SECONDARY is not TRUE
RNA exon variant	Gain-of-Function	RNAExonVariant	- SVTYPE = "RNAExonVariant" or "Fusion" - FILTER = "PASS" - Is a targeted RNA exon variant
Expression imbalance	Gain-of-Function	ExpressionImbalance	- SVTYPE = "RNAExonTiles" - FILTER = "PASS" - Record meets Targeted Isoforms Detected Requirement
Long deletion	Loss-of-Function	Truncating	- Positive mutation call - SVTYPE = "LongDel"

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Loss-of-function truncating de novo mutation	Loss-of-Function Unclassified	Truncating	- Positive mutation call - Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense - Occurs in a loss-of-function or unclassified gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Functional impact is missense - Transcript and codon position occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Transcript, location, and exon occur in predefined splice site hotspot list
Intronic hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and ID occur in predefined intronic hotspot list
Synonymous hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Functional impact is synonymous - Transcript and coding syntax occur in predefined synonymous hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a loss-of-function gene

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and coding syntax occur in MNV hotspot list
EGFR exon 19 deletion ^[1]	Gain-of-Function	EGFRExon19Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 744–761 of EGFR^[2]
EGFR exon 20 insertion ^[1]	Gain-of-Function	EGFRExon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion or nonframeshift block substitution - Insertion impacts codons 762–775 of EGFR or variant is in EGFR exon 20 insertion confirmed list^[2]
ERBB2 exon 20 insertion ^[1]	Gain-of-Function	ERBB2Exon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 770–783 of ERBB2^[2]
JAK2 exon 12 INDEL ^[1]	Loss-of-Function	JAK2Exon12Indel	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, or nonframeshift block substitution - INDEL impacts codons 536–547 of JAK2^[2]
KIT exon 11 deletion ^[1]	Gain-of-Function	KITExon11Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, or nonframeshift block substitution - Deletion impacts codons 550–580 of KIT^[2] - Alternatively, mutation occurs in splice site flanking the 5' end of exon 11
KIT exon 11 insertion ^[1]	Gain-of-Function	KITExon11Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, or nonframeshift block substitution - Insertion impacts codons 550–580 of KIT^[2]

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
MET exon 14 skipping ^[1]	Gain-of-Function	METExon14Skipping	- Positive mutation call - Location is splice site in MET exon 14, is intronic >= 4bp deletion in 30 nucleotides preceding MET exon 14, or variant is in MET Exon 14 Skipping confirmed list
PDGFRA exon 18 deletion ^[1]	Gain-of-Function	PDGFRAExon18Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, or nonframeshift block substitution - Deletion impacts codons 841–847 of PDGFRA^[2]

^[1] For more information, see “Ion Torrent™ class-based variants” on page 544.

^[2] In some cases the assay design may not cover the entire codon range.

OncoPrint™ Comprehensive Assay v3 GX

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number amplification	Gain-of-Function	Amplification	- SVTYPE = "CNV" - FILTER = "GAIN" - Occurs in a designated copy-gain gene
Long deletion	Loss-of-Function	Truncating	- Positive mutation call - SVTYPE = "LongDel"
Gene fusion	Gain-of-Function	Fusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Is a targeted fusion isoform
Nontargeted fusion	Gain-of-Function	NonTargetedFusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Non-Targeted field is present - NT_FUSION_IN_FRAME is not FALSE - NT_FUSION_DRIVER_INVOLVED is not FALSE - NT_FUSION_SECONDARY is not TRUE
RNA exon variant	Gain-of-Function	RNAExonVariant	- SVTYPE = "RNAExonVariant" or "Fusion" - FILTER = "PASS" - Is a targeted RNA exon variant

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Loss-of-function truncating de novo mutation	Loss-of-Function	Truncating	- Positive mutation call - Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense - Occurs in a loss-of-function gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Functional impact is missense - Transcript and codon position occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript, location, and exon occur in predefined splice site hotspot list
Intronic hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and ID occur in predefined intronic hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and coding syntax occur in MNV hotspot list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
EGFR exon 19 deletion ^[1]	Gain-of-Function	EGFRExon19Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 744–761 of EGFR^[2]
EGFR exon 20 insertion ^[1]	Gain-of-Function	EGFRExon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 762–775 of EGFR or variant is in EGFR exon 20 insertion confirmed list^[2]
ERBB2 exon 20 insertion ^[1]	Gain-of-Function	ERBB2Exon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 770–783 of ERBB2^[2]
KIT exon 11 deletion ^[1]	Gain-of-Function	KITExon11Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 550–580 of KIT^[2] - Alternatively, mutation occurs in splice site flanking the 5' end of exon 11
KIT exon 11 insertion ^[1]	Gain-of-Function	KITExon11Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 550–580 of KIT^[2]
MET exon 14 skipping ^[1]	Gain-of-Function	METExon14Skipping	- Positive mutation call - Location is splice site in MET exon 14, is intronic >= 4bp deletion in 30 nucleotides preceding MET exon 14, or variant is in MET Exon 14 Skipping confirmed list
PDGFRA exon 18 deletion ^[1]	Gain-of-Function	PDGFRAExon18Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 841–847 of PDGFRA^[2]

^[1] For more information, see “Ion Torrent™ class-based variants” on page 544.

^[2] In some cases the assay design may not cover the entire codon range.



OncoPrint™ Myeloid Assay GX v2

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Gene fusion	Gain-of-Function	Fusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Is a targeted fusion isoform
Nontargeted fusion	Gain-of-Function	NonTargetedFusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Non-Targeted field is present - NT_FUSION_IN_FRAME is not FALSE - NT_FUSION_DRIVER_INVOLVED is not FALSE - NT_FUSION_SECONDARY is not TRUE
RNA exon variant	Gain-of-Function	RNAExonVariant	- SVTYPE = "RNAExonVariant" or "Fusion" - FILTER = "PASS" - Is a targeted RNA exon variant
Loss-of-function truncating de novo mutation	Loss-of-Function	Truncating	- Positive mutation call - Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense - Occurs in a loss-of-function gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Functional impact is missense - Transcript and codon position occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Intronic hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and ID occur in predefined intronic hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a gain-of-function gene

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and coding syntax occur in MNV hotspot list
CEBPA-bZIP ^[1]	Loss-of-Function	CEBPA-bZIP	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, or nonframeshift block substitution - INDEL impacts codons 282–345 of CEBPA^[2]
CEBPA-N-terminal ^[1]	Loss-of-Function	CEBPA-N-terminal	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, nonframeshift block substitution, frameshift deletion, frameshift insertion, or frameshift block substitution - INDEL impacts codons 1–120 of CEBPA^[2]
JAK2 exon 12 INDEL ^[1]	Gain-of-Function	JAK2Exon12Indel	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, nonframeshift block substitution - INDEL impacts codons 536–547 of JAK2^[2]
KIT exon 11 deletion ^[1]	Gain-of-Function	KITExon11Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 550–580 of KIT^[2] - Alternatively, mutation occurs in splice site flanking the 5' end of exon 11
KIT exon 11 insertion ^[1]	Gain-of-Function	KITExon11Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 550–580 of KIT^[2]

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
FLT3 internal tandem duplication	Gain-of-Function	FLT3ITD	- Positive mutation call - Functional impact is a non-frameshift insertion in exon 14/15 of FLT3 or SVTYPE = "FLT3ITD"

[1] For more information, see "Ion Torrent™ class-based variants" on page 544.

[2] In some cases the assay design may not cover the entire codon range.

OncoPrint™ Precision Assay GX

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number amplification	Gain-of-Function	Amplification	- SVTYPE = "CNV" - FILTER = "GAIN" - Occurs in a designated copy-gain gene
Copy number deletion	Loss-of-Function	Deletion	- SVTYPE = "CNV" - FILTER = "LOSS" - Occurs in a designated copy-loss gene
Gene fusion	Gain-of-Function	Fusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Is a targeted fusion isoform
RNA exon variant	Gain-of-Function	RNAExonVariant	- SVTYPE = "RNAExonVariant" or "Fusion" - FILTER = "PASS" - Is a targeted RNA exon variant
Expression imbalance	Gain-of-Function	ExpressionImbalance	- SVTYPE = "RNAExonTiles" - FILTER = "PASS" - Record meets Targeted Isoforms Detected Requirement
Loss-of-function truncating de novo mutation	Loss-of-Function	Truncating	- Positive mutation call - Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense - Occurs in a loss-of-function gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Functional impact is missense - Transcript and codon position occur in predefined missense hotspot list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript, location, and exon occur in predefined splice site hotspot list
Intronic hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and ID occur in predefined intronic hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	- Positive mutation call - Transcript and coding syntax occur in MNV hotspot list
EGFR exon 19 deletion ^[1]	Gain-of-Function	EGFRExon19Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 744–761 of EGFR^[2]
EGFR exon 20 insertion ^[1]	Gain-of-Function	EGFRExon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 762–775 of EGFR or variant is in EGFR exon 20 insertion confirmed list^[2]
ERBB2 exon 20 insertion ^[1]	Gain-of-Function	ERBB2Exon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 770–780 of ERBB2^[2]

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
KIT exon 11 deletion ^[1]	Gain-of-Function	KITExon11Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 550–580 of KIT^[2] - Alternatively, mutation occurs in splice site flanking the 5' end of exon 11
KIT exon 11 insertion ^[1]	Gain-of-Function	KITExon11Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, nonframeshift block substitution - Insertion impacts codons 550–580 of KIT^[2]
MET exon 14 skipping ^[1]	Gain-of-Function	METExon14Skipping	- Positive mutation call - Location is splice site in MET exon 14, is intronic >= 4bp deletion in 30 nucleotides preceding MET exon 14, or variant is in MET Exon 14 Skipping confirmed list
PDGFRA exon 18 deletion ^[1]	Gain-of-Function	PDGFRAExon18Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift block substitution - Deletion impacts codons 841–847 of PDGFRA^[2]

^[1] For more information, see “Ion Torrent™ class-based variants” on page 544.

^[2] In some cases, the assay design may not cover the entire codon range.

For more information, see the *Genexus™ Software 7.0 Help* or contact support.

Custom panels

The OncoPrint™ Variant Annotator software module applies universal annotation rules for user-defined assays that include custom Ion AmpliSeq™ and Ion AmpliSeq™ HD panels from [Ampliseq.com](https://www.ampliseq.com). To apply the rules, the **Apply OncoPrint Variant Annotations** checkbox must be selected when the assay is created. For more information, see “Panel step assay options” on page 240.

IMPORTANT! Because these annotations are generic, the annotations can be different from those generated with a system-installed or fixed panel.

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number amplification	Gain-of-Function	Amplification	- SVTYPE = "CNV" - FILTER = "GAIN" - Occurs in a designated copy-gain gene
Copy number deletion	Loss-of-Function	Deletion	- SVTYPE = "CNV" - FILTER = "LOSS" - Occurs in a designated copy-loss gene
Copy number exon deletion	Loss-of-Function	ExonDeletion	- SVTYPE = "CNV" - FILTER = "LOSS" - SUBTYPE = "BigDel"
Copy number exon duplication	Loss-of-Function	ExonDuplication	- SVTYPE = "CNV" - FILTER = "GAIN" - SUBTYPE = "BigDup"
Gene fusion	Gain-of-Function	Fusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Is a targeted fusion isoform
Non-targeted fusion	Gain-of-Function	NonTargetedFusion	- SVTYPE = "Fusion" - FILTER = "PASS" - Non-Targeted field is present - NT_FUSION_IN_FRAME is not FALSE - NT_FUSION_DRIVER_INVOLVED is not FALSE - NT_FUSION_SECONDARY is not TRUE
RNA exon variant	Gain-of-Function	RNAExonVariant	- SVTYPE = "RNAExonVariant" or "Fusion" - FILTER = "PASS" - Is a targeted RNA exon variant

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Expression imbalance	Gain-of-Function	ExpressionImbalance	- SVTYPE = "RNAExonTiles" - FILTER = "PASS" - Record meets Targeted Isoforms Detected Requirement
Long deletion	Loss-of-Function	Truncating	- Positive mutation call - SVTYPE = "LongDel"
Loss-of-function truncating de novo mutation	Loss-of-Function Unclassified	Truncating	- Positive mutation call - Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense - Occurs in a loss-of-function or unclassified gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Functional impact is missense - Transcript and codon position occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Transcript, location, and exon occur in predefined splice site hotspot list
Intronic hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Transcript and ID occur in predefined intronic hotspot list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Synonymous hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Functional impact is synonymous - Transcript and coding syntax occur in predefined synonymous hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	- Positive mutation call - Function, transcript, and coding syntax occur in predefined truncating hotspot list - Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function Unclassified	Hotspot	- Positive mutation call - Transcript and coding syntax occur in MNV hotspot list
CEBPA-bZIP ^[1]	Loss-of-Function	CEBPA-bZIP	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, or nonframeshift block substitution - INDEL impacts codons 282–345 of CEBPA^[2]
CEBPA-N-terminal ^[1]	Loss-of-Function	CEBPA-N-terminal	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, nonframeshift block substitution, frameshift deletion, frameshift insertion, or frameshift block substitution - INDEL impacts codons 1–120 CEBPA^[2]
EGFR exon 19 deletion ^[1]	Gain-of-Function	EGFRExon19Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, or nonframeshift block substitution - Deletion impacts codons 744–761 of EGFR^[2]



