appliedbiosystems

Automated Axiom™ Analysis v3.1

USER GUIDE

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Revision history: MAN1000174

Revision	Date	Description
C00	October 2025	v3.1 release. Documented compatibility with Thermo Fisher's SwiftArrayStudio™ Microarray Analyzer. Also documented a new sample QC report option, viewing marker metrics summary as a table feature, and that the VCF Export now accepts and processes Dual workflow samples.
B00	March 2025	v3.0 release. Documented new library package management and disk usage features.
A00	May 2024	Initial release in Thermo Fisher Scientific document control system. MAN1000174 supersedes legacy Affymetrix publication number 703513 and Revision 3 is now A00 to comply with new numbering guidelines. v2.0 release. Documented new user interface and features, including the BloodGenomiX Reporter, Multiplate batching, Parallelization, and additional extension workflows.
2	January 2022	v1.1 release: Added Best Practices with Dual Workflow feature. Optional settings for Sample Name Mapping, Filter File, SNP List Filter, SNP Mapping File, and SNP Allele Swap are now enabled for arrays that have AxLE Export enabled. Option to include SNP Metrics in the AxLE SNP Map Export file.
1	October 2019	Initial release.

Important Software Licensing Information

Your installation and/or use of this Automated Axiom Analysis software is subject to the terms and conditions contained in the End User License Agreement (EULA) which is incorporated within the Automated Axiom Analysis software, and you will be bound by the EULA terms and conditions if you install and/or use the software.

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1 Before use

Introduction

Automated Axiom™ Analysis (AutoAx) enables analysis of CEL files upon completion of a plate scanning. AutoAx is compatible with Thermo Fisher's GeneTitan™ or SwiftArrayStudio™ .

IMPORTANT! AutoAx users are expected to be familiar with Axiom™ Analysis Suite and have its User Guide (P/N MAN0027928) on hand for reference and detailed instructions.

Applications included for SwiftArrayStudio users

- **SwiftArrayStudio™ Server** is dedicated to managing plates, instruments, storage and analysis of Microarray experiments.
- **SwiftArrayStudio™ Data Collection Software** is an integrated web application enabling users to:
  - Manage 1-4 instruments
  - Monitor plate runs remotely by viewing progress on the application dashboard
  - Sample and SwiftArray plate registration
  - View individual plates post image processing
  - Submit service and support tickets using SmartHelp™
- **Thermo Fisher Connect** is a service and support tool that runs in the background on the SwiftArrayStudio™ Server. It sends telemetry, core, and episodic data from each connected instrument. This compiled data and run information can be used for efficient troubleshooting and reduced instrument downtime.

Applications included for GeneTitan users

- **GCC File Uploader Service** sends a notification after the plate has been successfully completed. It then creates duplicates of the CEL and ARR files and uploades them to a designated Microarray Applications folder.
- **Automated Axiom™ (AutoAx)** is alerted that CEL files are available for analysis, then it runs the analysis based on the workflow that was selected for that specific array type.

  Note: Prior to the first GTMC instrument run, set up an analysis configuration and set it as the default workflow for each array to be run. When AutoAx runs and encounters the array, it will automatically run it. No further user input is required.

  Note: AutoAx requires very specific library files. Contact your Thermo Fisher support person for more information.

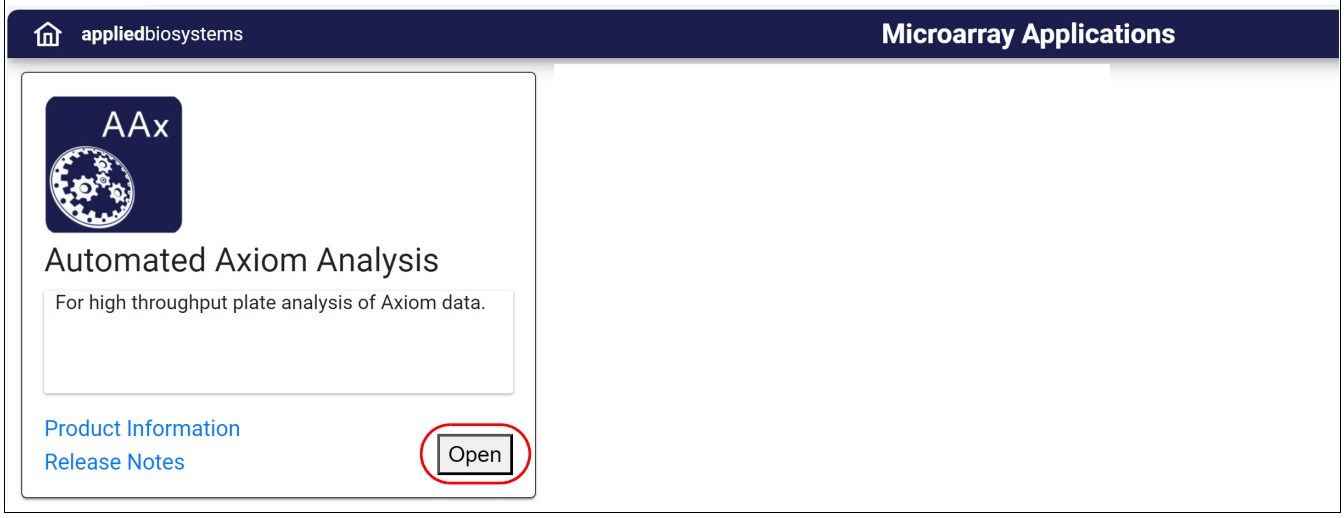
Open AutoAx

IMPORTANT! AutoAx has been validated to be compatible with Chrome. Using a different Internet browser is not recommended, as incompatibility issues may occur.

1. Click on the link provided by your IT team.

   The Microarray Applications web page opens and displays the **Automated Axiom™ Analysis** application card. (Figure 1)

Figure 1 Microarray Applications web page



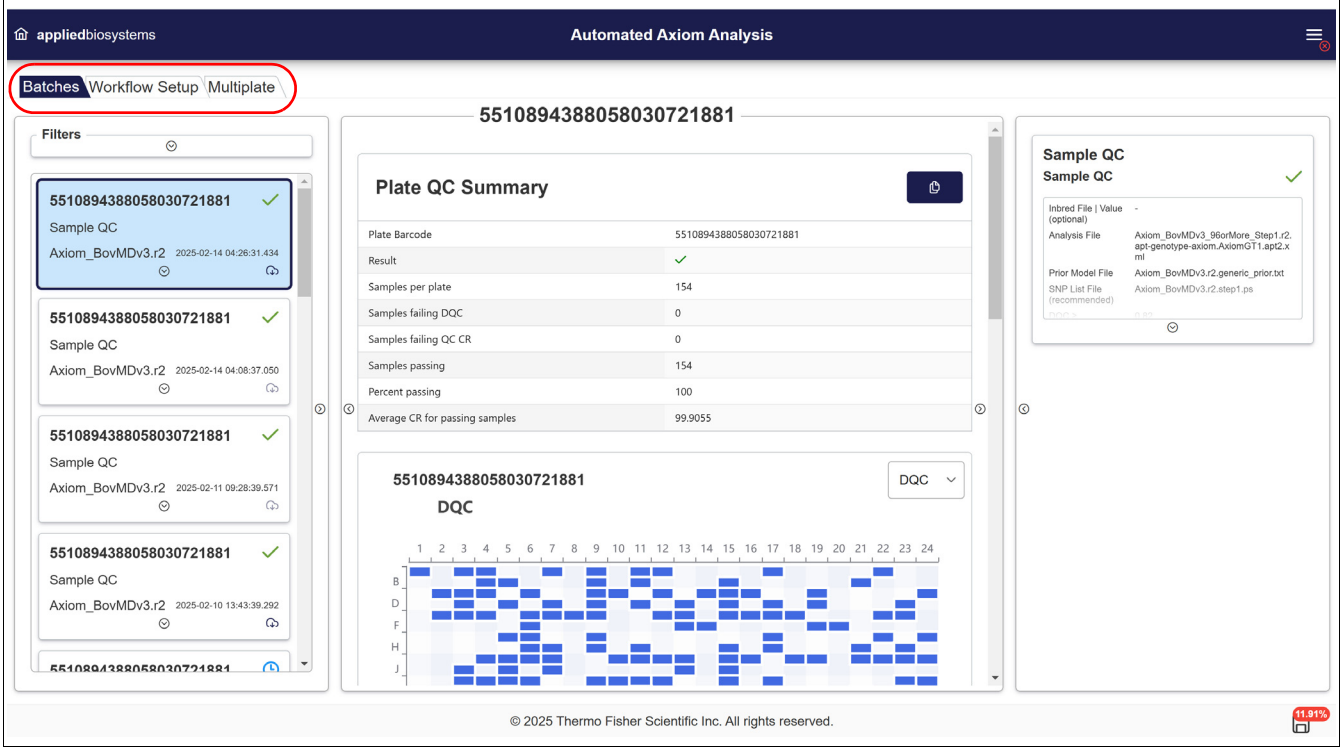
- Click **Product Information** to learn more about the software.
- Click **Release Notes** for information about this new release.

2. Click **Open**.

   The Automated Axiom™ Analysis web page opens (in the same browser window). The **Batches** window tab appears by default. (Figure 2)

Main window

Figure 2 Batches window tab - Populated example



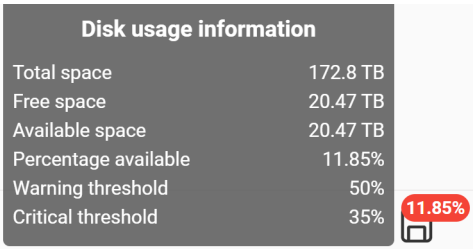
AutoAx consists of three main window tabs. The following chapters (starting with "Batches" on page 13) detail how to navigate and use each window's unique workflows, features, and options.

Monitor disk usage

Disk usage

displays the space currently available on the Server drive.

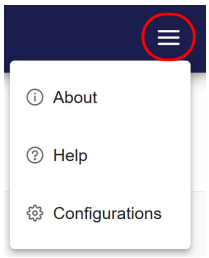
1. To view usage details, place the mouse pointer on the **Disk Usage** icon.



Note: To modify threshold alert settings, see "Disk Usage" on page 11.

AutoAx menu

1. Click on (upper right).



About

1. Click **About**.

   An About box appears.
2. Click on the drop-down arrow to display the multiple sub-applications (and their versions) that make up this release.
3. Click **OK** to close the window.