(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
EGFR exon 20 insertion ^[1]	Gain-of-Function	EGFRExon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, or nonframeshift block substitution - Insertion impacts codons 762–775 of EGFR or variant is in EGFR exon 20 insertion confirmed list^[2]
ERBB2 exon 20 insertion ^[1]	Gain-of-Function	ERBB2Exon20Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, or nonframeshift block substitution - Insertion impacts codons 770–783 of ERBB2^[2]
JAK2 exon 12 INDEL ^[1]	Loss-of-Function	JAK2Exon12Indel	- Positive mutation call - Functional impact is nonframeshift deletion, nonframeshift insertion, or nonframeshift block substitution - INDEL impacts codons 536–547 of JAK2^[2]
KIT exon 11 deletion ^[1]	Gain-of-Function	KITExon11Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, or nonframeshift block substitution - Deletion impacts codons 550–580 of KIT^[2] - Alternatively, mutation occurs in splice site flanking the 5' end of exon 11
KIT exon 11 insertion ^[1]	Gain-of-Function	KITExon11Insertion	- Positive mutation call - Functional impact is nonframeshift insertion, or nonframeshift block substitution - Insertion impacts codons 550–580 of KIT^[2]
MET exon 14 skipping ^[1]	Gain-of-Function	METExon14Skipping	- Positive mutation call - Location is splice site in MET exon 14, is intronic >= 4bp deletion in 30 nucleotides preceding MET exon 14, or variant is in MET Exon 14 Skipping confirmed list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
PDGFRA exon 18 deletion ^[1]	Gain-of-Function	PDGFRAExon18Deletion	- Positive mutation call - Functional impact is nonframeshift deletion, or nonframeshift block substitution - Deletion impacts codons 841–847 of PDGFRA^[2]
FLT3 internal tandem duplication	Gain-of-Function	FLT3ITD	- Positive mutation call - Functional impact is a non-frameshift insertion in exon 14/15 of FLT3 or SVTYPE = "FLT3ITD"

^[1] For more information, see "Ion Torrent™ class-based variants" on page 544.

^[2] In some cases the assay design may not cover the entire codon range.

Ion Torrent™ class-based variants

Ion Torrent™ Class-Based Variants are a set of logic-based rules that Genexus™ Software uses to computationally identify and annotate novel mutations in important genomic variant classes. The annotation logic is based on scientific literature evidence. Details about the logic follow.



CEBPA-N-terminal and CEBPA-bZIP mutations

- **Background**—The CEBPA gene encodes the enhancer binding protein alpha, a member of the basic region leucine zipper family of transcription factors that recognizes the CCAAT promoter [PMID:19706798]. CEBPA is an intron-less gene located on chromosome 19q13.1 [PMID:24056881] and it produces two protein isoforms—p42 and a shorter p30 that lacks the N-terminal transcriptional activation domain (TAD) of p42 [PMID: 17671234]. Both isoforms have the bZIP domain, which is involved in hetero or homo-dimerization with other CEBP family members and is required for DNA binding [PMID: 19706798]. Mutations in CEBPA are reported in 6–18% of all acute myeloid leukemia (AML) cases [PMID: 24071849, PMID: 18752591, PMID: 29861846, PMID: 20038735]. In AML, CEBPA mutations are observed to occur as either monoallelic (single mutant) or biallelic (double mutant) [PMID: 20439648, PMID: 20038735, PMID: 28179278]. Biallelic CEBPA mutations are heterozygous and observed in 52–78% of CEBPA mutated AML patients [PMID:20038735, PMID: 34448807, PMID: 31477806, PMID: 28250006]. Within these individuals, 85–100% of biallelic combinations involve an N-terminal frameshift on one allele and a C-terminal in-frame mutation on the other allele (referred to as an N/C mutant) [PMID: 17934489, PMID: 28179278, PMID:20038735, PMID: 34448807, PMID: 31477806, PMID: 28250006]. C-terminal in-frame mutations disrupt the bZIP domain which interferes with DNA binding and hetero/homo-dimerization. Monoallelic mutations in the bZIP region of CEBPA and biallelic CEBPA mutations are recognized as diagnostic entities for AML by the World Health Organization (WHO) [PMID: 35732831]. In-frame bZIP region mutations in CEBPA, whether they occur as monoallelic or biallelic mutations, are associated with a favorable prognosis in AML [PMID: 35797463].
- **CBV Criteria**—The boundaries defined to capture the CEBPA N-terminal mutations and the CEBPA-bZIP mutations as class-based variants are summarized in the following two rules.
 - a. CEBPA N-terminal mutation boundaries include frameshift mutations and in-frame INDELS within codon 1 and codon 120
 - b. CEBPA-bZIP mutation boundaries include in-frame INDELS that start at codon 282 and end at codon 345.

EGFR exon 19 deletion

- **Background**—Epidermal growth factor receptor (EGFR) is a transmembrane receptor protein made up of 1,210 amino acids and located on chromosome 7p11.2. EGFR has 31 exons—exons 2-4 and 8-12 encode the ligand binding domains, exons 5-7 and 13-16 encode the cysteine rich domain, exons 18–24 encode the tyrosine kinase domain (TKD), and exons 25-28 encode the autophosphorylation region [PMID: 17318210]. Mutations in EGFR often target the TKD which make up the ATP-binding pocket of the enzyme [PMID: 17318210]. Exon 19 deletions are INDELS that range between 15-18 bp in length and usually occur within codons 746-756 [PMID: 23768755, PMID: 22190593, PMID: 26933124, PMID: 24163741]. In non-small cell lung cancer (NSCLC), EGFR exon 19 deletions, located within codons 746-750, represent 45-50% of somatic mutations [PMID: 26933124, PMID: 24163741]. Exon 19 deletions result in constitutive activation of the receptor tyrosine kinase and hyperactivation of downstream signaling pathways [PMID: 29455648].
- **CBV Criteria**—The boundaries defined to capture EGFR exon 19 deletions as a class-based variant start at codon 744 and end at codon 761, which encompass common and rare INDELS [PMID: 30473385].

EGFR exon 20 insertion

- **Background**—Epidermal growth factor receptor (EGFR) is a transmembrane receptor protein made up of 1,210 amino acids located on chromosome 7p11.2. EGFR has 31 exons—exons 2-4 and 8-12 encode the ligand binding domains, exons 5-7 and 13-16 encode the cysteine rich domain, exons 18-24 encode the tyrosine kinase domain (TKD), and exons 25-28 encode the autophosphorylation region [PMID: 17318210]. Mutations in EGFR often target the TKD which make up the ATP-binding pocket of the enzyme [PMID: 17318210]. In-frame insertions within exon 20 of EGFR are the third most common type of mutation found in NSCLC, representing 4-12% of all EGFR mutations in NSCLC [PMID: 31208370]. EGFR exon 20 encompasses codons 762 to 823. Exon 20 insertions commonly involve codons 762 to 774 wherein codons 762-766 make up the c-helix and codons 767-774 make up the activation loop [PMID: 31208370, PMID: 30854234]. EGFR exon 20 insertions do not alter the receptor binding affinity; however, it is suggested that the location of exon 20 insertions stabilizes its active conformation [PMID: 30854234, PMID: 27843613].
- **CBV Criteria**—The boundaries defined to capture EGFR exon 20 insertions as a class-based variant start at codon 762 and end at codon 775. Additionally, known EGFR exon 20 insertions p.A763_Y764insFQEA and p.A763_Y764insLQEA, represented as c.2284-6_2284-5insTCCAGGAAGCCT and c.2284-9_2284-8insCCCTCCAGGAAG due to left alignment into the intronic region, are included in this rule.

The **Splice Site Size** annotation parameter value can be modified from its default value of 2 to a value of 6, so that the variant is annotated as expected. The current setting affects the annotation for EGFR p.A763_Y764insFQEA.

IMPORTANT! Changes to the Splice Site Size parameter will also impact the OncoPrint Variant Annotator annotations for all variants that are close to the splice site.

ERBB2 exon 20 insertion

- **Background**—Erb-b2 receptor tyrosine kinase 2 (ERBB2) is a transmembrane glycoprotein located on chromosome 17q12 [PMID: 25276427]. ERBB2 (also known as HER2) has 27 exons—exons 2-4 and 9-12 encode the extracellular receptor L domains, exons 5-8 encode the furin like domain, exons 13-16 encode the growth factor receptor domain IV, exon 17 encodes the transmembrane domain, and exons 18-24 encode the tyrosine kinase domain (TKD) [PMID: 22761469, PMID: 29420467]. In lung cancer, the most recurrent ERBB2 activating mutations include in-frame exon 20 insertions [PMID: 30425522]. ERBB2 exon 20 involves codons 770 to 831 and majority of exon 20 insertions occur between codons 775 and 781 within the kinase domain [PMID: 29686424, PMID:22761469]. Insertions at the C-terminal end of ERBB2 exon 20 induce a change in conformation of the α -C helix leading to a constitutively active formation which affects the drug-binding pocket [PMID: 29686424].
- **CBV Criteria**— The boundaries defined to capture ERBB2 exon 20 insertions as a class-based variant start at codon 770 and end at codon 783.

JAK2 exon 12 INDEL

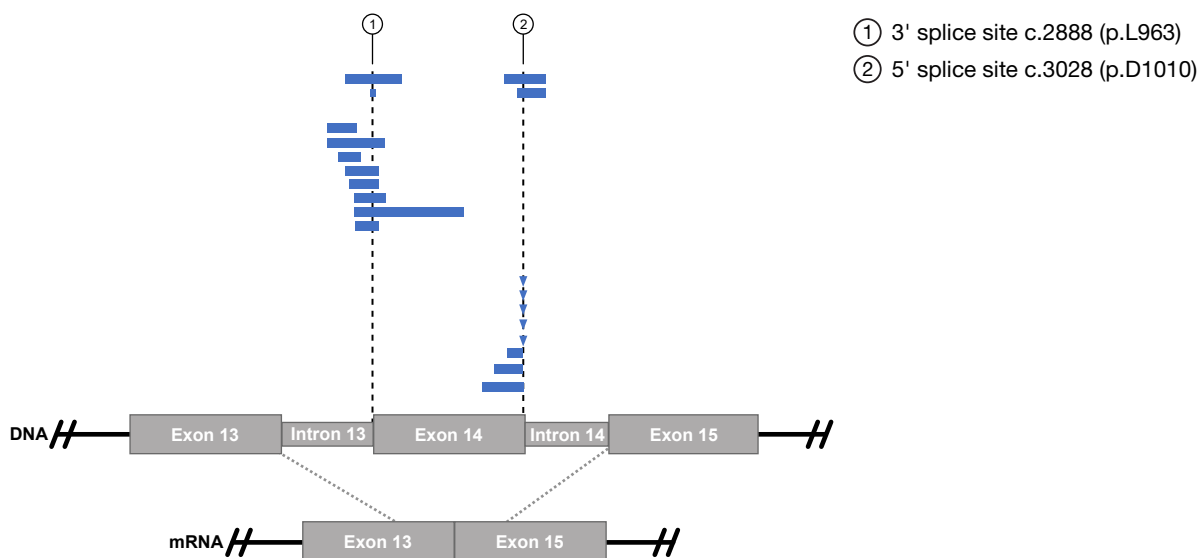
- **Background**—Janus kinase 2 (JAK2) is a non-receptor membrane associated protein tyrosine kinase (PTK) made up of 1,132 amino acids and located on chromosome 9p24. JAK2 belongs to the Janus kinase (JAK) family and has 25 exons—exons 3–7 encode the FERM domain, exons 9–11 encode the SH2-pseudokinase domain, exons 12–19 encode the kinase-like or pseudokinase domain (JH2), and exons 19–25 encode the C-terminal tyrosine kinase domain (JH1) [PMID: 23592968; PMID: 17639043; PMID: 29379470]. In addition, codons 523–544 in exon 12 encode the SH2-pseudokinase linker region and codons 817–839 in exon 19 encode the linker between the JH1 and JH2 protein kinase regions [PMID: 23592968, PMID: 17639043]. Mutations in JAK2 often target exon 12 including the region adjacent to the start of the pseudo-kinase domain [PMID: 21674578]. JAK2 exon 12 mutations (missense and in-frame deletions) are prevalent in myeloproliferative neoplasms including polycythemia vera [PMID: 21674578].
- **CBV Criteria**—The boundaries defined to capture JAK2 exon 12 in-frame insertions and deletions (INDELS) as a class-based variant start at codon 536 and end at codon 547. These boundaries capture the most common JAK2 exon 12 INDELS, including N542_E543del, E543_D544del, F537_K539delinsL and R541_E543delinsK mutations.