Help

1. Click **Help**.

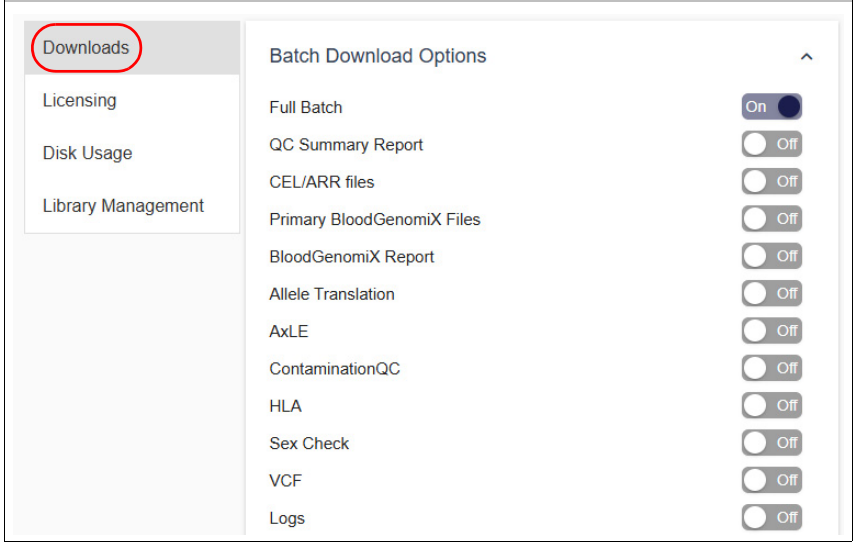
   The Thermo Fisher Scientific *Microarray Analysis Software* web page opens.

Configurations

1. Click **Configurations**.

   The Configurations window opens starting with **Downloads**. (Figure 3)

Figure 3 Configuration window - Downloads



Downloads

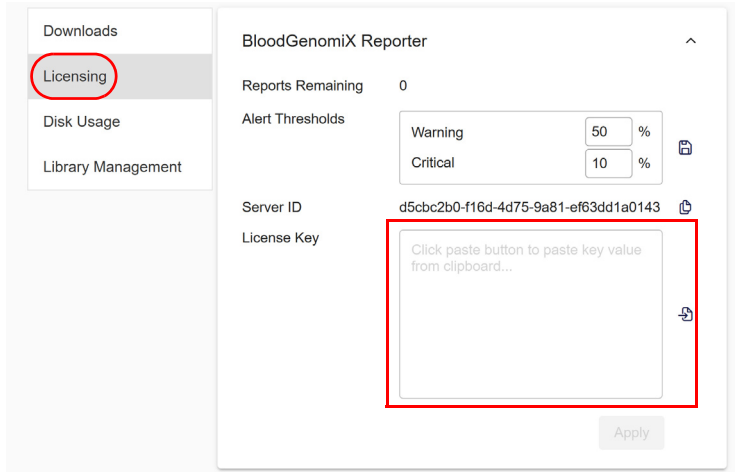
1. At the **Batch Download Options** list, click on a option's toggle to turn it on or off. **Note:** Each toggled option is auto-saved and used for future batch downloads.

   To exit the Configurations window, click the **Batches** tab.

Licensing

- View how many purchased BloodGenomix reports are remaining.
- Use the **Alert Thresholds** fields to modify alert threshold, then click to save.
- To copy the **Server ID** string to the Clipboard, click .
- To paste the License key string, click the License Key's icon. After the **Apply** button is enabled, click on it. The **Reports Remaining** count updates.

Figure 4 Configurations window - Licensing



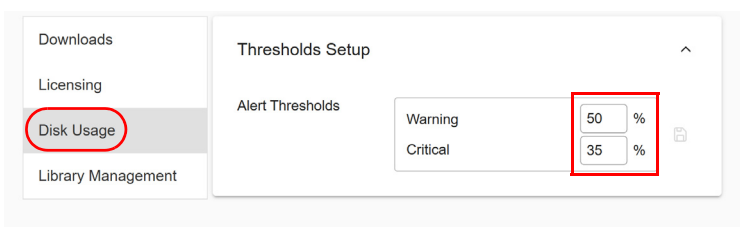
To exit the **Configurations** window, click the **Batches** tab.

Disk Usage

Click **Disk Usage** to view the current alert threshold settings. These settings can be changed.

1. Use the Alert Threshold fields to enter a different alert threshold, then click .

Figure 5 Configurations window - Disk Usage

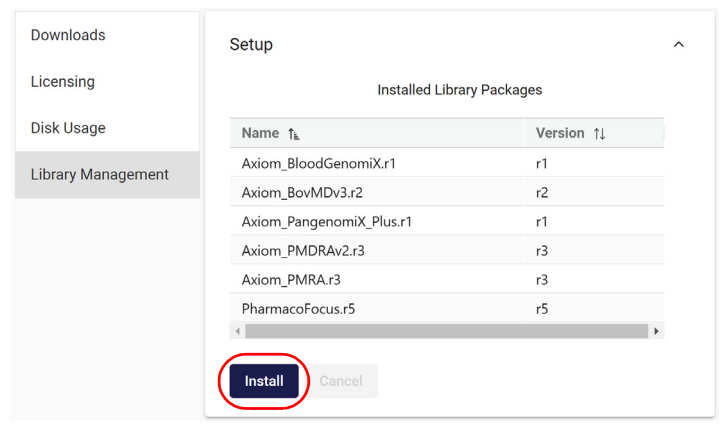


To exit the Configurations window, click the **Batches** tab.

Library Management

1. Click **Library Management** to view, install, or delete a library file.

Figure 6 Configurations window - Library Management



How to install a new library file

1. Click the **Install** button.

   An Explorer window appears.

2. Navigate to the library zip package, click to select it, then click **Open**.

   An uploading progress bar appears.

IMPORTANT! Only one library package can be installed per array.

Note: BEFORE installing the library file package, make sure it contains the **annot.db** file. If it does not, contact the site's Thermo Fisher Field Support representative.

To exit the Configurations window, click the **Batches** tab.

How to delete a library file

1. Place the mouse pointer on the library file to be deleted, then click on the icon.

   A Confirmation message appears.

2. Click **OK**.

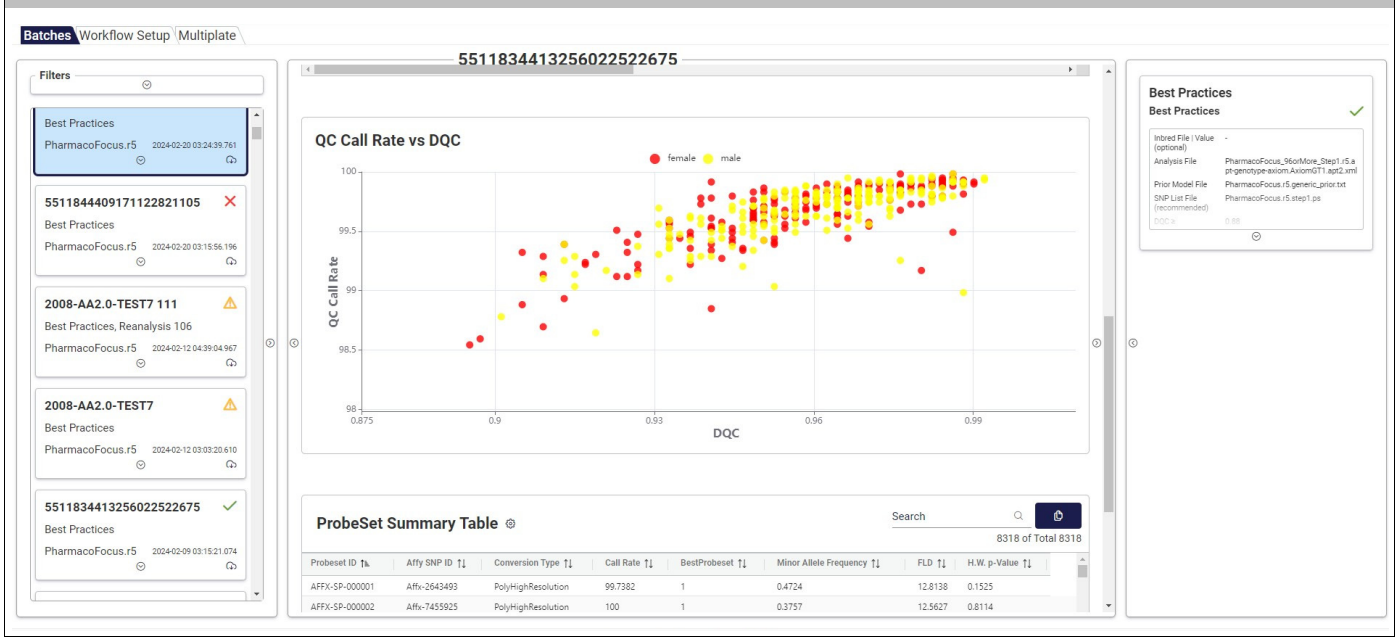
   The library file is deleted.

   To exit the **Configurations** window, click the **Batches** tab.

2 Batches

Use the **Batches** window tab to view the comprehensive results of a successfully completed batch run, as shown in Figure 7.

Figure 7 Batches window tab - Populated example

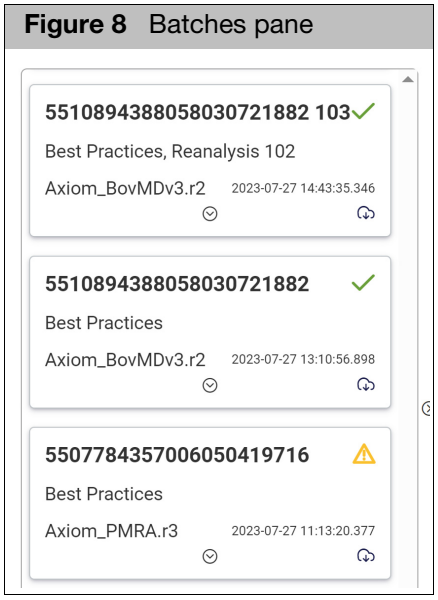


Batches window tab

Batches pane

- Batches are displayed in the far left pane. (Figure 8)
- The card with a blue background represents the batch currently displayed.

Figure 8 Batches pane



- Card symbols:

  = **Completed**

  = **Error**

  = **Warning**

  = **Download**

  = **Running**

  = **Pending**

- Click the batch card's **Download** icon to copy its data to the **Downloads** folder.
- Click each card's **down arrow** button to display additional batch information, including batch ID and GeneTitan™/SwiftArrayStudio™ instrument warnings and error details.
- Click the pane's **right arrow** button to view all batch cards. (Figure 9) Click on the **left arrow** button (far middle right) to return to the default three pane view.

  Note: If the batch is a multiplate batch, the barcode is replaced with a user defined batch name.
- Click on the **Switch to Table View** button to view all cards in a table view. (Figure 10) Click on a column header(s) to sort the data. Click on the **Cloud** icon next to the batch to download it.