KIT exon 11 insertion and KIT exon 11 deletion

- **Background**—KIT, also known as CD117, is a proto-oncogene receptor tyrosine kinase made up of 976 amino acids located on chromosome 4q12 [PMID: 16689459]. KIT is a member of the PDGFR type III receptor tyrosine kinase family [PMID: 26579483; PMID: 26309392]. KIT has 21 exons—exons 1–9 encode the extracellular ligand-binding region containing five Ig-like domains, exon 10 encodes the transmembrane (TM) domain, and exon 11 encodes a cytoplasmic juxtamembrane region. The intracellular region of KIT is encoded by exons 12–21, wherein, exons 12–14 encode the tyrosine kinase domain 1, exons 15–16 encode the kinase insert (KI) region, and exons 17–19 encode the tyrosine kinase domain 2 [PMID: 24449920, PMID: 27536065, PMID: 31378162, PMID: 15342366, PMID: 17555444]. Recurrent somatic KIT alterations are observed in both solid and hematological cancers and include activating mutations such as single nucleotide variants (SNVs), small duplications, and complex in-frame insertions or deletions (INDELS). Mutations in KIT exons 8, 9, 11, and 17 disrupt the auto-inhibitory mechanisms and lead to constitutive activity [PMID: 27536065]. While mutations in exons 8 and 17 are common in myeloid cancers, 60–70% of somatic mutations (including SNVs and INDELS) in gastrointestinal stromal tumor (GIST) occur in exon 11, which encodes the juxtamembrane domain [PMID: 27536065, PMID: 22588877, PMID: 23678293; NCCN-GIST].
- **CBV Criteria**—The boundaries defined to capture KIT exon 11 insertions and deletions as a class-based variant start at codon 550 and end at codon 580.

MET exon 14 skipping

- Background**—The MET proto-oncogene is a receptor tyrosine kinase made up of 1,390 amino acids and is located on chromosome 7q31 [PMID: 15735036]. MET has 21 exons containing three main structural domains—an extracellular sema domain in exon 2, a juxtamembrane domain in exon 14, and a tyrosine kinase domain in exons 15-21 [PMID: 28376232, PMID: 9380410]. Splice site mutations flanking exon 14 are observed in 3–4% of non-small cell lung cancer (NSCLC). These mutations include canonical splice site mutations affecting exon 14 and deletions that extend into the splicing motifs within intron 13 [PMID: 25971938, PMID: 27343443]. Such mutations disrupt splicing leading to the formation of an alternative transcript that joins exon 13 directly to exon 15 and skips exon 14 entirely. The MET exon 14 skipping transcript lacks the juxtamembrane domain that contains the recognition motif for ubiquitin-dependent proteolysis and thus leads to a marked increase in steady-state level of the MET protein [PMID: 28164087].
- CBV Criteria**—The boundaries defined to capture MET exon 14 skipping as a class-based variant can be summarized into four individual rules.
 - Has transcript = NM_000245.4, exon = 14, and location equal to splice site_3 (at c.2888) or splice site_5 (at c.3028),
 - Is an intronic deletion ≥ 4 bp impacting 30 nucleotides preceding exon 14
 - Is a missense variant at c.3028 [PMID: 27343443, PMID: 26729443, PMID: 25971938, PMID: 25898962, PMID: 25898965, PMID: 31472177],
 - Is one of the following variants with confirmed skipping defined as c.3028delG [PMID: 25971938, PMID: 27343443], c.3012_3027delAGCTACTTTTCCAGAA, c.3021_3028delTCCAGAAG [PMID: 31472177], or c.3026_3027delAA [PMID: 26729443].





PDGFRA exon 18 deletion

- **Background**—Platelet derived growth factor receptor alpha (PDGFRA) is a cell surface tyrosine kinase receptor made up of 1089 amino acids and located on chromosome 4q12. PDGFRA contains 23 exons –exons 1-2 encode the signaling peptide, exons 3-10 encode the extracellular domain, exon 11 encodes the transmembrane domain, exon 12 encodes the juxtamembrane domain, and exons 13-23 encode the kinase domain [PMID: 33449152]. Mutations in PDGFRA often target the activation loop located within exon 18 and, to a lesser extent, target the juxtamembrane domain and the N-lobe of the kinase domain found within exons 12 and 14 [PMID: 30778083, PMID: 33449152, PMID: 15146165]. Activating PDGFRA exon 18 mutations such as V561D, N659K, D842V, and in-frame deletions are observed in 30-40% of KIT-negative gastrointestinal stromal tumors (GISTs) [PMID: 17193822, PMID: 15928335, PMID: 12522257, PMID: 23970477]. Specifically, PDGFRA in-frame deletions in codons 841-847 make up 23% of all exon 18 mutations [PMID: 15146165].
- **CBV Criteria**—The boundaries defined to capture PDGFRA exon 18 deletions as a class-based variant start at codon 841 and end at codon 847.

Variant identifiers for OncoPrint™ panels

Hotspots BED files

The BED file specification (<http://genome.ucsc.edu/FAQ/FAQformat.html#format1.7>) indicates that the fourth column is the name of the BED line. The values in this column are used to label the variant region in the UCSC genome browser or the Integrative Genomics Viewer (IGV). The labels are used in Genexus™ Software to populate the ID field in the output VCF files and the Variant ID columns.

OncoPrint™ panel hotspots files include genomic representations that correspond to somatic variants that have often been observed in cancer samples and therefore are likely to be relevant to the cancer phenotype. These files also include variants that are observed less often, or are implicated in literature reports as functionally relevant, such as activating or inactivating variants.

When possible, variants in the hotspots files are assigned an identifier consistent with a publicly available data source, preferentially COSMIC (<https://cancer.sanger.ac.uk/cosmic>) but also including dbSNP (<https://www.ncbi.nlm.nih.gov/snp/>) and ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/>). If a variant cannot be found in one of these databases, the variant can receive an arbitrarily assigned identifier to help with variant calling interpretation and troubleshooting. Such identifiers, for example, BT144, OM3324, OMINDEL700, MAN103, are designed to be consistent across OncoPrint™ panels. When hotspots files are updated, Thermo Fisher Scientific reviews the COSMIC database to determine whether variants with such identifiers have been assigned COSMIC IDs and sometimes replaces them with the more meaningful COSMIC IDs.

COSMIC ID changes due to database updates

The COSMIC database updates approximately twice a year, and these updates involve the addition of newly curated variants, removal of a smaller number of variants, and changes in genomic representations to a very small number of variants. Therefore, there is a chance that the variant identifiers in the hotspots files are not synchronized with – or are no longer found on – the COSMIC website. Additionally, before COSMIC version 90, the same normalized genomic variant (see <https://pubmed.ncbi.nlm.nih.gov/25701572/>) can have had multiple redundant legacy COSMIC variant identifiers (COSM); since COSMIC version 90, these identifiers have been replaced with one consistent COSV identifier, to which multiple legacy COSMIC IDs can map.

Fusions files

A comprehensive, universally recognized database of oncogenic gene fusion breakpoints does not exist. Therefore, gene fusion isoform identifiers are generated by concatenating the two gene symbols with a hyphen, and then combining the first letter of the 5' partner with the last retained 5' exon along with the first letter of the 3' partner with the first retained 3' exon. For example, EML4-ALK.E6A17 involves a fusion between the sixth exon of EML4 with the 17th exon of ALK. Fusion isoforms involving junctions between incomplete exons, or involving intronic insertions can include “ins” or “del” modifiers followed by the number of nucleotides removed or added. Additionally, assay names can include additional identifiers such as COSMIC COSF ids or GenBank accession numbers.

System-installed barcode sets

The Genexus™ Software includes system-installed barcode sets as presets to use in assays. System-installed barcode sets are locked and cannot be changed.

System-installed barcode set	Description
Ion AmpliSeq™ HD Dual Barcode Kit 1–24	A set of 24 unique dual-barcode primers designed and validated for optimal performance with Ion GeneStudio™ S5 Series sequencers. For use with Ion AmpliSeq™ HD assays.
IonCode™ Barcodes 1-32	A unique set of 32 barcode adapters designed for optimal performance with Ion GeneStudio™ S5, Ion PGM™, and Ion Proton™ sequencers.
IonCode™ Barcode Adapters 1–384 Kit	A unique set of 384 barcode adapters designed for optimal performance with Ion GeneStudio™ S5, Ion PGM™, and Ion Proton™ sequencers.
Ion Select BC Set-1	A unique set of 16 barcode adapters supplied with the Ion PGM™ Select Library Kit.
Ion SingleSeq™ Barcode	A unique set of barcode adapters optimized for use with Ion ReproSeq™ libraries.
Ion Torrent™ Dual Barcode Kit 1–96	A set of 96 unique dual-matched barcode adapters designed to enable optimal performance of amplicon libraries and Ion semiconductor sequencers. For use with Ion AmpliSeq™ HD and OncoPrint™ assays.



(continued)

System-installed barcode set	Description
Ion Torrent™ Genexus™ Barcodes 1–32 HD	A unique set of 32 barcode adapters designed and validated for optimal performance with the Genexus™ Integrated Sequencer. For use with Ion AmpliSeq™ HD assays.
Ion Torrent™ Genexus™ Barcodes 1–96 AS	A unique set of barcode adapters designed and validated for optimal performance with the Genexus™ Integrated Sequencer.
Ion Torrent™ Genexus™ Barcodes 1–96 AS (48 Barcodes Plate)	A unique set of barcode adapters designed and validated for optimal performance with the Genexus™ Integrated Sequencer.
Ion Xpress™ Barcode Adapters 1–96 Kit	A unique set of 96 barcode adapters for use with Ion AmpliSeq™ libraries and Ion Xpress™ Plus fragment libraries.

System-installed filter chains

Genexus™ Software includes system-installed filter chains to use as presets in assays to filter in or out variants of interest in run results. The filter chains can also be used to dynamically change the view of the variants as you view results.

The available filter chain version depends on the assay parameter set that is installed in the software. We recommend that you use the filter chain version that is compatible with your assay. Some system-installed assays specify the compatible filter chain by default. Some filter chains are installed with an assay definition file.

System-installed filter chains are locked and cannot be changed. You cannot edit system-installed filter chains. You can create user-defined filter chains to refine the results in the software.

System-installed filter chain	Available versions	Genome reference	Description
Default Germline Variants view	7.0	hg19	This is the default filter chain in the custom Ion AmpliSeq™ assays that is used to refine sample results for germline CNV workflows. It returns CNVs based on the specific filter conditions and all “Present” SNV/INDEL and fusion variants. This filter chain is based on the hg19 genome reference. To pass this filter, CNVs must meet specific confidence thresholds. You can enable or disable the default filter chain during assay creation and also within the CNV results table.
Default Germline Variants view GRCh38	7.0	GRCh38	This is the default filter chain in the custom Ion AmpliSeq™ assay to that is used refine sample results for germline CNV workflows. It returns CNVs based on the specific filter conditions and all “Present” SNV/INDEL and fusion variants. This filter chain is based on the GRCh38 genome reference. To pass this filter, CNVs must meet specific confidence thresholds. You can enable or disable the default filter chain during assay creation and also within the CNV results table.
Default Somatic Variants view	7.0	hg19	This is the default filter chain in the custom Ion AmpliSeq™ assays that is used to refine sample results for somatic CNV workflows. This filter chain is based on the hg19 genome reference. This filter chain defines criteria for both copy number amplification (gain) and depletion (loss). Only targets that meet these criteria are reported in the results table when the filter chain is applied. You can enable or disable the default filter chain during assay creation and also within the CNV results table.
Default Somatic Variants view GRCh38	7.0	GRCh38	This is the default filter chain in the custom Ion AmpliSeq™ assays that is used to refine copy sample results for somatic CNV workflows. This filter chain is based on the GRCh38 genome reference. This filter chain defines criteria for both copy number amplification (gain) and depletion (loss). Only targets that meet these criteria are reported in the results table when the filter chain is applied. You can enable or disable the default filter chain during assay creation and also within the CNV results table.
OncoPrint™ BRCA	5.16 ^[1]	hg19	This filter chain removes any variants in the sample ID amplicons that are not in the <i>BRCA1</i> and <i>BRCA2</i> genes.

(continued)

System-installed filter chain	Available versions	Genome reference	Description
OncoPrint™ Extended	7.0^[1] 6.8^[1] 5.16 5.14	hg19	This filter chain includes variants that are annotated with the OncoPrint™ Variant Annotator software module in specific OncoPrint™ panels and variants that can be relevant to cancer due to their inclusion in one or more of the following classes. - CNV variants with FILTER value of GAIN or LOSS. - LOH variants - Likely somatic mutations based on dbSNP, 5000Exomes, gnomAD, and UCSC Common SNPs annotation source databases. The minor allele frequencies range lies between 0.0 and 1.0E-6. Mutations must also be nonsynonymous and occur in exonic or splice-site regions. - Variants with ClinVar annotations of pathogenic or likely pathogenic.
OncoPrint™ Variants	7.0 6.8 5.16 5.14^[1]	hg19	This filter chain includes variants that are annotated with the OncoPrint™ Variant Annotator software module in specific OncoPrint™ panels.
TMB GRCh38 (Non-germline Mutations)	7.0 ^[1]	GRCh38	This filter chain returns results for somatic variants based on dbSNP, 5000Exomes, gnomAD, and UCSC Common SNPs annotation source databases. The minor allele frequencies range is between 0.0 and 1.0E-6. This filter chain also filters out variants of homopolymer lengths >4.
TMB (Non-germline Mutations)	7.0^[1] 6.8^[1]	hg19	This filter chain returns results for somatic variants based on dbSNP, 5000Exomes, gnomAD, and UCSC Common SNPs annotation source databases. The minor allele frequencies range is between 0.0 and 1.0E-6. This filter chain also filters out variants of homopolymer lengths >4.
Variant Matrix Summary	6.8 5.16 5.14^[1]	hg19	This filter chain filters in the following variant types in a visualization. - Positive hotspot calls - Positive gene-level CNV GAIN or LOSS calls <i>FLT3-ITD</i> variants - Non-wild-type intragenic structural variations - For TagSeq Breast and TagSeq Pan-Cancer assays: non-hotspot variants in TP53 with QUAL >50 and Allele Frequency >1.0 (Breast) or 0.5 (Pan Cancer) Variant types returned include SNVs/INDELs, CNVs, fusions, and RNAExonVariants.

^[1] Installed with an assay definition file.

System-installed filters

Genexus™ Software includes system-installed filters that you can add to user-defined filter chains. You can include a filter chain in an assay that is used for a sequencing run.

A filter chain is a set of filters that the software uses to narrow the list of variants that are included in results. When you create a filter chain, you can include filters to dynamically change the view of variants in results, or to filter in or filter out variants from results.

The system-installed filters are based on public and proprietary annotation sources and data types that are included in the software. For optimal filtering results, use the same filter as the annotation source used in the analysis.

The system-installed filters are listed in the software based on filter categories.

- Filters for genes and variants
- Filters for population frequencies
- Filters for metrics
- Filters for annotation sources
- Filters for functional scores
- Other filters that do not fit the above categories

These filters are described in this module in alphabetical order.

5000Exomes AfricanAmerican MAF filter

This filter can be added to filter chains in the software to filter in minor allele frequencies (MAF), SNVs, and INDELs for specific population groups. The filter is derived from the 5000Exomes annotation database.

Filter conditions include the ability to set the minor allele frequency range from 0.0 to 0.5.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	• Returns all variants that do not have a 5000Exome annotation. • Returns all CNV and fusions variants.



5000Exomes EuropeanAmerican MAF filter

This filter can be added to filter chains in the software to filter in minor allele frequencies (MAF), SNVs, and INDELs for specific population groups. The filter is derived from the 5000Exomes annotation database.

Filter conditions include the ability to set the minor allele frequency range from 0.0 to 0.5.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	• Returns all variants that do not have a 5000Exome annotation. • Returns all CNV and fusions variants.

5000Exomes Global MAF filter

This filter can be added to filter chains in the software to filter in minor allele frequencies (MAF), SNVs, and INDELs for specific population groups. The filter is derived from the 5000Exomes annotation database.

Filter conditions include the ability to set the minor allele frequency range from 0.0 to 0.5.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	• Returns all variants that do not have a 5000Exome annotation. • Returns all CNV and fusions variants.

Allele Frequency filter

This filter can be added to filter chains in the software to report the frequency of alleles observed in raw data.

Filter conditions include the ability to set allele frequency ranges from 0.0 to 1.0. The option to include or exclude unannotated variants in filtered results is also available.

The filter returns allele frequencies of alleles observed in raw data.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) within the specified threshold.
NOT is selected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) outside of the specified threshold.
Include Unannotated is selected.	Returns all CNV and fusions variants.

Allele Ratio filter

This filter can be added to filter chains in the software to narrow results to nonreference allele frequencies.

Filter conditions include the ability to set allele ratio ranges from 0.0 to 1.0. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

The filter returns all variants that have at least one nonreference allele reported with a frequency in the selected filter range.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all CNV and fusion variants.

Allele Read-Count filter

This filter can be added to filter chains in the software to set the minimum count for genotype alleles.

Filter conditions include the ability to set allele read-count ranges from 0 to 100,000. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

The filter removes variant candidates that do not have the required numbers of supporting reads.

The variants returned by the filter depend on the options that are selected.

The filter returns all variants that are not SNV/INDEL, MNV, REF, NOCALL, FLT3ITD, and ProcControl regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) within the specified threshold.
NOT is selected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) outside of the specified threshold.
Include Unannotated is selected.	Returns all CNV and fusions variants.

Alternate Allele Count filter

This filter can be added to filter chains in the software to set the minimum count for alternate alleles.

Filter conditions include the ability to set alternate allele read-count ranges from 0 to 100,000. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

The filter removes variant candidates that do not have the required numbers of reads.

The variants returned by the filter depend on the options that are selected.

The filter returns all the variants that are not SNV/INDEL, MNV, REF, NOCALL, FLT3ITD, and ProcControl regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) within the specified threshold.
NOT is selected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) outside of the specified threshold.
Include Unannotated is selected.	Returns all CNV and fusions variants.

Clinvar filters

This filter can be added to filter chains in the software to evaluate the impact of variants that match those in the National Center for Biotechnology Information (NCBI) ClinVar database.

In Genexus™ Software, updated versions of the ClinVar database are available. For more information, see “Updated versions of annotations” on page 276.

One or more of the impact values can be included in the filter chain. The complete list can be viewed when the **All** option is selected.

The filter returns all variants with the selected impact values that match those in the NCBI ClinVar database.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a ClinVar annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a ClinVar annotation.
Include Unannotated is selected.	• Returns all CNV and fusions variants • Returns all the variants that do not have a ClinVar annotation.

CNV Confidence Range filter

This filter can be added to filter chains in the software to return copy number variants (CNV) with confidence levels between 10 and 10,000,000 and other variants.

Filter conditions include the ability to set the CNV confidence range from 10 to 1.0E7. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

The filter returns copy number variants with high confidence levels and other variants.

The variants returned by the filter depend on the options that are selected.

The filter returns all variants that are not CNV or the CONFIDENCE field is absent regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all SNV/INDEL and fusion variants.

CNV Confidence Range—CNVs Only filter

This filter can be added to filter chains in the software to return copy number variants (CNV) with confidence levels between 0 and 10,000,000.

Filter conditions include the ability to set the CNV confidence range from 10 to 1.0E7. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

The filter returns only copy number variants with high confidence levels.

The variants returned by the filter depend on the options that are selected.

The filter returns all variants that are not CNV regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all SNV/INDEL and fusion variants.

CNV Somatic Confidence Range filter

This filter can be added to filter chains in the software to make ploidy estimates with lower and upper confidence values on somatic samples. The filter detects extra copies over the expected normal ploidy value (2 for autosomes, 2 for X chromosomes in females, and 1 for X chromosomes in males). The 5% lower confidence bound value is the ploidy estimate where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate where it is 95% certain that the true ploidy is below that value. The lower bound is the most important for gains. The upper bound is most important for losses.

By default, the filter is set to detect gains and losses using the confidence interval values of 5% confidence interval for Minimum Ploidy Gain over the expected value and 95% confidence interval for Minimum Ploidy Loss under the expected value. The default boundary values are set to 0.0.

This filter can be set to return the samples that do not meet both of the thresholds for Minimum Ploidy Gain and Minimum Ploidy Loss. When there are multiple filters in a filter chain, the software setting of (A or B), NOT (A or B) is calculated as (NOT A) and (NOT B). Because of this logic, the setting to return the samples that do not meet both threshold settings is NOT with OR. For example, NOT **Minimum Ploidy Gain (5% CI) over expected OR Minimum Ploidy Loss (95% CI) under expected**.

If you set the **Minimum Ploidy Gain (5% CI) over expected** to **2.0** ($2+2=4$) and the **Minimum Ploidy Loss (95% CI) under expected** to **0.0**, you can expect the following example CNV call CI data:

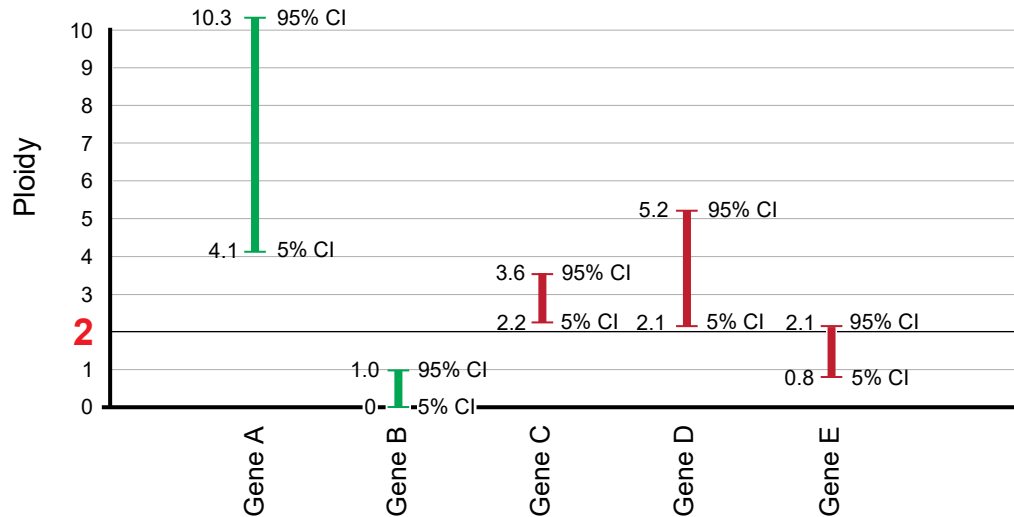
Gene A—A gene with suspected gain of 5% CI = 4.1 and 95% CI = 10.3 filtered in.

Gene B—A gene with suspected loss of 5% CI = 0 and 95% CI = 1.0 filtered in.

Gene C—A gene with suspected gain of 5% CI = 2.2 and 95% CI = 3.6 filtered out (2.2 is <4).

Gene D—A gene with suspected gain of 5% CI = 2.1 and 95% CI = 5.2 filtered out (2.1 is <4).

Gene E—A gene with 5% CI = 0.8 and 95% CI = 2.1 filtered out (0.8 is less than 4 and 2.1 is >2).



The variants returned by the filter depend on the options that are selected.

This filter returns all the variants that are not CNV or the metric (VCN5/VCN95) is unavailable, regardless of the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants greater than or equal to the specified threshold.
NOT is selected.	Returns all variants less than the specified threshold.
Include Unannotated is selected.	Returns all the variants where the metric (VCN5/VCN95) is not available.

CNV Somatic Confidence—CNVs Only filter

This filter can be added to filter chains in the software to make ploidy estimates with lower and upper confidence levels.

Filter conditions include the ability to enable and set the range for Minimum Ploidy Gain (5% CI) over expected, to enable and set the Minimum Ploidy Loss (95% CI) under expected ploidy values. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

The filter returns only somatic copy number variants within the lower and upper limits of the set confidence ranges.

This filter can be set to return the samples that do not meet both of the thresholds for Minimum Ploidy Gain and Minimum Ploidy Loss. When there are multiple filters in a filter chain, the software setting of (A or B), NOT (A or B) is calculated as (NOT A) and (NOT B). Because of this logic, the setting to return the samples that do not meet both threshold settings is NOT with OR. For example, NOT **Minimum Ploidy Gain (5% CI) over expected OR Minimum Ploidy Loss (95% CI) under expected**.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants greater than or equal to the specified threshold.
NOT is selected.	Returns all variants less than the specified threshold.
Include Unannotated is selected.	Returns all variants where the metric (VCN5/VCN95) is not available.

CNV p-value filter

This filter can be added to the filter chain in the software to review the CNV results that meet the specific p-value threshold. You can adjust the threshold.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants (CNV and LOH) below or equal to the specified threshold.
NOT is selected.	Returns all variants (CNV and LOH) above the specified threshold.
Include Unannotated is selected.	Returns all SNV/INDEL and fusion variants.

COSMIC filters

These filters can be added to filter chains in the software to compare variants to the catalog of somatic mutations in tumor tissue as compiled by the COSMIC database.