Figure 9 Expanded (All) cards view

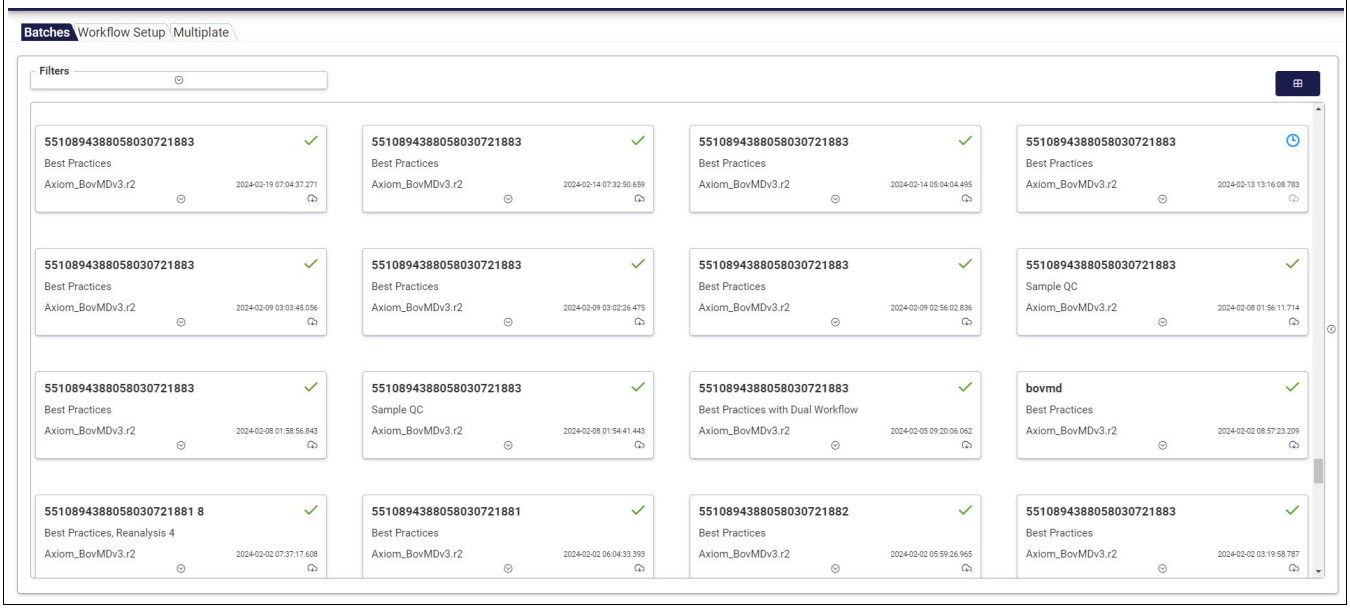
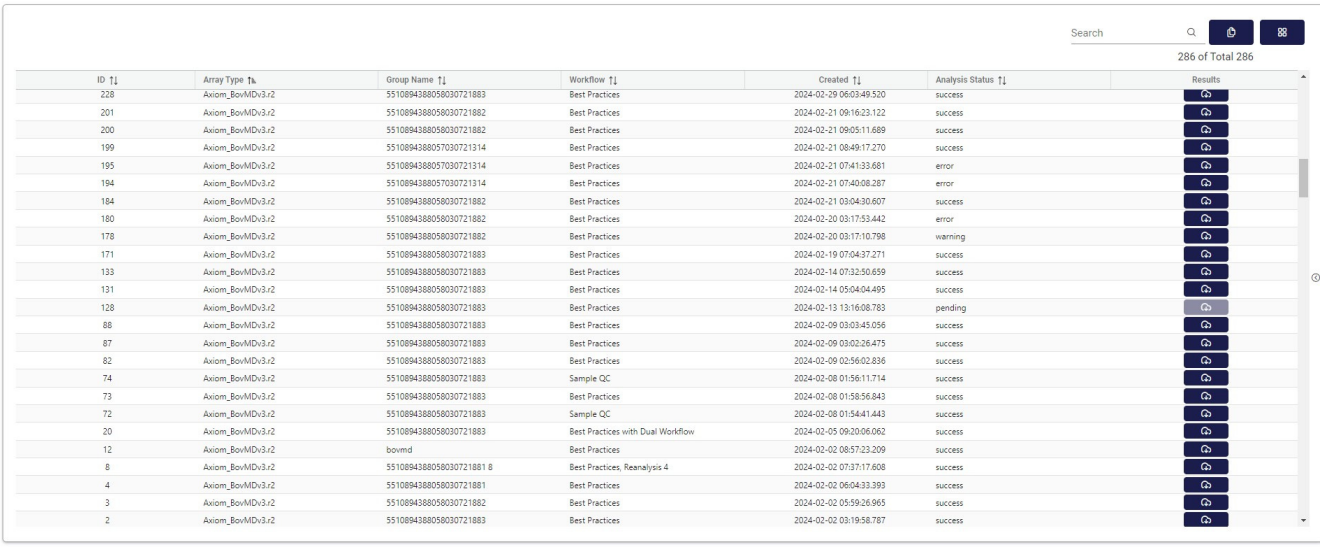


Figure 10 Expanded (All) Table view

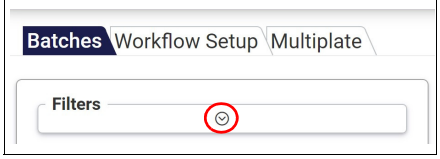


ID	Array Type	Group Name	Workflow	Created	Analysis Status	Results
228	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-29 06:03:49.520	success	
201	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-21 09:16:23.122	success	
200	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-21 09:05:11.689	success	
199	Axiom_BovMDv3.r2	5510894388057030721314	Best Practices	2024-02-21 08:49:17.270	success	
195	Axiom_BovMDv3.r2	5510894388057030721314	Best Practices	2024-02-21 07:41:33.681	error	
194	Axiom_BovMDv3.r2	5510894388057030721314	Best Practices	2024-02-21 07:40:08.287	error	
184	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-21 03:04:30.607	success	
180	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-20 03:17:53.442	error	
178	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-20 03:17:10.798	warning	
171	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-19 07:04:37.271	success	
133	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-14 07:32:50.659	success	
131	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-14 05:04:04.495	success	
128	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-13 13:16:08.783	pending	
88	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-09 03:03:45.056	success	
87	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-09 03:02:26.475	success	
82	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-09 02:56:02.836	success	
74	Axiom_BovMDv3.r2	5510894388058030721883	Sample QC	2024-02-08 01:56:11.714	success	
73	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-08 01:58:56.843	success	
72	Axiom_BovMDv3.r2	5510894388058030721883	Sample QC	2024-02-08 01:54:41.443	success	
20	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices with Dual Workflow	2024-02-05 09:20:06.062	success	
12	Axiom_BovMDv3.r2	bovmd	Best Practices	2024-02-02 08:57:23.209	success	
8	Axiom_BovMDv3.r2	5510894388058030721881 8	Best Practices, Reanalysis 4	2024-02-02 07:37:17.608	success	
4	Axiom_BovMDv3.r2	5510894388058030721881	Best Practices	2024-02-02 06:04:33.393	success	
3	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-02 05:59:26.965	success	
2	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-02 03:19:58.787	success	

Filter batches

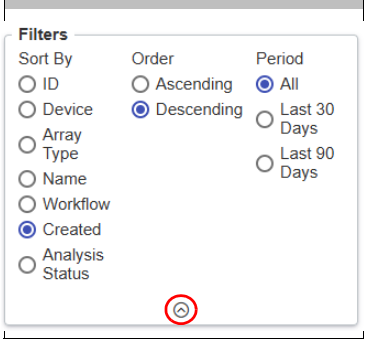
1. At the main **Batches** window tab, click the **down arrow** button under **Filters** (Figure 11) to display the filters.

Figure 11 Filters button



2. Click on the radio button(s) (Figure 12) to customize filtering options, then click the **up arrow** button to hide filter(s).

Figure 12 Filters options



View a selected batch

Plate QC summary table

The **Plate QC Summary** table (Figure 13) contains detailed QC information for each plate that is run in the batch. **Note:** Displayed information varies with each analysis run.

Figure 13 Plate QC Summary table

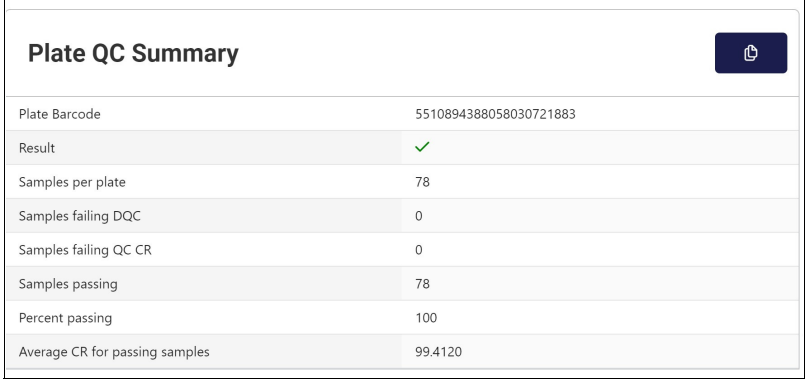


Plate Barcode	5510894388058030721883
Result	✓
Samples per plate	78
Samples failing DQC	0
Samples failing QC CR	0
Samples passing	78
Percent passing	100
Average CR for passing samples	99.4120

The **Plate QC Summary** table columns and its descriptions are detailed in Table 1.

Table 1 Plate QC Summary table

Column	Description
Plate ID	SwiftArrayStudio™ plate identification number that was analyzed in the batch.
Plate Barcode	Barcode of the plate that was analyzed in the batch.
Result	Based on QC thresholds set in analysis configuration template. **Note:** A plate is either Pass or Fail.
Samples per plate	Count of samples in the plate.
Number of Dual Workflow samples	Records the samples that went through the Dual Workflow analysis.
Samples failing DQC	Count of samples with DQC values that were lower than threshold set for Dish QC.
Samples failing QC CR	Count of samples with QC CR values that were lower than threshold set for QC Call Rate.
Samples passing	Count of samples that pass DQC, QC Call Rate and Plate QC.
Percent passing	Percent of samples that pass DQC, QC Call Rate and Plate QC.
Average CR for passing samples	Average call rate for samples that pass QC.
Number of samples failing ContaminationQC *	Count of samples that did not pass ContaminationQC thresholds.
Number of BGX Reports used *	Number of BGX reports used per plate.
Sex Check *	Number of samples with confirmed computed sex (requires user provided annotations within the plate registration file).