Filter conditions include the ability to select all COSMIC values and to include or exclude unannotated variants in filtered results.

In addition, one or more specific annotation values can be included in the filter chain. Examples include ER-positive_carcinoma and basal_cell_carcinoma. A partial list can be viewed when the option of **All** is selected.

The filter returns variants that match those in the COSMIC database. In sample results, the CNVs tab shows the **Variant ID** in the **COSMIC** column. The **Variant ID** may not be available when you create a filter chain.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a COSMIC annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.

(continued)

Options	Variants returned
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a COSMIC annotation.
Include Unannotated is selected.	Returns all variants that do not have a COSMIC annotation.

dbSNP filters

These filters can be added to filter chains in the software to compare single nucleotide polymorphism (SNP) variants in samples against the dbSNP database. The filters detect SNP variants that match those in the dbSNP database, which can be included or excluded from filtered results.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants that have a dbSNP annotation.
NOT is selected.	Returns all variants that do not have a dbSNP annotation, including CNV and fusions.
Include Unannotated is selected.	Returns all CNV and fusion variants.

Default DNA and Fusions View filter

This filter can be added to filter chains in the software to detect all fusion variants, expression control markers, assay 5'/3' markers, and all DNA variants that the predefined Default DNA and Fusions View filter chain detects.

Filter conditions include the ability to select specific annotations (**Present** or **Absent**), to include all default DNA and fusions view variants (in), or to exclude all Default DNA and Fusions view variants (NOT in) in filtered analysis results. In addition, unannotated variants can be included or excluded in filtered analysis results.

The filter detects the fusion variants, expression control markers, assay 5'/3' markers, and all DNA variants, which can be included or excluded in filtered analysis results.

The variants returned by the filter depend on the options that are selected.



This filter returns all SNV/INDEL, CNV, EXPR_CONTROL, ASSAYS_5P_3P, RNA_HOTSPOT, and GENE_EXPRESSION variants regardless of the selected options.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all fusion variants.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that are not fusion.
Include Unannotated is selected.	Returns all variants that are not fusion.

Default Fusions View filter

This filter can be added to filter chains in the software to detect all fusion variants, expression control markers, and assay 5'/3' markers that the predefined Default Fusions View filter chain detects.

Filter conditions include the ability to select specific annotations (**Present** or **Absent**), to include all Default Fusions View variants (in), or to exclude all Default Fusions View variants (NOT in) in filter analysis results. In addition, unannotated variants can be included or excluded in filtered analysis results.

The filter detects fusion variants, expression control markers, and assays 5'/3' markers, which can be included or excluded in filtered analysis results.

The variants returned by the filter depend on the options that are selected.

The filter returns all the SNV, INDEL, CNV, EXPR_CONTROL, ASSAYS_5P_3P, RNA_HOTSPOT, and GENE_EXPRESSION variants regardless of the selected options.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all fusion variants.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.

(continued)

Options	Variants returned
NOT is selected and In is selected.	Returns all variants that are not fusion.
Include Unannotated is selected.	Returns all variants that are not fusion.

DGV filters

These filters can be added to filter chains in the software to detect human genomic structural variants that match those in the Database of Genomic Variants (DGV). The filters detect human genomic structural variants as defined by DGV, which can be included or excluded from filtered results.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all CNV variants that have a DGV annotation.
NOT is selected.	Returns all variants that do not have a DGV annotation, including SNV/INDELs and fusions.
Include Unannotated is selected.	Returns all SNV/INDEL and fusion variants.

DRA filter

This filter can be added to filter chains in the software to annotate disease research categories as defined by the Disease Research Area (DRA) annotation source database.

Filter conditions include the ability to search for all (in) or none (NOT in) and to include or exclude unannotated variants in filtered results.

In addition, one or more specific annotation values can be included in the filter chain. Examples include acquired immunodeficiency syndrome and adenoma.

This filter annotates disease research areas that match the DRA database, and can be included or excluded.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a DRA annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.



(continued)

Options	Variants returned
NOT is selected and In is selected.	Returns all variants that do not have a DRA annotation.
Include Unannotated is selected.	Returns all variants that do not have a DRA annotation.

DrugBank filters

These filters can be added to filter chains in the software to detect variants that are correlated with drugs and drug targets listed in the DrugBank database.

Filter conditions include the ability to select specific values, to include all DrugBank values (in), or to exclude all DrugBank values (NOT in). In addition, unannotated variants can be included or excluded in filtered results.

This filter detects variants that are correlated with drugs and drug targets in DrugBank, which can be included in or excluded from filtered results.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a DrugBank annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a DrugBank annotation.
Include Unannotated is selected.	Returns all variants that do not have a DrugBank annotation.

ExAC filters

These filters can be added to filter chains in the software to detect rare gene variants for specific population groups. The filter detects rare gene variant results that align with the Exome Aggregation Consortium (ExAC) database, which can be included or excluded from filtered results. The filters are derived from the ExAC database.

Filter conditions include the ability to set a range from 0.0 to 1.0.

The variants returned by the filter depend on the options that are selected.

The filter returns all variants with hit level allele regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	• Returns all variants without hit level allele for ExAC annotations. • Returns all variants without ExAC annotations. • Returns all CNV and fusions variants.

Expected Normal Ploidy Buffer filter

This filter can be added to filter chains in the software to detect mosaic chromosomal aneuploidies in research samples. You can set a buffer for normal ploidy values (2 for autosomes, 2 for X chromosomes in females, and 1 for X and Y chromosomes in males) to identify the most likely aneuploidy samples. Ploidy calls do not have to be integers because mosaicism detection allows decimal-level copy number gain or loss calls.

By default, the filter is set to filter out all copy number variant segments of gain or loss within 0.2 ploidy value of expected normal. Gains must be above ploidy 2.2 and below ploidy 1.8 to remain filtered in on autosome and female X, and ploidy 1.2 and ploidy 0.8 on male Y. Filter conditions include the ability to change the amount of expected normal ploidy buffer by adjusting the *from* and *to* values. The expected normal ploidy buffer overrides any confidence filtering that may be in effect within the buffer zone.

This filter can be set to return the samples that do not meet both of the thresholds for expected normal ploidy buffer and confidence. When there are multiple filters in a filter chain, the software setting of (A or B), NOT (A or B) is calculated as (NOT A) and (NOT B). Because of this logic, the setting to return the samples that do not meet both the expected normal ploidy buffer and confidence thresholds is NOT with OR. For example, NOT expected normal ploidy buffer OR confidence.

If default settings are applied, ploidy values of 1.8 to 2.2 are filtered out as normal for autosomes and X chromosomes in females, and 0.8 to 1.2 are filtered out as normal for X and Y chromosomes in males. Values outside this range are detected as chromosomal ploidy samples.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Applies only to aneuploidy run results.

FATHMM Scores filters

These filters can be added to filter chains in the software to calculate functional analysis through Hidden Markov Models (FATHMM) scores for coding variants, nonsynonymous single-nucleotide variants (nsSNVs), and noncoding variants.

Filter conditions include the ability to set a range from 0.0 to 1.0 for the score.

The filter includes FATHMM scores for coding variants and non-coding variants.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all CNV and fusions variants.

Filter filter

This filter can be added to filter chains in the software to show or exclude specific annotations.

Filter conditions include the ability to include all (**In**), none (**NOT In**), or specific annotations from the list, and to include or exclude unannotated variants in results when you apply the filter chain.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a FILTER column value in the VCF file.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.

(continued)

Options	Variants returned
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a FILTER column value in the VCF file.
Include Unannotated is selected.	Returns all variants that do not have a FILTER column value in the VCF file.

Filtered Coverage filter

This filter can be added to filter chains in the software to review results that meet a specific coverage threshold. By default, the filter reports all results with a coverage ≥ 100 . You can adjust the minimum threshold. The minimum threshold value that is set is included.

The option to include or exclude unannotated variants in filtered results is also available.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants greater than or equal to the specified threshold.
NOT is selected.	Returns all variants less than the specified threshold.
Include Unannotated is selected.	Returns all SNV/INDEL and fusion variants.

Fold Difference filter

This filter can be added to filter chains in the software to review the CNV results that meet the specific fold difference threshold. You can adjust the threshold.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants below or equal to the specified threshold.
NOT is selected.	Returns all variants above the specified threshold.
Include Unannotated is selected.	Returns all SNV/INDEL and fusions variants.

Functional Scores filter

This filter can be added to filter chains in the software to provide functional scores based on SIFT, PolyPhen, and Grantham scores. SIFT and PolyPhen scores are predictions of the functional effect of a variant on a protein. A Grantham score attempts to predict the distance between two amino acids, in an evolutionary sense (lower is less distance, higher is greater distance).

Filter conditions include the ability to include or exclude SIFT, PolyPhen, and Grantham scores, and to set ranges for those scores. The range boundaries are included. SIFT and PolyPhen ranges can be set from 0.0 to 1.0, and Grantham can be set from 0.0 to 215.0.

The filter returns a functional score based on the three (or fewer) scores.

This filter can be set to return the variants that do not meet all three threshold scores set in the filter. When there are multiple filters in a filter chain, the software setting of (A or B), NOT (A or B) is calculated as (NOT A) and (NOT B). Because of this logic, the setting to return the samples that do not meet all threshold settings is NOT with OR. For example, NOT **SIFT**, **PolyPhen** OR **Grantham**.

The variants returned by the filter depend on the options that are selected.

The filter returns all the variants that are not SNV/INDEL, MNV, REF, NOCALL, FLT3ITD, and ProcControl regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) within the specified threshold including boundaries.
NOT is selected.	Returns all variants (SNV/INDEL, MNV, REF, and NOCALL) outside of the specified threshold.
Include Unannotated is selected.	Returns all CNV and fusion variants.

Fusion Read Counts filter

This filter can be added to filter chains in the software to produce counts for fusion reads.

Filter conditions include the ability to set a minimum count of 0 or higher, to include the boundary value for the count, and to include or exclude unannotated variants in filtered analysis results.

This filter returns results that meet the fusion read minimum count value.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants greater than or equal to the specified threshold.
NOT is selected.	Returns all variants less than the specified threshold.
Include Unannotated is selected.	Returns all SNV/INDEL and CNV variants.

Gene Symbol filter

This filter can be added to filter chains in the software to report on specific genes. This filter returns the gene symbols of genes that are found in your samples.

Filter conditions include the ability to select all (**In**), none (**NOT In**), or specific gene symbols from the list, and to include or exclude unannotated variants in filtered analysis results.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a Gene Model annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a Gene Model annotation.
Include Unannotated is selected.	Returns all variants that do not have a Gene Model annotation.

gnomAD filters

These filters can be added to filter chains in the software to detect rare gene variants for specific population groups. The filters are derived from the genome aggregation database (gnomAD) database. The filter detects rare gene variant results that align with the gnomAD database.

Filter conditions include the ability to set a range from 0.0 to 1.0. The range boundaries are included. Unannotated variants can be included or excluded in filtered results.

The variants returned by the filter depend on the options that are selected.

The filter returns all variants with hit level allele regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all variants without hit level allele for gnomAD annotations. Returns all variants without gnomAD annotation. Returns all CNV and fusions variants.

Genomic Coordinates filter

This filter can be added to filter chains in the software to narrow analysis results to specific genomic regions. The filter can also be used to identify regions that are targeted by one or more amplicons in a user-defined assay.

The filter value is one chromosome region, or multiple regions that are separated by an OR operator or a comma without spaces.

This filter returns variants in samples that are in the range for the genomic coordinates that you set.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns variants that are within the specified regions.
NOT is selected.	Returns variants that are <i>not</i> within the specified regions.

Gene Ontology (GO) filters

These filters can be added to filter chains in the software to detect various gene ontologies (functions of specific genes).

Filter conditions include the ability to search for all (**In**) or none (**NOT In**), and to include or exclude unannotated variants in filtered results.

In addition, one or more specific annotation values can be included in the filter chain.

This filter returns gene ontologies of samples based on the selected options.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for the selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a GO annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values
NOT is selected and Contains is selected.	Return all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a GO annotation.
Include Unannotated is selected.	Returns all variants that do not have a GO annotation.

Homopolymer Length filter

This filter can be added to filter chains in the software to detect INDEL variants of specific homopolymer lengths.

Filter conditions include the ability to set a range from 0 to 1,000 for homopolymer length. The range boundaries are included. The option to include or exclude unannotated variants in filtered results is also available.

This filter returns INDEL variants that meet the specified homopolymer length.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all CNV and fusion variants.

Hotspot filter

This filter can be added to filter chains in the software to show or exclude variants that are hotspots. The filter returns variants that overlap hotspots that are defined in an uploaded hotspots file.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all hotspot variants.
NOT is selected.	Returns all variants not included in the hotspots file.
Include Unannotated is selected.	Returns all variants not included in the hotspots file.

Location filter

This filter can be added to filter chains in the software to detect variants in specific positions, such as exonic. Filter conditions include the ability to select all locations or specific locations in filtered analysis results.

The filter returns variants in samples that match the selected filter locations.

The variants returned by the filter depend on the options that are selected.

The filter returns all CNV and fusion variants regardless of the selected options.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a location annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a location annotation.
Include Unannotated is selected.	Returns all variants that do not have a location annotation.

Minor Allele Frequency filter

This filter can be added to filter chains in the software to detect variants with minor allele frequencies that match those in the dbSNP database from the 1000 Genomes Project.

Filter conditions include the ability to set the minor allele frequency range from 0.0 to 0.5.

The variants returned by the filter depend on the options that are selected.

The filter applies the following conditions regardless of the selected options.

- The hit level allele filter threshold is matched against the exact allele.
- $RMA \neq 1$ — The filter threshold is not matched if it is a reference allele.
- Returns all the variants that are not SNV/INDEL, MNV, REF, NOCALL, FLT3ITD, ProcControl.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.

(continued)

Option	Variants returned
Include Unannotated is selected.	• Returns all variants without hit level allele for dbSNP annotations. • Returns all variants with hit level allele and RMA = 1 for dbSNP annotations. • Returns all variants without dbSNP annotation. • Returns all CNV and fusions variants.

OMIM filters

These filters can be added to filter chains in the software to detect variants that match those in the Online Mendelian Inheritance in Man® (OMIM) database.

Filter conditions include the ability to search for all (In) or none (NOT In) and to include or exclude unannotated variants in filtered results.

In addition, one or more specific annotation values can be included in the filter chain. Examples include 5-hydroxytryptamine (serotonin) receptor-1A and A kinase (PRKA) anchor protein 17A. A partial list of specific annotation values can be viewed when the option of **All** is selected.

The filter returns variants that match those in the OMIM database.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have an OMIM annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have an OMIM annotation.
Include Unannotated is selected.	Returns all variants that do not have an OMIM annotation.

Oncomine™ filter

This filter can be added to filter chains in the software to detect variants that are annotated with the Oncomine™ Variant Annotator software module in specific Oncomine™ panels. This is the default filter for most Oncomine™ assays. It presents Oncomine™ driver variants that are based on their Oncomine™ gene class and Oncomine™ variant class annotations.

Oncomine™ assays for Ion AmpliSeq™ HD panels in Genexus™ Software include the Oncomine™ Variant Annotator software module. The software module integrates analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types, and annotates variants relevant to cancer with Oncomine™ gene class and Oncomine™ variant class information. For more information and a full list of annotation rules for each Oncomine™ assay, see “Annotation criteria for the Oncomine™ Variant Annotator” on page 527, or contact a Field Bioinformatics Specialist (FBS).

The Oncomine™ filter returns variants that have Oncomine™ gene class and Oncomine™ variant class annotation.

This filter can be used only if an Oncomine™ assay was used. You can apply filter chains that include the Oncomine™ filter only if the assay that was used for the analysis includes the Oncomine™ Variant Annotator software module. Examples of filter chains that include this filter are the Oncomine™ Variants and Oncomine™ Extended filter chains.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants that are annotated with the Oncomine™ Variant Annotator software module.
NOT is selected.	Returns all variants that do not have Oncomine™ Variant Annotator software module annotations.
Include Unannotated is selected.	Returns all variants that do not have Oncomine™ Variant Annotator software module annotations.

Pfam filters

These filters can be added to filter chains in the software to detect protein domain families in coded proteins as defined by the Pfam database.

Filter conditions include the ability to search for all (In) or none (NOT In) and to include or exclude unannotated variants in filtered results.

In addition, one or more specific annotation values can be included in the filter chain. Examples include 7 transmembrane receptor (Secretin family) and Activator of apoptosis harakiri. A partial list of specific annotation values can be viewed when the option of **All** is selected.

The filter returns variants that match the variants in the Pfam database.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a Pfam annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a Pfam annotation.
Include Unannotated is selected.	Returns all variants that do not have a Pfam annotation.

Phred QUAL Score filter

This filter can be added to filter chains in the software to filter by the Phred QUAL score. Phred QUAL scores are defined as a property that is logarithmically related to the base-calling error probabilities.

The filter returns Phred Qual scores for variants calls. Scores are calculated in the following way.

- For variants, the score is computed by the posterior probability that the variant allele frequency is greater than the minimum allele-frequency that is specified for the variant type.
- For reference calls, the score is computed by posterior probability that the variant allele frequency is below this threshold.

Posterior probability is computed conditional on the reads observed and includes sampling variability.

Filter conditions include the ability to set a range from 0.0 to 1,000,000.0. The range boundaries are included. In addition, unannotated variants can be included or excluded in filtered results.

The variants returned by the filter depend on the options that are selected.

The filter returns the Phred Qual Score not equal to '.' regardless of the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all fusion variants.

PhyloP Scores filter

This filter can be added to filter chains in the software to measure evolutionary conservation at individual alignment sites. Interpretations of the scores are compared to the evolution that is expected under neutral drift. Positive scores measure conservation, which is slower evolution than expected, at sites that are predicted to be conserved. Negative scores measure acceleration, which is faster evolution than expected, at sites that are predicted to be evolving rapidly.

This filter is useful to evaluate the signatures of selection at particular nucleotides or classes of nucleotides. For example, as with third codon positions, or first positions of miRNA target sites, the absolute values of phyloP scores represent $-\log$ p-values under a null hypothesis of neutral evolution.

Filter conditions can be set to a score range from -20.0 to 30.0 . The range boundaries are included.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all CNV and fusion variants.

Possible Polyploidy Alleles filter

This filter can be added to filter chains in the software to detect alleles that have passed all variant calling filters used by the Torrent Variant Caller (TVC) module but missed by genotype.

This filter is useful for identification of variants beyond diploid genomes, that is, cancer cells or polyploidy species.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that are Possible Polyploidy allele.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that are not Possible Polyploidy allele.
Include Unannotated is selected.	Returns all variants that are not Possible Polyploidy allele.
YES is selected.	Find alleles that have passed all filters.

(continued)

Options	Variants returned
NO is selected.	Find alleles that have <i>not</i> passed all filters, in addition to genotype.
Both YES and NO are selected.	Find all variants, including Possible Polyploidy allele and not Possible Polyploidy allele.

PValue filter

This filter can be added to filter chains in the software to narrow results based on the p-value of variants. The p-value quantifies the statistical significance of variant calls.

Filter conditions include the ability to set a range from 0.0 to 1.0.

The filter returns p-values for variants calls in the samples.

The variants returned by the filter depend on the options that are selected.

Option	Variants returned
NOT is deselected.	Returns all variants within the specified range, including the range boundaries.
NOT is selected.	Returns all variants outside of the specified range.
Include Unannotated is selected.	Returns all fusions variants.

UCSC Common SNPs filter

This filter can be added to filter chains in the software to detect variants that match those in the UCSC Genome Browser Common SNPs database. The filter returns variants in samples that match the UCSC Common SNPs database.

The variants returned by the filter depend on the options that are selected.

This filter returns all the variants that are not REF, SNV, and NOCALL regardless of the selected options.

Option	Variants returned
NOT is deselected.	Returns all variants that are annotated with a dbSNP annotation and the COMMON flag is present.
NOT is selected.	Returns all variants that are annotated with a dbSNP annotation and the COMMON flag is absent.
Include Unannotated is selected.	- Returns all variants that have a dbsnp annotation but the COMMON flag is absent. - Returns all variants that do not have a dbsnp annotation. - Returns all variants other than REF, SNV, and NOCALL.

User Classification filter

This filter can be added to filter chains in the software to narrow results to variants that for which a **User Classification** is added. These classifications are added to results by users.

The classifications, and any unique classifications that users have defined and saved before the creation of the filter chain, are available in this filter.

Filter conditions include the ability to include all classifications (**In**), none (**NOT In**), or to select specific classifications in filtered results.

The filter returns variants that are classified by software users.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that are classified.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that are not classified.
Include Unannotated is selected.	Returns all variants that are not classified.

Variant Effect filter

This filter can be added to filter chains in the software to detect the effect of variants on coding sequences.

Filter conditions include the ability to select all (**In**), none (**NOT In**), or to select specific values, and to include or exclude unannotated variants in filtered analysis results.

The filter returns variants in samples that match the selected variant effects.

The variants returned by the filter depend on the options that are selected.

The filter returns all CNV and fusion variants regardless of the selected options.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a variant effect annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a variant effect annotation.
Include Unannotated is selected.	Returns all variants that do not have a variant effect annotation.

Variant Subtype filter

This filter can be added to filter chains in the software to detect variant subtypes.

Filter conditions include the ability to select all or specific variant subtypes in filtered analysis results.

The filter returns variants that match the selected specific variant subtypes.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a subtype annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a subtype annotation.
Include Unannotated is selected.	Returns all variants that do not have a subtype annotation.

Variant Type filter

This filter can be added to filter chains in the software to detect variants that match the variant types.

Filter conditions include the ability to select all variant types or specific variant types in filtered analysis results.

Note: The FLT3ITD variant type is detected only in OncoPrint™ Myeloid Assay analysis workflows.

The variants returned by the filter depend on the options that are selected.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a variant type.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a variant type.
Include Unannotated is selected.	Returns all variants that do not have a variant type.

Zygosity filter

This filter can be added to filter chains in the software to detect variants that are homozygous or heterozygous.

Filter conditions include the ability to search for both homozygous and heterozygous variants, or either of them, in filtered analysis results.

The filter returns variants in analyses that are homozygous or heterozygous.

The variants returned by the filter depend on the options that are selected.

This filter returns all CNV and fusions variants regardless of the selected options.

Options	Variants returned
NOT is deselected and Specific Annotations is selected.	Returns all variants with an exact match for selected value or values.
NOT is deselected and Contains is selected.	Returns all variants that match the selected value or values pattern.
NOT is deselected and In is selected.	Returns all variants that have a zygosity annotation.
NOT is selected and Specific Annotations is selected.	Returns all variants that do not exactly match the selected value or values.

(continued)

Options	Variants returned
NOT is selected and Contains is selected.	Returns all variants that do not match the selected value or values pattern.
NOT is selected and In is selected.	Returns all variants that do not have a zygosity annotation.
Include Unannotated is selected.	Returns all variants that do not have a zygosity annotation.

System-installed plugins

Genexus™ Software includes system-installed plugins that expand the analysis capabilities of the software. System-installed plugins are locked and cannot be changed.

System-installed plugin	Description
coverageAnalysis	This plugin provides statistics and graphs that describe the level of sequence coverage that is produced for targeted genomic regions.
CustomerSupportArchive	This plugin generates a downloadable archive that a technical support representative can use to troubleshoot and diagnose problems with sequencing runs or with Genexus™ Software. The archive contains log files and other technical data about the software and other files from sequencing runs on a Genexus™ Integrated Sequencer. For more information, see “Download a Customer Support Archive” on page 586.
generateGenome	This plugin generates a consensus genome for an individual sample that is based on a reference genome and variants that are called in a sequencing run. The full reference genome is returned in a compressed FASTA file with regions that are not sufficiently covered, or otherwise of high uncertainty, denoted by 'N' characters for bases. It is therefore primarily suited to small genomes, such as viruses, but may also be used with larger targeted genomes that have multiple contigs or chromosomes.
IonReporterUploader	This plugin allows you to directly transfer sample results files that are generated in Genexus™ Software to Ion Reporter™ Software.
molecularCoverageAnalysis	This plugin provides statics and graphs that describe the level of sequence molecular coverage that is produced for targeted genomic regions.
sampleID	This plugin produces a unique identification code (sampleID) for each barcode in a sample.

System-installed sample attributes

The following table lists and describes examples of system-installed sample attributes. System-installed sample attributes cannot be edited.

When samples are created, the attributes that are shown in the software depend on the application category that is selected. User-defined sample attributes are not listed in this table.

System-installed sample attribute	Description
Adverse Events	The adverse events associated with the sample.
Age at Diagnosis (Yrs)	The age at diagnosis, in years, of the individual from whom the sample was collected.
Biological Sex (display)	The biological sex of the individual from whom the sample was collected. Select Male , Female , or Unknown .
Cancer Grade	The grade of the cancer of the collected sample. Select GX , G0 , G1 , G2 , G3 , or G4 .
Cancer M Stage (Metastatic)	The metastatic stage of the cancer. Select MX , M0 , or M1 .
Cancer N Stage	The stage of the cancer that indicates lymph node involvement. Select NX , N0 , N1 , N2 , or N3 .
Cancer Sample Site	Location from where the sample was collected. Select Primary or Metastasis .
Cancer Stage^[1]	The stage of the cancer from which the sample was collected. Select Stage 0 - Stage IV , Unknown , or Other .
Cancer T Stage	The size and extent of the tumor. Select TX , T0 , T1 , T2 , T3 , or T4 .
Cancer Type^[1]	The type of cancer that is represented by the collected sample. Select the type of solid or hematologic cancer. If cancer type is unknown, select Unknown Primary Origin .
Case ID	A unique identifier for the research experiment.
Case Location (Country)	The country in which the sample originated or is located.
Case Location (State/Province/Territory)	The state, province, or territory where the sample is located.