* **BloodGenomiX Reporter analyses only.**

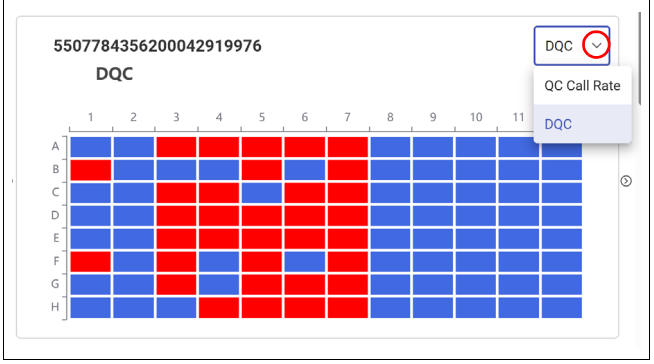
Copy the table

1. Click the button to copy the table.
2. Open Word or an Excel file.
3. Press **Ctrl+v** to paste the table data.

Plots

1. The **DQC** plot appears by default. Click the drop-down (Figure 14) to select a different plot.

Figure 14 Plots drop-down



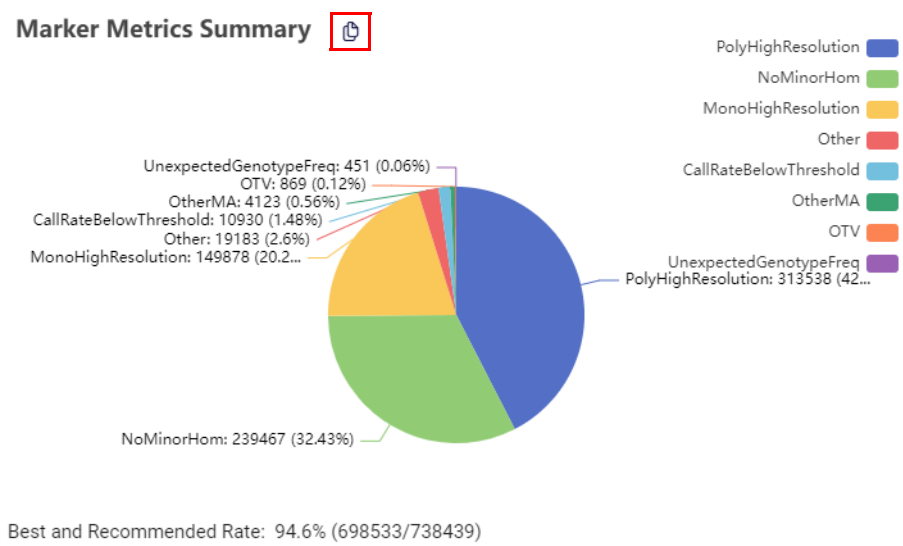
- Each sample value is shown in its well location.
- Wells with failed samples are shown in **red**.
- A white well denotes an empty well or one that was not calculated.
- Place the mouse pointer on a well to view its details.

  Note: Batches that include a Copy Number will display MAPD and WavinessSD plate plots. BloodGenomiX Reporter analyses will display ContaminationQC plate plots.

Marker metrics summary

1. Hover over the **Marker Metrics Summary** ⓘ button (Figure 15) to display its pop up window.

Figure 15 Marker Metrics Summary

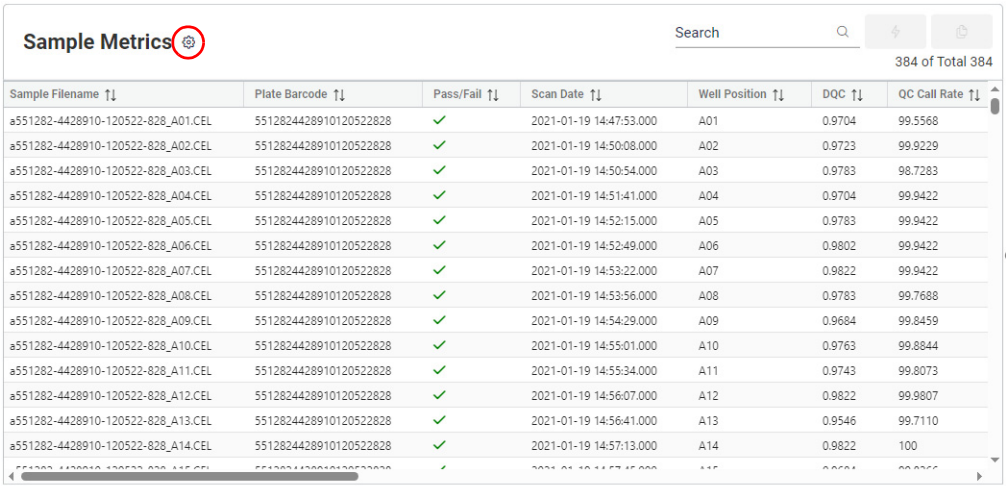


2. Click outside the pop up window to hide it.
3. Optional: To view the Marker Metrics Summary's graphic results as a conventional table, click its icon, then paste (Ctrl v) in Excel or Notepad.

Sample metrics table

The **Sample Metrics** table (Figure 16) and its default columns and descriptions are detailed in Table 2.

Figure 16 Sample Metrics table



Sample Filename	Plate Barcode	Pass/Fail	Scan Date	Well Position	DQC	QC Call Rate
a551282-4428910-120522-828_A01.CEL	5512824428910120522828	✓	2021-01-19 14:47:53.000	A01	0.9704	99.5568
a551282-4428910-120522-828_A02.CEL	5512824428910120522828	✓	2021-01-19 14:50:08.000	A02	0.9723	99.9229
a551282-4428910-120522-828_A03.CEL	5512824428910120522828	✓	2021-01-19 14:50:54.000	A03	0.9783	98.7283
a551282-4428910-120522-828_A04.CEL	5512824428910120522828	✓	2021-01-19 14:51:41.000	A04	0.9704	99.9422
a551282-4428910-120522-828_A05.CEL	5512824428910120522828	✓	2021-01-19 14:52:15.000	A05	0.9783	99.9422
a551282-4428910-120522-828_A06.CEL	5512824428910120522828	✓	2021-01-19 14:52:49.000	A06	0.9802	99.9422
a551282-4428910-120522-828_A07.CEL	5512824428910120522828	✓	2021-01-19 14:53:22.000	A07	0.9822	99.9422
a551282-4428910-120522-828_A08.CEL	5512824428910120522828	✓	2021-01-19 14:53:56.000	A08	0.9783	99.7688
a551282-4428910-120522-828_A09.CEL	5512824428910120522828	✓	2021-01-19 14:54:29.000	A09	0.9684	99.8459
a551282-4428910-120522-828_A10.CEL	5512824428910120522828	✓	2021-01-19 14:55:01.000	A10	0.9763	99.8844
a551282-4428910-120522-828_A11.CEL	5512824428910120522828	✓	2021-01-19 14:55:34.000	A11	0.9743	99.8073
a551282-4428910-120522-828_A12.CEL	5512824428910120522828	✓	2021-01-19 14:56:07.000	A12	0.9822	99.9807
a551282-4428910-120522-828_A13.CEL	5512824428910120522828	✓	2021-01-19 14:56:41.000	A13	0.9546	99.7110
a551282-4428910-120522-828_A14.CEL	5512824428910120522828	✓	2021-01-19 14:57:13.000	A14	0.9822	100

Table 2 Sample Metrics table

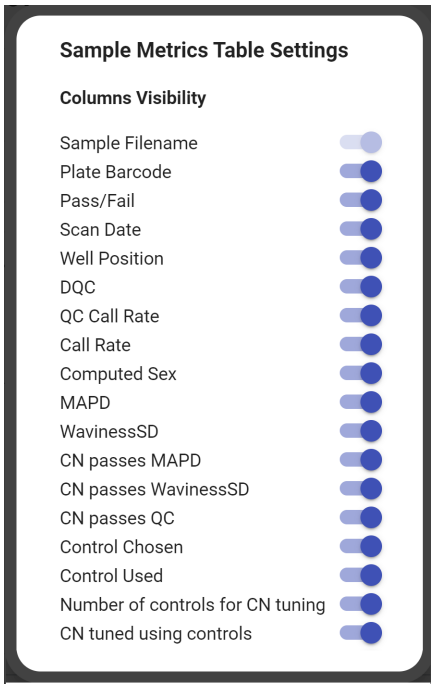
Column	Description
Sample Filename	Name of CEL file.
Pass/File	Sample quality control status. ✓ = The Sample QC step deems it meets the minimum thresholds for DQC, QC call_rate, and average call rate for passing samples. ✗ = The sample did not pass Sample QC.
Plate Barcode	Barcode of the array plate.
Scan Date	Date the array was scanned (year-month-day).
Well Position	Location of sample in the array plate e.g A1, B1, etc.
DQC	Dish QC of sample. **Note:** Dish QC measures the amount of overlap between two homozygous peaks created by non-polymorphic probes. A DQC of 1 equals no overlap (Good), while a DQC of 0 denotes a complete overlap (Bad).
QC Call Rate (QCCR)	QC Call Rate of a sample. **Note:** The QC call rate is computed over a representative set (usually 20k) of well-behaved probesets. A Low QC call rate over well-behaved probesets suggest sample quality issues.
Call Rate	Call rate of a sample. **Note:** The Call rate is the percentage of samples assigned a genotype (other than a No Call).
Computed Sex	Computed sex of the sample.
Dual Workflow *	Records the samples that went through the Dual Workflow analysis. **Note:** A [●] icon appears in the Dual Workflow column to show the sample met the Dual Workflow criteria and was analyzed. * Best Practices with Dual workflow only.

Note: Columns in the Sample Metrics table are Workflow specific.

Sample Metrics Table Settings

1. Click the **Sample Metrics** icon.
2. At the pop up window (Figure 17), click on the toggles to show or hide the columns listed. **Note:** Columns Visibility preferences must be set for each workflow. **Sample Filename** cannot be toggled off.

Figure 17 Sample Metrics Table Settings window



3. Click outside the pop up window to close it and save the toggled selections.

Filter a Sample Metrics table

1. Enter a word or value in the **Search** field (upper right).

   If a match is found, only entered search results are displayed.
2. Optional: Return the table to its original unfiltered state by highlighting and deleting the **Search** entry.

   The Sample Metrics table now returns to its default ALL records view.

Sort a Sample Metrics table

1. Click on a column header to sort it either in ascending or descending order.

   A small arrow appears on the header indicting the sorting order.
2. Click the header again to change the sorting order.

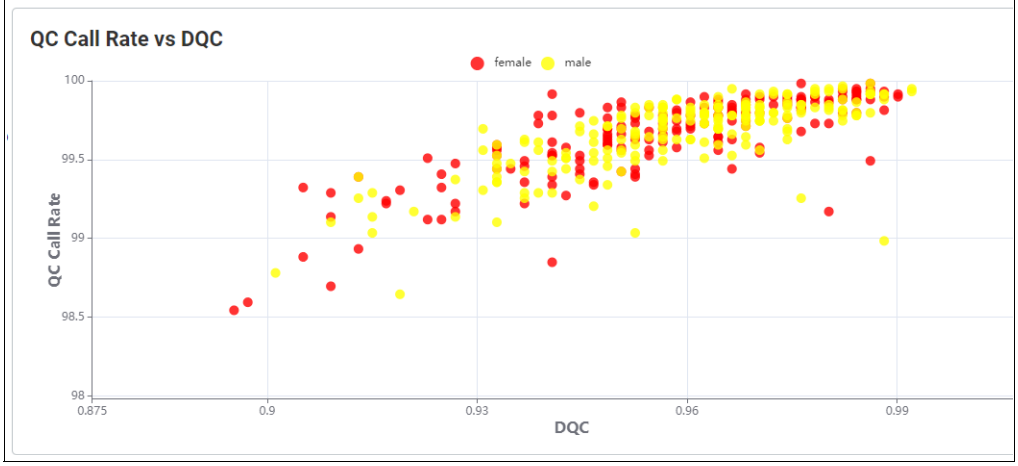
Copy a table

1. Click to select the row(s) you want to copy, then click the button.
2. Open Word or an Excel file.
3. Press **Ctrl v** to paste the table data.

Scatter plot

A **QC Call Rate** vs **DQC** Scatter plot appears below the Sample Metrics table. Plots are color-keyed by gender, as shown in Figure 18.

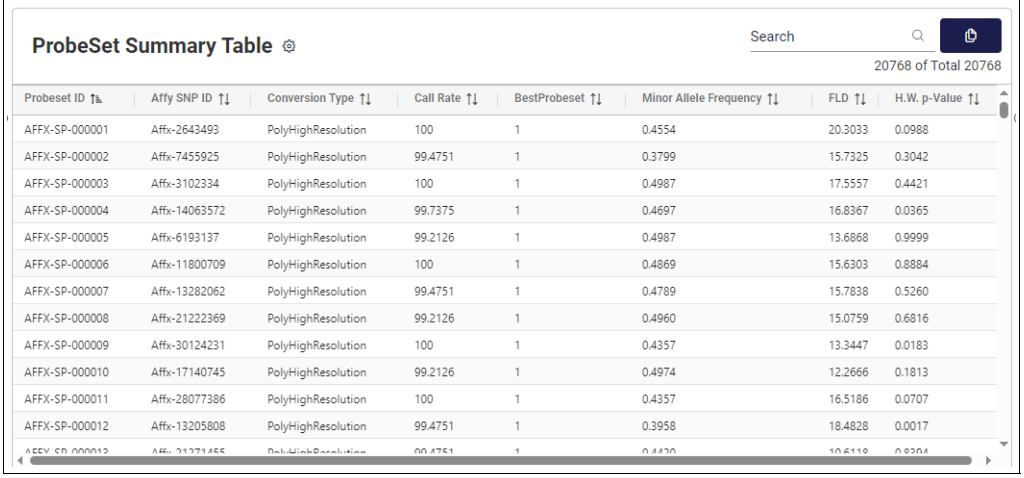
Figure 18 Scatter plot - QC Call Rate vs DQC



ProbeSet Summary table

The ProbeSet Summary table's (Figure 19) columns and descriptions are detailed in Table 3.

Figure 19 ProbeSet Summary Table



ProbeSet Summary Table

Search

20768 of Total 20768

Probeset ID	Affy SNP ID	Conversion Type	Call Rate	BestProbeset	Minor Allele Frequency	FLD	H.W. p-Value
AFFX-SP-000001	Affx-2643493	PolyHighResolution	100	1	0.4554	20.3033	0.0988
AFFX-SP-000002	Affx-7455925	PolyHighResolution	99.4751	1	0.3799	15.7325	0.3042
AFFX-SP-000003	Affx-3102334	PolyHighResolution	100	1	0.4987	17.5557	0.4421
AFFX-SP-000004	Affx-14063572	PolyHighResolution	99.7375	1	0.4697	16.8367	0.0365
AFFX-SP-000005	Affx-6193137	PolyHighResolution	99.2126	1	0.4987	13.6868	0.9999
AFFX-SP-000006	Affx-11800709	PolyHighResolution	100	1	0.4869	15.6303	0.8884
AFFX-SP-000007	Affx-13282062	PolyHighResolution	99.4751	1	0.4789	15.7838	0.5260
AFFX-SP-000008	Affx-21222369	PolyHighResolution	99.2126	1	0.4960	15.0759	0.6816
AFFX-SP-000009	Affx-30124231	PolyHighResolution	100	1	0.4357	13.3447	0.0183
AFFX-SP-000010	Affx-17140745	PolyHighResolution	99.2126	1	0.4974	12.2666	0.1813
AFFX-SP-000011	Affx-28077386	PolyHighResolution	100	1	0.4357	16.5186	0.0707
AFFX-SP-000012	Affx-13205808	PolyHighResolution	99.4751	1	0.3958	18.4828	0.0017

1. Click the adjacent icon to display the ProbeSet Summary Table Settings window. (Figure 20) Click on a column's toggle to turn its visibility on or off. **ProbeSet ID** cannot be toggled off.

   Note: Columns Visibility preferences must be set for each workflow.

Figure 20 ProbeSet Summary Table Settings window

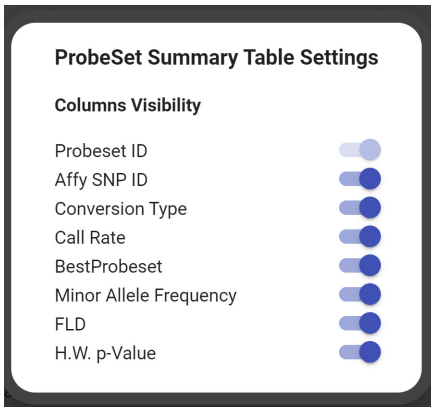


Table 3 ProbeSet Summary Table

Column	Description
ProbeSet ID	The Axiom unique identifier for the set of probes used to detect a particular Single Nucleotide Polymorphism (SNP probe sets only).
Affy SNP ID	The Axiom unique identifier for the set of probes used to detect a particular Single Nucleotide Polymorphism (SNP).
Conversion Type	ProbeSet classification
Call Rate (CR)	The percentage of samples with a genotype call other than "No Call" for the SNP.
BestProbeSet	BestProbeSet flag is available when multiple probesets are mapped to the same SNP (Affy-SNP-ID) by a ps2snp file. A probeset is selected based on the priority order of the conversion types. BestProbeSet flag is 1 when it is the best or only probeset for a SNP. Otherwise the flag is 0.
Minor Allele Frequency	The allele frequency for the A allele is calculated as: $P_A = \frac{(\#\ AA\ Calls + 0.5 * AB\ Calls)}{Total\ \#\ Calls}$ Where the Total # Calls does not include the No Calls. The B allele frequency is . $P_B = 1 - P_A$ The minor allele frequency is the Min(P_A, P_B).

Table 3 ProbeSet Summary Table

Column	Description
FLD	Fisher's Linear Discriminant (FLD) is a measure of the cluster quality of a probeset. High-quality probeset clusters have well-separated centers, and the clusters are narrow. FLD is measured from the SNP posteriors produced during genotyping. FLD is undefined if either the heterozygous or one of the homozygous clusters is empty.
H.W. p-Value	Hardy Weinberg p-value is a measure of the significance of the discrepancy between the observed ratio or heterozygote calls in a population and the ratio expected if the population was in Hardy Weinberg equilibrium. There are two statistical tests used for HWE. When AA, AB, and BB counts are all >=10, a Chi-squared test is used. When one or more of the AA, AB, and BB counts are <10, an Exact test is used. An Exact test means that the p-value is calculated exactly and not approximated from a population distribution.

Filter a ProbeSet Summary Table

1. Enter a word or value in the **Search** field (upper right).

   If a match is found, only the entered search results are displayed.

   To **Clear** (return the table to its original state), delete the search entry.

Sort a ProbeSet Summary Table

1. Click on a column header to sort it either in ascending or descending order.

   A small arrow appears on the header indicting the new sorting order.
2. Click the header again to change the sorting order.

Copy a table

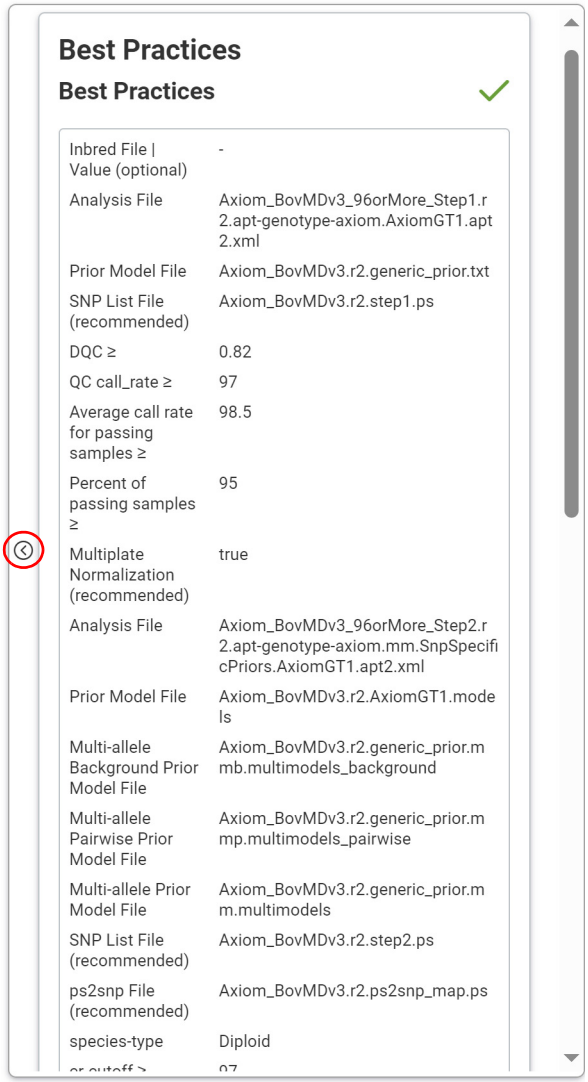
1. Click the button to copy the current selection shown on the table.
2. Open Word or an Excel file.
3. Press **Ctrl v** to paste the table data.

Batch analysis parameters

The batch analysis and extension parameters can be found on the far right pane of the Batches window tab. (Figure 21)

1. Click the **down arrow** button to view the batch's analysis or extension details.
2. Use the pane's scroll bar to view all the reported analyses or click the **left arrow** button to view it in a full window.
3. Click the **right arrow** button to return to the pane's (far right pane) default view.