(continued)

System-installed sample attribute	Description
% Cellularity	The percentage of tumor cells over normal cells in the collected sample. This is a whole number between 1 and 100. The % Cellularity attribute applies only to FFPE samples. IMPORTANT! • If not set, % Cellularity is assumed to be 100% in calculations that use this attribute. • % Cellularity is a required attribute for CNV analyses. Do not leave the field blank. • (FFPE samples only) If % Cellularity value is set to <100, then the magnitude of copy number gain or loss can be decreased. For more information, see “CNVs table” on page 153.
Collection Date ^[1]	The date, in year-month-day format, that the sample was collected. Click  Calendar to select the date in the correct format.
Date of Birth	The date, in year-month-day format, of the birth of the individual from whom the sample was collected.
Estrogen Receptor Status (Breast Cancer)	The Estrogen Receptor (ER) status of the collected sample. This refers to the presence or absence of estrogen receptors on the surface of cells in a sample. Select ER-Positive, ER-Negative, or Unknown.
Gender ^[1]	The biological sex of the collected sample. Select Female, Male, or Unknown. IMPORTANT! Male or Female must be selected for correct measurement of sex chromosome copy number variation.
Is Deceased?	Indicates whether the sample is collected from an individual who is deceased. Select Yes, No, or Unspecified.
Lab Location (Country)	Select the country where the laboratory is located.
Lab Location (State/Province/Territory)	Select the state, province, or territory where the laboratory is located.
Laboratory Name	Enter the name of the laboratory associated with the collected sample.
% Necrosis	The percentage of cellular necrosis in the collected sample. Enter a whole number between 1 and 100.
Notes	An open-entry field used to enter more information about the collected sample.

(continued)

System-installed sample attribute	Description
Progesterone Status (Breast Cancer)	Progesterone Receptor (PR) status of the collected sample. Select PR-Positive , PR-Negative , or Unknown .
Sample Collected By	The name of the laboratory or organization that collected the sample.
Sample ID DNA Barcode	The identifier of the DNA barcode used in the sequencing run.
Sample Name^[1]	A unique identifier representing the collected sample. IMPORTANT! To prevent erroneous sample selection during run planning, ensure that you assign a unique and distinguishable sample name for each sample. The sample name can contain only alphanumeric characters (0–9, Aa–Zz), periods (.), underscores (_), or hyphens (-), cannot contain spaces, and is limited to a maximum of 50 characters. Note: • Samples that have been used in a run cannot be deleted. • To prevent duplication, the software checks all sample names and returns an error message if a nonunique sample name is detected.
Sample Type^[1]	A term that describes the collected sample, for example, bone marrow. Select a sample type from the list, or select Other, then enter a user-defined sample type.
Treatment	The current or prior treatment that is related to the cancer sample.
Treatment Response	The response to treatment that is related to the cancer sample.

^[1] Required attributes within one or more application types.



Troubleshooting

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Information is provided to help you troubleshoot issues. If you need more assistance, contact Technical Support.

Contact Support

You can access Genexus™ Software technical support from the software.

1. Scroll to the bottom of the screen.
2. Click **Support** to open <https://www.thermofisher.com/contactus>, where you can request help and technical support.

Download a Customer Support Archive

A Customer Support Archive (CSA) file is a downloadable archive that a support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Software. The archive contains log files and other technical data about the software and other files from sequencing runs.

CSA files are generated for sequencing runs in which the CustomerSupportArchive plugin is run. The plugin can be run automatically during the sequencing run, or manually after a sequencing run is complete.

1. Click **Results ▶ Run Results**, then click a run name of interest.
2. In the **Run Results** screen, click ... **(More Options) ▶ CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.

An XZ compressed TAR archive (TXZ) file is downloaded to your computer and is ready to be shared with a support representative.

Your support representative might recommend one of the alternative ways to download a CSA file.

- In the **Sample Results** screen, place the pointer over the row of the sample result of interest, then click **CSA**.
- In the **Sample Results** screen, click a sample name, then from any tab in the **Results** screen, click **... (More Options) ▶ CSA**.
- Click **Results ▶ Run Results**, then place the pointer over the row of the run result of interest, then click **CSA**.

Download instrument diagnostic information

Support representatives may request that you share instrument diagnostics information to help diagnose issues remotely. You can download a ZIP file in Genexus™ Software that contains the instrument diagnostic information, then share the ZIP file with support representatives.

Before you can download the diagnostics ZIP file, you must first upload the instrument diagnostic information from the Genexus™ Integrated Sequencer or from the Genexus™ Purification System to Genexus™ Software.

- To upload the diagnostic information from the sequencer instrument, on the **System Tools** touchscreen, touch **Upload diagnostics**. The diagnostic information is uploaded to Genexus™ Software. For more information, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. [MAN0017910](#)).
- To upload the diagnostic information from the purification instrument, see the *Genexus™ Purification System User Guide* (Pub No. [MAN0018475](#)).

1. In the menu bar, click  **(Settings) ▶ Services**.

2. Scroll to the bottom of the screen to the **Download Instrument Diagnostics** section.

3. In **Select Instrument**, select the instrument.

4. Click **Download**.

The diagnostic ZIP file that contains the instrument diagnostics information and factory test logs is downloaded to your computer.

Troubleshoot Genexus™ Software

Observation	Possible cause	Recommended action
Cannot sign in to the Genexus™ Software	You have either forgotten your password or are signed out due to several failed login attempts.	Contact the Genexus™ Software system administrator.

Observation	Possible cause	Recommended action
Failure of multiple samples import	One or more entries in the sample import file contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors.	Check each entry for correct formatting, correct any errors, and repeat the import.
	Blank rows were copied into the sample import file from a different source.	Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean sample import template, and be careful to copy only those rows that contain actual data.
	The sample import file contains a nonunique sample name.	Every sample name in the software must be unique. Ensure that the sample import file does not contain any duplicate sample names, then repeat the import. Note that the system check is not case-sensitive, so a sample name of ABC1 conflicts with abc1.
	The headings in the sample import file do not match the sample attributes in the software.	The headings in the sample import file must exactly match the sample attribute names in the software. Check the headings for spelling or other errors. Map a sample attribute if needed.
Library batch import fails	One or more entries in the library batch import file contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors.	Check each entry for correct formatting, correct any errors, and repeat the import.
	Blank rows were copied into the library batch import file from a different source.	Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean library batch import template, and be careful to copy only those rows that contain actual data.
	The library batch import file contains a nonunique Library Batch ID .	Each Library Batch ID in the software must be unique. Ensure that the library batch import file does not contain any duplicate IDs, then repeat the import. The system check is not case sensitive. For example, a Library Batch ID of ABC1 conflicts with abc1.
	A sample name entered in the library batch import library batch import file does not match a sample name that is listed in the Manage Samples screen.	Ensure that the sample names entered into the library batch import file are correct and match a sample name in the software.

Observation	Possible cause	Recommended action
Library batch import fails (continued)	The Barcode ID name format does not exactly match the format that is used in the Prepare Library Batch dialog box.	Use the name format following the Barcode ID name format found in the Barcode Set reference lists (Settings ▶ References ▶ Barcode Set), for example: IonDual_0101 through IonDual_0196, or IonHDdual_0101 to IonHDdual_0132.
	An invalid library, control, or panel kit barcode has been entered in the library batch import file.	Ensure that you have correctly entered a valid kit barcode in the appropriate table cell of the library batch import file.
	The library batch import file template that you used is from a previous software version.	New fields in the library batch import template file can be added with new software versions. Ensure that you download the template file from the current software version.
Cannot upload my hotspots file or panel files	Issues with BED file format or the files do not end in .bed .	Ensure that your files use the correct BED format and have a .bed extension.
Library QC metrics failed	Library metrics failed in a run, while Run QC and Control QC metrics pass.	Make library QC settings less restrictive.
		Check sample concentration and quality.
		Make a new sample preparation if quality is questionable, if possible.
	Library metrics failed in a run, while Run QC and Control QC metrics also failed.	Confirm that correct consumables were loaded in the run.
		Upload instrument diagnostics for review by support personnel.
Loading metrics are reported as 0	- Chip failure - Defective pariposer - Chip leak	Refer to the troubleshooting sections in the Genexus™ Integrated Sequencer user guide for possible causes of chip or pariposer failure.
	Sample preparation failure resulting in loss of sample	Refer to the troubleshooting sections in for sample, library, and template preparation in the <i>Ion AmpliSeq™ Library Preparation on the Ion Chef™ System User Guide</i> (Pub. No. MAN0013432).
I want more/less stringent variant calling	Parameters chosen while setting up your assay are not appropriate.	Create a new assay with new variant calling parameters, then reanalyze using the new assay. You have an option to reanalyze from the Results screen.
Variants tab is missing hotspot entries Details: The remaining entries are present.	Hotspot BED file contains entries that are incorrectly formatted.	Check that BED file entry is correctly formatted. See the following examples: SNP entry: chr1 2337276 2337277 SVA_322 0 + REF=C;OBS=T;ANCHOR=G AMPL Deletion entry: chr1 201341175 201341180 SVA_497 0 + REF=AGAAG;OBS=;ANCHOR=C AMPL Insertion entry: chr1 236978992 236978992 SVA_621 0 + REF=;OBS=TCTG;ANCHOR=T AMPL
		Ensure that the REF values match the actual reference coordinate of hg19.

Observation	Possible cause	Recommended action
Target summary does not meet expectation Details: For example: - the number of targets does not match - coverage uniformity is below expected value	Incorrect barcode used for sample preparation.	Verify that each Sample name-to-Barcode ID pair is entered correctly, then repeat the run using the corrected sample library. Re-prepare the sample library from the sample if necessary.
	Wrong panel was selected while setting up your assay or wrong assay was selected while setting up your run plan.	If you start a run with the wrong panel or assay, there is no way to reanalyze using the software. Use your library to prepare another template reaction.
No information for my loci of interest in the results	The wrong hotspot or panel file is associated with the assay.	1. Check the correct hotspot file, target region file, and panel files are associated with the assay. If either is incorrect, create a new assay. 2. Plan a new run for the library preparation or sample with the correct assay. 3. Repeat the run for library preparation or sequencing of the sample.



Documentation and support

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■ Customer and technical support	591

Related documentation

Related software documentation is available at <http://www.thermofisher.com/ngssoftwaresupport>.

Document	Publication number
<i>Oncomine™ BRCA Assay GX User Guide</i>	MAN0018514
<i>Genexus™ Integrated Sequencer User Guide</i>	MAN0017910
<i>Genexus™ Integrated Sequencer Quick Reference</i>	MAN0017912
<i>Genexus™ Deep Clean Kit User Guide</i>	MAN1000882
<i>Genexus™ Purification System User Guide</i>	MAN0018475
<i>Ion Reporter™ Software 5.20 User Guide, or later.</i>	MAN0028231
<i>Oncomine™ Precision Assay GX User Guide</i>	MAN0018508
<i>Oncomine™ Comprehensive Assay v3 GX User Guide</i>	MAN0018512
<i>Oncomine™ TCR Beta-LR Assay GX User Guide</i>	MAN0018513
<i>Ion AmpliSeq™ Designer Help</i>	MAN0018937
<i>Ion AmpliSeq™ Library Preparation on the Ion Chef™ System User Guide</i>	MAN0013432

Customer and technical support

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 - User guides, manuals, and protocols
 - Certificates of Analysis
 - Safety Data Sheets (SDSs; also known as MSDSs)

Note: For SDSs for reagents and chemicals from other manufacturers, contact the manufacturer.

Genexus™ Software definitions

adapter dimer (ISPs)

Primer and adapter sequences that anneal during library preparation or templating to form a short fragment that can be sequenced. These adapter dimer ISPs generate unusable reads that the software filters out of the final sequencing data. High levels of adapter dimer ISPs can reduce the number of valid reads.

aligned read length

The aligned length of a read at a given accuracy threshold is defined as the greatest position in the read at which the accuracy in the bases up to and including the position meets the accuracy threshold. For example, the AQ17 length of a read is the greatest length at which the read error rate is 2% or less. The "perfect" length is the longest perfectly aligned segment. For all of these calculations, the alignment is constrained to start from position 1 in the read—that is, no 5' clipping is allowed.

aligned reads

The number of bases covered by reads aligned to the reference sequence.

AQ score

An alignment quality (AQ) score defines read accuracy when compared to the reference sequence. The discrepancy can be biological (real variant) or technical (sequencing error). For example, an AQ score of 17 represents 2% aligned read error rate, while an AQ score of 20 represents 1% aligned read error rate when compared to the reference sequence.

archive

The process that occurs when data for runs on the Ion Torrent™ Genexus™ Integrated Sequencer are moved to the external storage and is no longer available for review on the sequencer.

average base coverage depth

The average number of reads of all targeted reference bases.

average base read depth

The average number of reads of all targeted reference bases that were read at least once.

backup

The process of data being copied to an external storage device or system from the Ion Torrent™ Genexus™ Integrated Sequencer. Backups of data are scheduled based on backup settings, and data that is backed up is not deleted from the sequencer. When data are backed up, the same data remains on both the Genexus™ Integrated Sequencer and on the external storage system.

BAM file

A BAM (binary alignment map) file (.bam) is the binary version of a SAM (sequence alignment map) file (.sam). A SAM file is a tab-delimited text file that contains sequence alignment data. A BAM file contains aligned reads sorted by reference location.

barcode

A barcode is a machine-readable code in the form of numbers and a pattern of parallel lines of varying widths, printed on and identifying a product.

There are several applications for barcodes. Libraries can be molecularly barcoded with unique nucleic acid sequence identifiers. Library barcodes are used during data analysis to sort the sequencing results from sequencing reactions that contain combined libraries. Chips and sample tubes also contain unique numeric barcodes that aid in the setup of the experimental analysis workflow.

base calling

Base calling is the process by which raw data from the sequencing instrument is converted to nucleotide sequences. This is performed by base calling software that is usually run from the instrument itself.

basecalling input file

Signal processing input files are converted to a single condensed basecalling input file that represents the processed signal. Basecalling input files are required files for basecalling.

bead loading

The percentage of chip wells that contain live ISPs. The percentage value considers only potentially addressable wells. Bead loading is calculated as number of loaded ISPs divided by the number of potentially addressable wells.

bp

Abbreviation for "base pair(s)".

cellularity (%)

The percentage of tumor cells in a given sample.

BED file

Browser Extensible Data file—BED file—defines chromosome positions or regions.

CNV

Copy number variation (CNV) is the variation in copy number of any given gene between two samples. CNV is a phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals in the human population.

CRAM file

A CRAM (Compressed Reference-oriented Alignment Map) file (.cram) is a file format that is similar to the BAM file format but includes improved file compression and a restructured column-oriented binary container format. As data volumes increase in genetic software, the CRAM format was designed to keep file sizes manageable by reducing the disk space used by alignment data.

contig

A contig (from contiguous) is a set of overlapping DNA segments that together represent a consensus region of DNA.

control sequence

Control nucleic acid sequences can be added to DNA or RNA samples to facilitate post-sequencing data analysis. Two types of control sequences can be used during sample preparation. ERCC RNA Spike-In Mix is used with RNA samples to achieve a standard measure for data comparison across gene expression experiments. Ion AmpliSeq™ Sample ID Panel, comprised of nine specially designed primers, can be added prior to template amplification to generate a unique ID for each sample during post-sequencing analysis.

CSV file

A comma-separated values (CSV) file is a delimited text file in which each line represents a data record with information fields separated by a comma. A CSV file stores tabular data (numbers and text) in plain text. Each line of the file is a data record.

CSV files are easily opened using spreadsheet software, such as Excel™ or Apache® OpenOffice™ Calc, where each comma-separated field is listed in a separate column.

DAT file

A DAT file is a generic data file. It contains data that may be opened or referenced by a specific application. While some DAT files can be opened directly, most contain program data that is referenced the program when it is running. Therefore, most DAT files are not meant to be opened manually.

empty wells

Wells on a chip that do not contain an ISP.

enrichment (%)

Predicted number of live ISPs that have a key signal identical to the library key signal. The percent enrichment value reported is the number of loaded ISPs that are library ISPs, after taking out the test fragment ISPs.

FASTA file

A FASTA file is a text-based format for representing either nucleotide sequences or peptide sequences, in which base pairs or amino acids are represented using single-letter codes. A sequence in FASTA format begins with a single-line description, followed by lines of sequence data.

FASTQ file

A FASTQ file is a text-based format for storing both a biological sequence (usually nucleotide sequence) and its corresponding quality scores. Both the sequence letter and quality score are each encoded with a single ASCII character for brevity.

FD (flow disruptiveness)

A data filtering parameter that is used instead of INDEL, SNP, and MNP.

final library reads

Number (and percentage) of reads, passing all filters that are recorded in the unmapped BAM file. This value may be different from the total number of reads located in the Library Summary Section due to specifications associated with read trimming beyond a minimal requirement resulting in total number of reads being slightly less than Final Library Reads.

flow

A flow is the event of exposing a chip to one particular dNTP (deoxyribonucleotide triphosphate), followed by a washing step.

flow order

The order in which a chip is exposed to each particular dNTP. The default Samba flow order consists of a 32-base sequence, repeated. This flow order resists phase errors by providing opportunities for out-of-phase molecules to catch up and is designed to sample all dimer (nucleotide pair) sequences efficiently. Samba is the default flow order because it improves sequencing accuracy for longer reads by resisting phase errors.

flow transfer

Progress of the sequencing run expressed as number of total flows completed. For example, a sequencing run set to 500 flows shows 250/500 flows in the status column of the **Runs in Progress** table when the sequencing run is half completed.

fusions

A targeted sequencing technique used for detection and annotation of gene fusions (or translocation of genetic material) in samples.

homologous recombination repair

Homologous Recombination Repair (HRR) is a pathway to repair double-stranded DNA breaks which are generated as by-products of cellular metabolism, DNA replication errors, or exogenous factors.

hotspots file

A BED or a VCF file that defines variants. Specifying a hotspots file to use in a run enables the Torrent Variant Caller (TVC) module to identify variants that may be present in sample DNA. A hotspots file instructs the Torrent Variant Caller (TVC) module to include these positions in its output files, including evidence for a variant and the filtering thresholds that disqualified a variant candidate. A hotspots file does not affect other parts of the analysis pipeline.

If you do not specify a hotspots file, the software tells only the difference between your sequence and the reference genome.

IA

Isothermal application (IA) is a template preparation technique that uses nonemulsion isothermal reaction to clonally amplify DNA onto an ISP surface.

INDEL

INDEL is an abbreviation used to designate an insertion or deletion of bases in the genome of an organism.

intermediate files

Intermediate files contain information used for debugging runs.

ISPs

Ion Sphere™ Particles (ISPs) are particles that contain bound copies of a single (ideally) DNA fragment amplified during template preparation.

key signal

Average 1-mer signal in the library key.

library ISPs

Live ISPs that have a key signal identical to the library key signal.

LIMS metadata

Laboratory Information Management System (LIMS) software is used for recording sequencing metadata. Entered text is associated with the Planned Run and can be extracted using APIs for LIMS consumption.

live ISP

An ISP with a signal of sufficient strength and composition to be associated with the library or test fragment key.

low quality ISP

An ISP with a low or unrecognizable signal.

library key

A short known sequence of bases used to distinguish a library fragment from a test fragment (for example, "TCAG").

MAPD

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on Aneuploidy run results and other runs that detect CNVs.

MAPD is one metric that is used to determine whether the panel data are useful for copy number variation (CNV) run results.

MAPD is defined as the Median of the Absolute values of all Pairwise Differences between log2 ratios of each tile for a run. Tiles roughly correspond to amplicons in an Ion AmpliSeq™ assay. Each pair is defined as adjacent in terms of genomic distance. Tiles corresponding to copy number amplicons and other amplicons are being treated equally as no differences in variability are seen between these types. Then, any two log2 ratios that are adjacent on the genome are a pair. Except at the beginning and the end of a chromosome, every log2 ratio belongs to two pairs.

Formally, if x_i is the log2 ratio at position i , with i ordered by genomic position:

```
MAPD = median ( | x(i - 1) - x(i) | )
```

MAPD is an estimate of copy number variability in each sequencing run that is similar to standard deviation (SD). If one assumes the log2 ratios are distributed normally with mean 0 and a constant SD, then MAPD/0.95 is approximately equal to SD. However, unlike SD, using MAPD is robust against high biological variability in log2 ratios induced by known conditions such as cancer.

Regardless of the source of the variability, increased variability decreases the quality of CNV calls.

mapped reads

Sequencing reads that have been mapped to the reference sequence.

mean raw accuracy

Average raw accuracy of 1-mers at a specific position in the read.

microsatellite instability

Microsatellite instability (MSI) is a form of genomic instability in the replication of repetitive DNA that leads to the appearance of multiple alleles at microsatellite loci. MSI markers can be detected with some DNA and DNA and Fusions assays.

missense SNV

A point mutation that changes the amino acid of the respective protein. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

MNP

Multiple nucleotide polymorphism (MNP) is a genetic mutation in an allele that differs from the reference allele of the same length by >1 nucleotide.

nonsense SNV

A point mutation that changes one of the 20 amino acids into a stop codon, hence a shorter or unfinished protein product. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

on target reads

Sequencing reads mapped to any target region of the reference. A read is considered to be on target if at least one aligned base overlaps a target region. A read that overlaps a targeted region but where only flanking sequence is aligned, for example, due to poor matching of 5' bases of the read, is not counted.

primer dimer

A primer dimer (PD) is a potential by-product in PCR, a common biotechnological method. As its name implies, a PD consists of primer molecules that have attached (hybridized) to each other because of strings of complementary bases in the primers.

primer dimer ISP

An ISP that carries an insert length of less than 8 base pairs.

Q score

Phred quality score (Q score) is used to measure the accuracy of the nucleotide sequence generated by the sequencing instrument. The Q score represents the probability that a given base is called incorrectly by the sequencer.

read length

The length of called reads measured in base pairs.

read

The sequence of a section of a unique fragment obtained after the end of the sequencing process.

reference library

A consensus nucleotide sequence that represents the genome of a particular species. The results from a sequencing run are compared to the reference library to identify sequence variants.

SAM file

A SAM (sequence alignment map) file (.sam) is a tab-delimited text file that contains sequence alignment data. It stores biological sequences aligned to a reference sequence.

sample

Genetic material from one source (for example, DNA from one individual).

signal processing input files

Signal processing input files consist of the raw voltage measurement data collected during the sequencing run. These files are required to reanalyze the run from signal processing.

SNP

Single nucleotide polymorphism (SNP) is a genetic mutation in an allele that differs from the reference allele of the same length by one nucleotide.

SSH

SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives users, particularly system administrators, a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

structural variants

Genetic mutations that cause a change in the organism's chromosome structure, such as insertions, deletions, copy number variations, duplications, inversions, and translocations.

target base coverage

Summary statistics for targeted base reads of the reference. A base covered by multiple target regions is counted only once per sequencing read.

target regions file

A BED file that specifies regions that a panel represents such as the amplified regions that are used with target sequencing. The complete software analysis pipeline, including plugins, is restricted to include only these specified regions instead of the entire reference library.

test fragment

A test fragment (TF) is a known nucleotide sequence that is used to monitor system characteristics.

test fragment ISPs

Live ISPs with a key signal that is identical to the test fragment key signal.

test fragment key

A test fragment key (TF key) is the nucleotide sequence that is used to identify test fragment reads.

TF key peak counts

Signal strength of the first three bases of the TF key.

total reads

Total number of filtered and trimmed reads independent of length reported in the output BAM file.

TSV file

A tab-separated values (TSV) file is a tab-delimited file that is used with spreadsheet software. TSV files are essentially text files, and the raw data can be viewed by text editors, though they are often used when moving raw data between spreadsheets. See also *VCF file*.

uBAM file

An unmapped BAM (uBAM) file is a variant form of the BAM file format in which the read data does not contain mapping information. This is basically an "off-label" use of the BAM format (which was specifically designed to contain mapping information) that is used for data management reasons. It allows you to attach metadata to the reads from as early on in the analysis process as possible.

unaligned reads

Nucleotide bases covered by reads that are not aligned to the reference.

uniformity of base coverage

The percentage of bases in all targeted regions (or whole genome) covered by at least 0.2X the average base coverage depth.

usable sequence / usable reads

Usable reads consist of library ISPs that pass the polyclonal, low quality, and primer dimer filters.

VCF file

A variant call format (VCF) file specifies a variant of interest and its location. This file stores the differences between the BAM file and the reference file.

wells with ISPs

Number of wells that were determined to be positive for the presence of an ISP within the well. Wells that contain ISPs have a delayed pH change due to the presence of an ISP that slow the detection of the pH change from the solution.

XLS file

Files that use the .xls extension. XLS files can be created by Excel™ and other similar spreadsheet programs.