Figure 21 Batch analysis parameters



Best Practices

Best Practices

Inbred File | Value (optional)	-
Analysis File	Axiom_BovMDv3_96orMore_Step1.r2.apt-genotype-axiom.AxiomGT1.apt2.xml
Prior Model File	Axiom_BovMDv3.r2.generic_prior.txt
SNP List File (recommended)	Axiom_BovMDv3.r2.step1.ps
DQC ≥	0.82
QC call_rate ≥	97
Average call rate for passing samples ≥	98.5
Percent of passing samples ≥	95
Multiplate Normalization (recommended)	true
Analysis File	Axiom_BovMDv3_96orMore_Step2.r2.apt-genotype-axiom.mm.SnpSpecificPriors.AxiomGT1.apt2.xml
Prior Model File	Axiom_BovMDv3.r2.AxiomGT1.models
Multi-allele Background Prior Model File	Axiom_BovMDv3.r2.generic_prior.mmb.multimodels_background
Multi-allele Pairwise Prior Model File	Axiom_BovMDv3.r2.generic_prior.mmp.multimodels_pairwise
Multi-allele Prior Model File	Axiom_BovMDv3.r2.generic_prior.mm.multimodels
SNP List File (recommended)	Axiom_BovMDv3.r2.step2.ps
ps2snp File (recommended)	Axiom_BovMDv3.r2.ps2snp_map.ps
species-type	Diploid
cr cutoff ≥	97

3 Workflow Setup

The **Workflow Setup** window tab (Figure 22) contains three different customizable workflows and supported extensions for the Best Practices workflows.

- "Sample QC"
- "Best Practices" on page 30
- "Best Practices with Dual Workflow (Agbio Arrays only)" on page 35

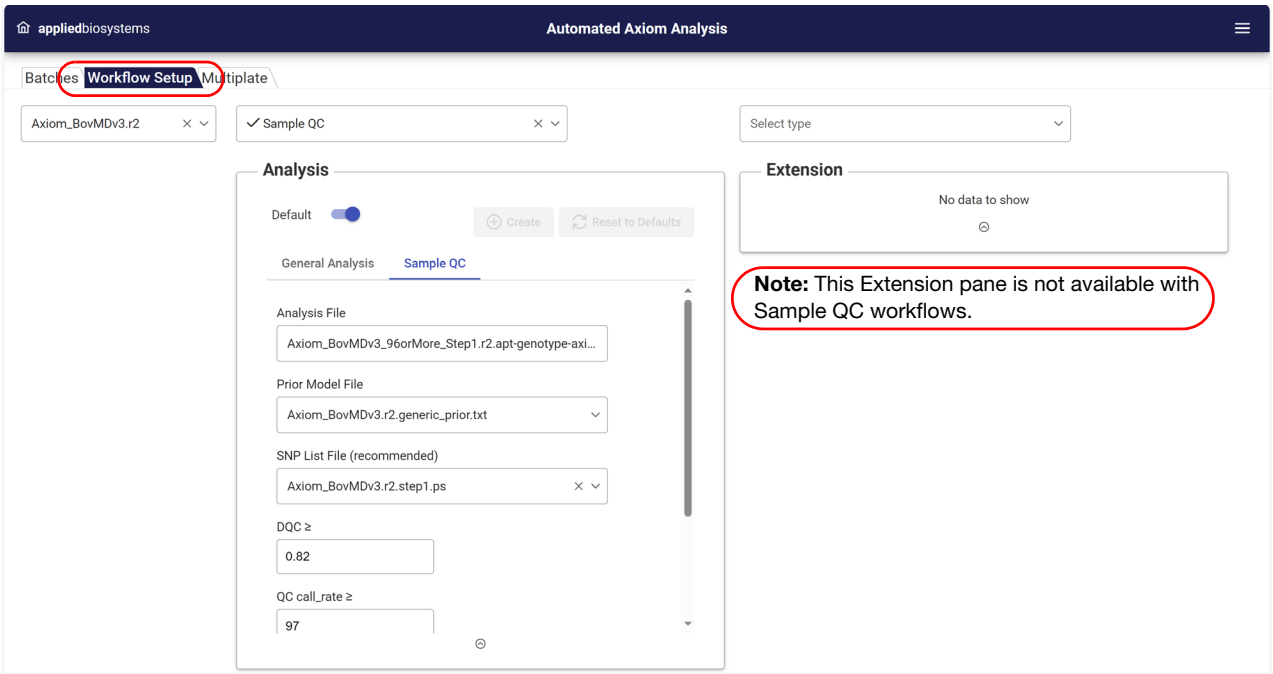
Note: The *BloodGenomiX Reporter* and *BloodGenomiX Reporter XY* workflows are enabled for Blood Transfusion arrays. For more information, see Appendix A.

Sample QC

1. Click the **Workflow Setup** tab.

   The **Workflow Setup** page appears. (Figure 22)

Figure 22 Workflow Setup window tab - Populated example



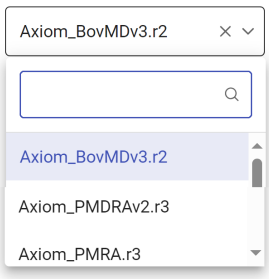
Customize an analysis

IMPORTANT! **BEFORE** changing any analysis setting, refer to the *Axiom Genotyping Data Solutions Guide* (MANN0018363) for analysis setting values and details.

Select an array

1. Click the **Array Type** drop-down (Figure 23) to select the array you want to use. Use the search field to locate a different array.

Figure 23 Array Type



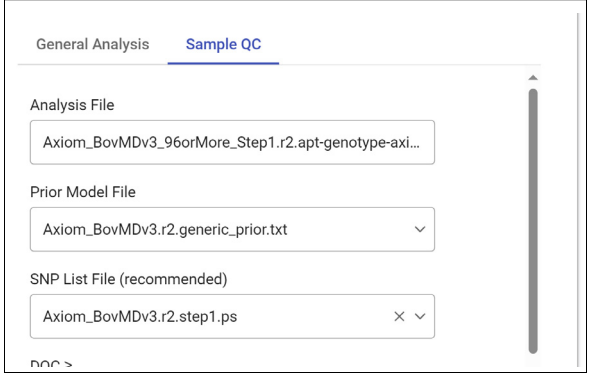
Note: The Workflow drop-down displays **Sample QC** by default. (Figure 24)

Figure 24 Workflow drop-down

Analysis pane

The Analysis pane features a **Sample QC** tab and **General Analysis** tab that features **Inbred File** options.

Figure 25 Analysis options



Note: To maintain all factory default settings, make sure the **Default** toggle is on (Figure 26)

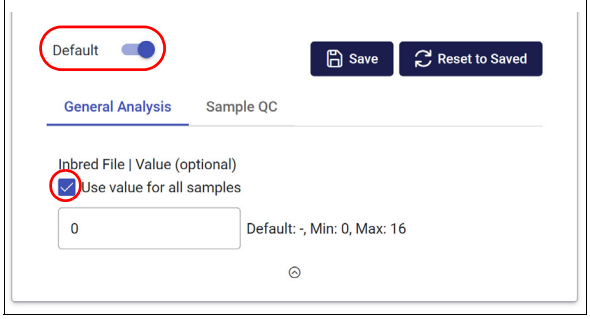
- **Analysis File**: Displays the array you selected.
- **Prior Model File**: Displays the default *Prior Model File* associated with the selected array. Click the drop-down to select a different file or add a new file.
- **SNP List File (recommended)**: Displays the default *SNP List File* associated with the selected array. Click the drop-down to select a different file or add a new file.
- **DQC ≥**: Displays the default *DQC* value associated with the selected array. To enter a new value, click inside the text box.
- **QC call_rate ≥**: Scroll down to display. Displays the default *QC_call_rate value* associated with the selected array. To enter a new value, click inside the text box.
- **Average call rate for passing samples ≥**: Scroll down to display. Displays the default *Average call rate for passing samples* value associated with the selected array. To enter a new value, click inside the text box.
- **Percent of passing samples ≥**: Scroll down to display. Displays the *Percent of passing samples* value associated with the selected array. To enter a new value, click inside the text box.
- To add a file to one of the above entries, see "Add a file not listed" on page 39.
- To change a value, see "Enter a new value" on page 40.

General Analysis tab options (Optional)

Inbred File value for all samples

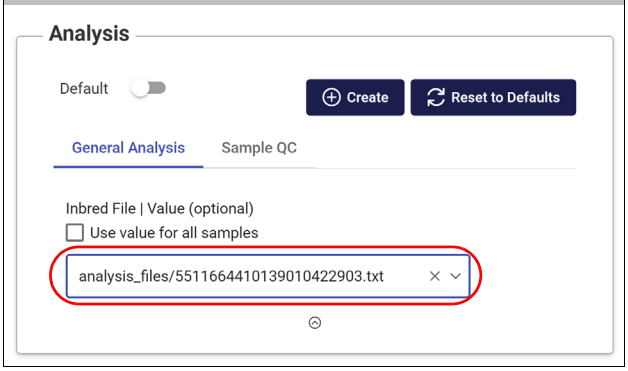
1. Click the check box, then enter a value. (Figure 26)

Figure 26 General Analysis pane - Inbred File



To add an Inbred file not listed in the drop-down (Figure 27), click **Add File**, then see "Add a file not listed" on page 39 for detailed instructions.

Figure 27 General Analysis options



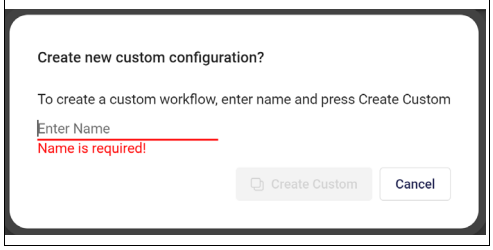
Save a custom configuration

1. Click **Create**.

   Note: If modifying an existing custom workflow, click **Save**.

   After clicking **Create**, a message window appears. (Figure 28)

Figure 28 Message window



2. Enter a workflow name, then click **Create Custom.**

   The new value is displayed.

Save an existing custom configuration

Once you modify an existing custom configuration, the **Save** button is enabled.

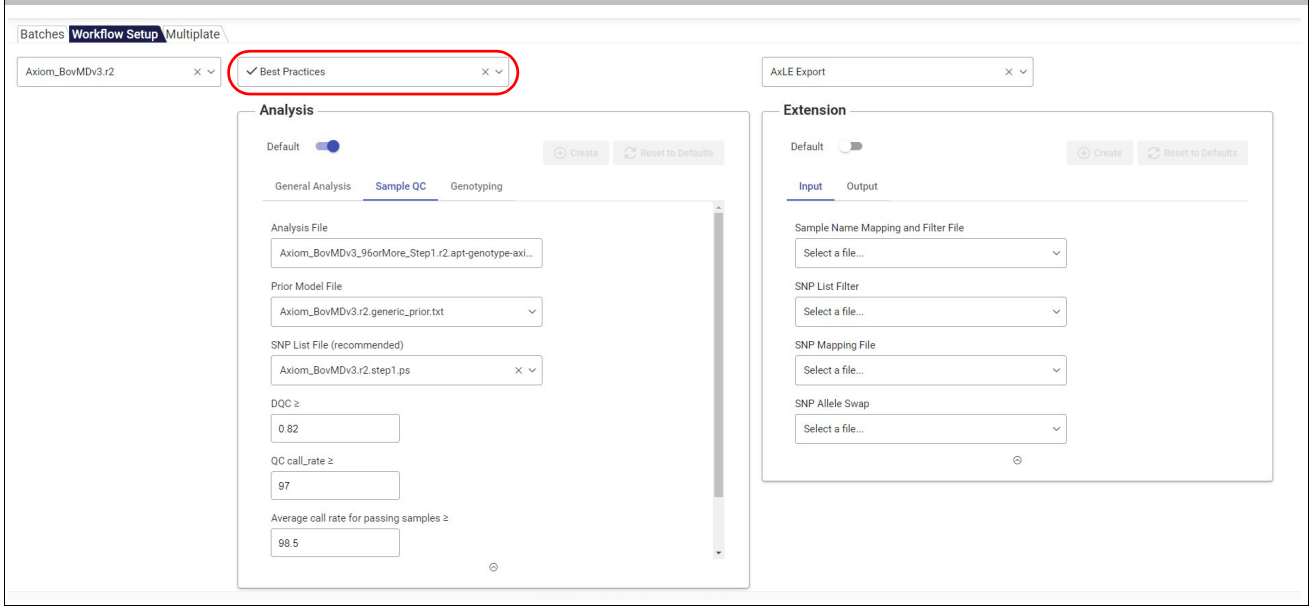
1. Click **Save**.

   The custom configuration is now saved.

Best Practices

1. From the **Workflow Setup** tab, click the **Workflow** drop-down, then select **Best Practices**. (Figure 29)

Figure 29 Workflow Setup window tab - Populated example



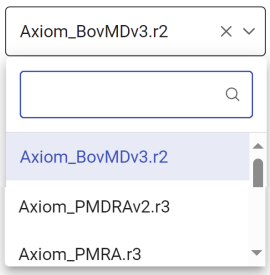
Customize an analysis

IMPORTANT! **BEFORE** changing any analysis setting, refer to the *Axiom Genotyping Data Solutions Guide* (MANN0018363) for analysis setting values and details.

Select an array

1. Click the **Array Type** drop-down (Figure 23) to select the array you want to use. Use the search field to locate a different array.

Figure 30 Array Type



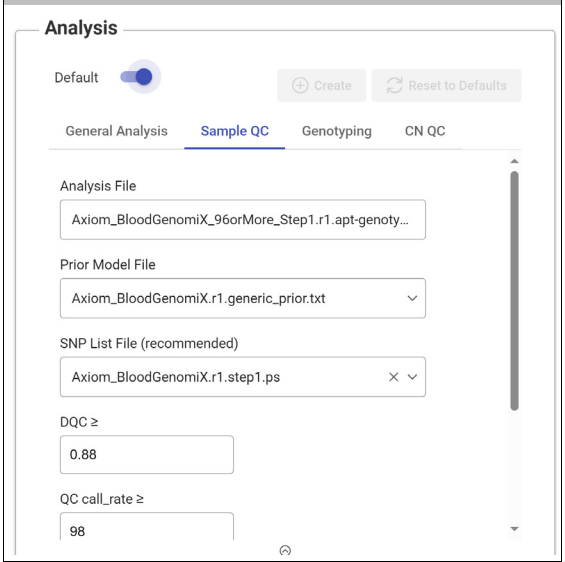
2. Click the Workflow drop-down to select **Best Practices.**

Analysis pane

The Analysis pane features **Sample QC**, **Genotyping**, and **CN QC** tabs. The **General Analysis** tab features **Inbred File** options.

Note: Make sure the **Default** toggle is on (Figure 31) if you want to keep the factory default settings and you are not going to modify them.

Figure 31 Analysis options



Sample QC options

- **Analysis File**: Displays the array you selected.
- **Prior Model File**: Displays the default *Prior Model File* associated with the selected array. Click the drop-down to select a different file or add a new file.
- **SNP List File (recommended)**: Displays the default *SNP List File* associated with the selected array. Click the drop-down to select a different file or add a new file.
- **DQC ≥**: Displays the default *DQC* value associated with the selected array. To enter a new value, click inside the text box.
- **QC call_rate ≥**: Scroll down to display. Displays the default *QC_call_rate value* associated with the selected array. To enter a new value, click inside the text box.
- **Average call rate for passing samples ≥**: Scroll down to display. Displays the default *Average call rate for passing samples* value associated with the selected array. To enter a new value, click inside the text box.
- **Percent of passing samples ≥**: Scroll down to display. Displays the *Percent of passing samples* value associated with the selected array. To enter a new value, click inside the text box.
- To add a file to one of the above entries, see "Add a file not listed" on page 39.
- To change a value, see "Enter a new value" on page 40.

Genotyping options

1. Click the **Genotyping** tab to display the options listed below:

- **Multiplate Normalization (recommended)** becomes enabled for batches using more than one plate.
- **CN Reference Model File**: Contains reference information for CN analysis step.
- **CN Priors File**: Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
- **Analysis File**: This field displays the array you selected in the Array Type drop-down.
- **Prior Model File**: This field displays the default Prior Model File associated with the selected array. Click the drop-down to select a different file or add a new file.
- **SNP List File (recommended)**: This field displays the default SNP List File associated with the selected array. Click the drop-down to select a different file or add a new file.
- **ps2snp File**: If multiple probeset designs exist on the array for a given SNP (for example, one forward and one reverse strand design), then the ps2snp file is used by the SNP classification step to identify the best performing probeset for the SNP.
- **Genotype Frequency File**: If the library package supports a check for unexpectedly high call frequency for specific genotypes, this optional file specifies the maximum expected frequency for reviewed genotypes.
- **species-type**: Species type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.

- **cr-cutoff**: Minimum acceptable call rate.
- **fld-cutoff**: For autosomal probesets, minimum acceptable FLD value for cluster separation.
- **het-so-cutoff**: Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.
- **hom-ro1-cutoff**: Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 1 genotype cluster.
- **hom-ro2-cutoff**: Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.
- **hom-ro-3-cutoff**: Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.
- **recommended**: ProbeSets having ConversionTypes in this recommended set will be included in the recommended probeset list.

CN QC options

1. Click the **CN QC** tab to display the options listed below:

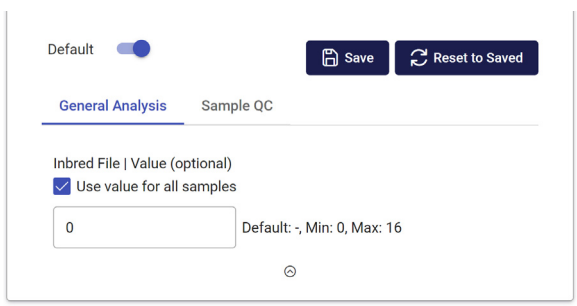
- **MAPD**: For arrays that support copy number (CN) analysis, the Median Absolute Pairwise Difference of log2ratio signals of adjacent copy number (CN) probesets must be less than this value to make CN calls.
- **Waviness SD**: For arrays that support copy number (CN) analysis, the Waviness Standard Deviation of log2 ratio signals of copy number (CN) probesets must be less than this value to make CN calls.

General Analysis options (Optional)

Inbred File value for all samples:

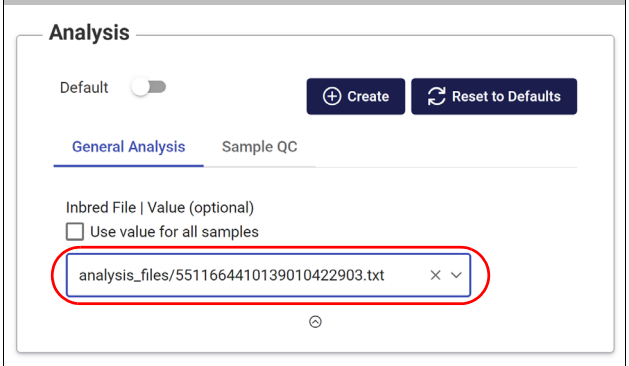
1. Click the check box, then enter a value. (Figure 32)

Figure 32 General Analysis pane - Inbred File



To add an Inbred file not listed in the drop-down (Figure 33), click **Add File**, then see "Add a file not listed" on page 39 for detailed instructions.

Figure 33 General Analysis options



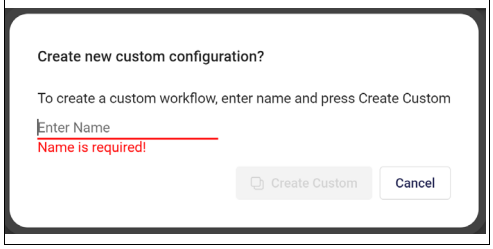
Save a custom configuration

1. Click **Create**.

   Note: If modifying an existing custom workflow, click **Save**.

   After clicking **Create**, a message window appears. (Figure 34)

Figure 34 Message window



2. Enter a workflow name, then click **Create Custom.**

   The new value is displayed.

Save an existing custom configuration

Once an existing custom configuration has been modified, the **Save** button is enabled.

1. Click **Save**.

   The custom configuration is now saved.

Extension pane

The **Extension** pane (right) manages additional features and settings available for export or analysis within the Best Practices Workflow.

Extensions are enabled when available, as they vary depending on the array design. **Note:** Not every extension is available for every array design.

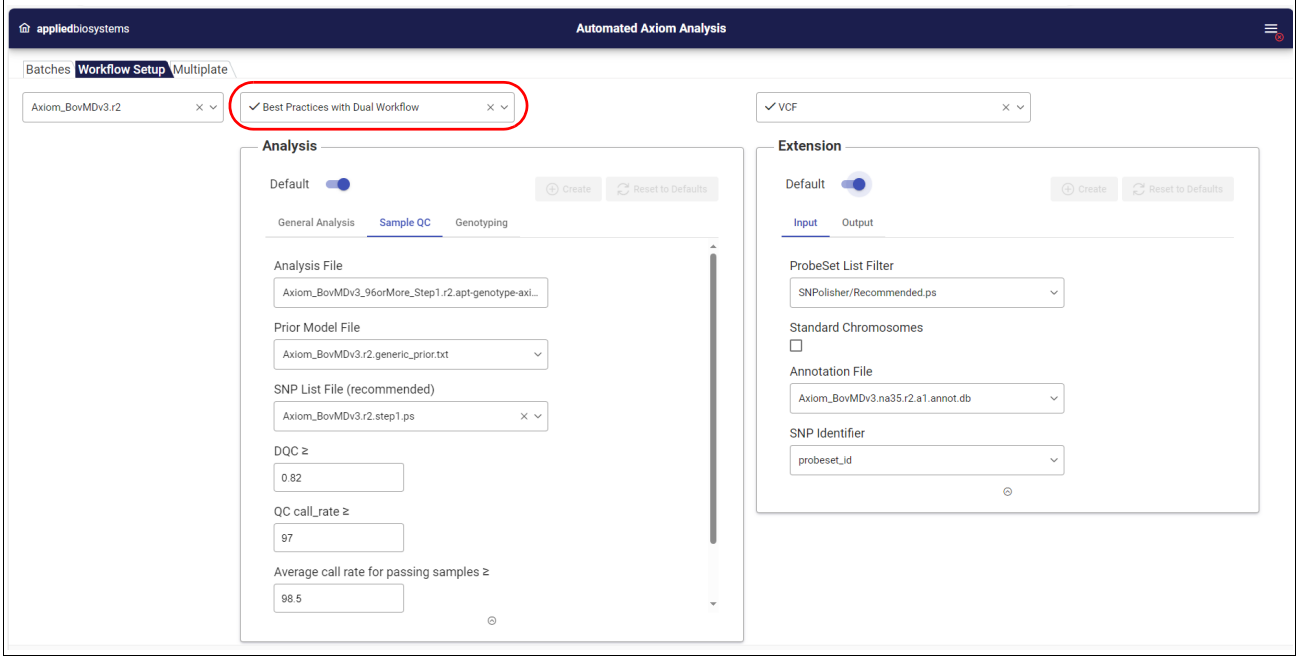
To use the Extension pane, see "How to use the extension pane" on page 65.

Best Practices with Dual Workflow (Agbio Arrays only)

For selected arrays, Best Practices with Dual Workflow is an additional analysis option. For samples that pass DQC and do not make the QCCR, Automated Axiom™ will re-analyze those samples within the same batch. For more information, see the *Axiom Genotyping Data Solutions Guide* (MAN0018363).

1. From the **Workflow Setup** tab, click the **Workflow** drop-down, then select **Best Practices with Dual Workflow**. (Figure 35)

Figure 35 Workflow Setup window tab - Populated example



Customize an analysis

IMPORTANT! **BEFORE** changing any analysis setting, refer to the *Axiom Genotyping Data Solutions Guide* (MANN0018363) for analysis setting values and details.

Select an array

1. Click the **Array Type** drop-down (Figure 36) to select the array you want to use. Use the search field to locate a different array.

Figure 36 Array Type

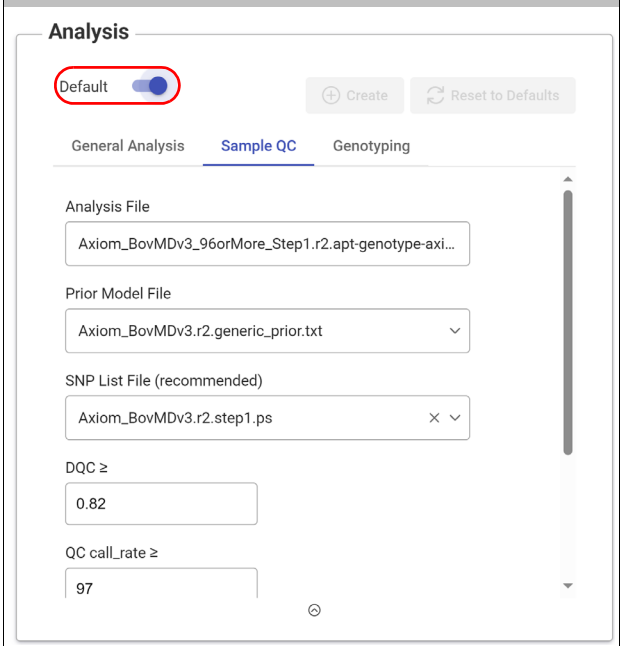
2. Click the Workflow drop-down to select **Best Practices with Dual Workflow.**

Analysis pane

The Analysis pane features **Sample QC**, **Genotyping**, and a **General Analysis** tab features **Inbred File** options.

Note: Make sure the **Default** toggle is on (Figure 37) if you want to keep the factory default settings and you are not going to modify them.

Figure 37 Analysis options



Sample QC options

- **Analysis File**: Displays the array you selected.
- **Prior Model File**: Displays the default *Prior Model File* associated with the selected array. Click the drop-down to select a different file or add a new file.
- **SNP List File (recommended)**: Displays the default *SNP List File* associated with the selected array. Click the drop-down to select a different file or add a new file.
- **DQC ≥**: Displays the default *DQC* value associated with the selected array. To enter a new value, click inside the text box.
- **QC call_rate ≥**: Scroll down to display. Displays the default *QC_call_rate value* associated with the selected array. To enter a new value, click inside the text box.
- **Average call rate for passing samples ≥**: Scroll down to display. Displays the default *Average call rate for passing samples* value associated with the selected array. To enter a new value, click inside the text box.
- **Percent of passing samples ≥**: Scroll down to display. Displays the *Percent of passing samples* value associated with the selected array. To enter a new value, click inside the text box.
- To add a file to one of the above entries, see "Add a file not listed" on page 39.
- To change a value, see "Enter a new value" on page 40.

Genotyping options

1. Click the **Genotyping** tab to display the options listed below:

- **Multiplate Normalization (recommended)** becomes enabled for batches using more than one plate.
- **CN Reference Model File**: Contains reference information for CN analysis step.
- **CN Priors File**: Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
- **Analysis File**: This field displays the array you selected in the Array Type drop-down.
- **Prior Model File**: This field displays the default Prior Model File associated with the selected array. Click the drop-down to select a different file or add a new file.
- **SNP List File (recommended)**: This field displays the default SNP List File associated with the selected array. Click the drop-down to select a different file or add a new file.
- **ps2snp File**: If multiple probeset designs exist on the array for a given SNP (for example, one forward and one reverse strand design), then the ps2snp file is used by the SNP classification step to identify the best performing probeset for the SNP.
- **Genotype Frequency File**: If the library package supports a check for unexpectedly high call frequency for specific genotypes, this optional file specifies the maximum expected frequency for reviewed genotypes.
- **species-type**: Species type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.
- **cr-cutoff**: Minimum acceptable call rate.

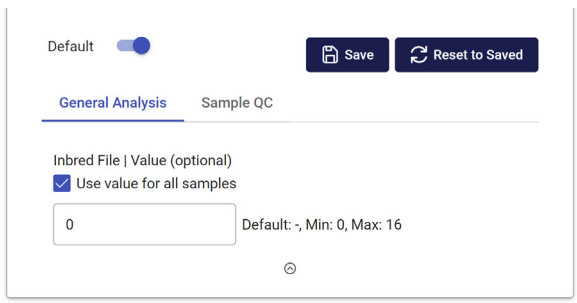
- **fld-cutoff**: For autosomal probesets, minimum acceptable FLD value for cluster separation.
- **het-so-cutoff**: Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.
- **hom-ro1-cutoff**: Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 1 genotype cluster.
- **hom-ro2-cutoff**: Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.
- **hom-ro-3-cutoff**: Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.
- **recommended**: ProbeSets having ConversionTypes in this recommended set will be included in the recommended probeset list.

General Analysis options (Optional)

Inbred File value for all samples

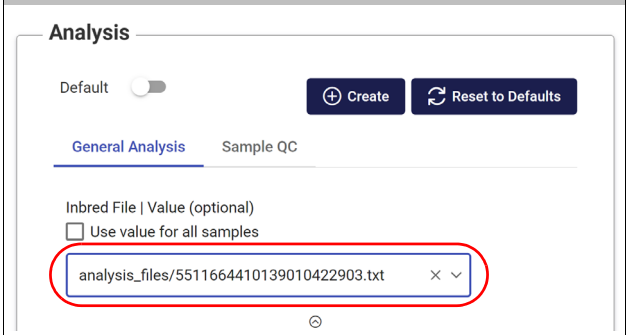
1. Click the check box, then enter a value. (Figure 38)

Figure 38 General Analysis pane - Inbred File



To add an Inbred file not listed in the drop-down (Figure 39), click **Add File**, then see "Add a file not listed" on page 39 for detailed instructions.

Figure 39 General Analysis options



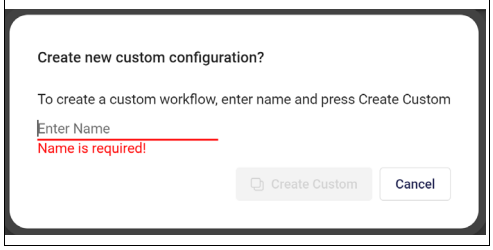
Save a custom configuration

1. Click **Create**.

   Note: If modifying an existing custom workflow, click **Save**.

   After clicking **Create**, a message window appears. (Figure 40)

Figure 40 Message window



2. Enter a workflow name, then click **Create Custom.**

   The new value is displayed.

Save an existing custom configuration

Once you modify an existing custom configuration, the **Save** button is enabled.

1. Click **Save**.

   The custom configuration is now saved.

Extension pane

The Extension pane (right) manages additional features and settings available for export or analysis within the Best Practices with Dual Workflow.

Extensions are enabled when available, as types vary depending on the array design. **Note:** Not every extension is available for every array design.

To use the Extension pane, see "How to use the extension pane" on page 65.

Add a file not listed

1. To select a file not listed on the drop-down menu, click **Add File**.

   An Explorer window appears.

2. Select the file you want, then click **Open**.

   If you are modifying an existing custom configuration the **Save** button is now enabled. If you are modifying a factory default workflow the **Create** button is enabled.

Enter a new value

1. To change a default value, click inside its text box.
2. Enter a new value.

   If you are modifying an existing custom configuration the **Save** button is now enabled. If you are modifying a factory default workflow the **Create** button is enabled.

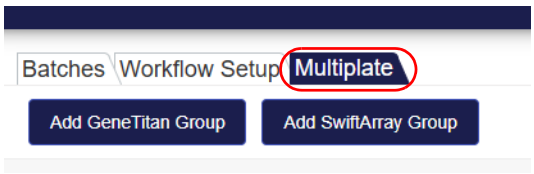
4 Multiplate

Use the Multiplate function to manually analyze batches made of 1 to 50 plates. Batches can be set up to include any extensions available for the selected array type.

GeneTitan multiplate

1. Click the **Multiplate** tab. (Figure 41)

Figure 41 Workflow Setup tab

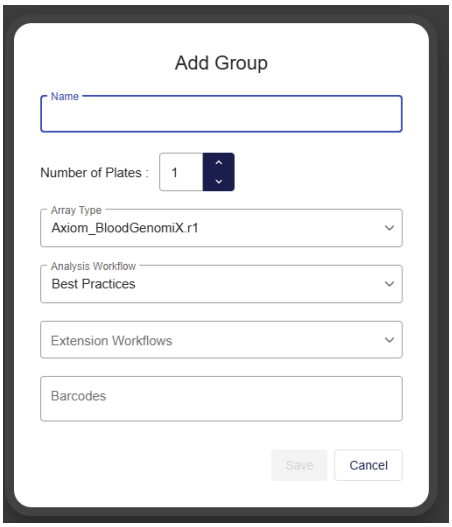


The **Multiplate** window tab appears.

2. Click **Add GeneTitan Group**.

The **Add Group** window appears. (Figure 42)

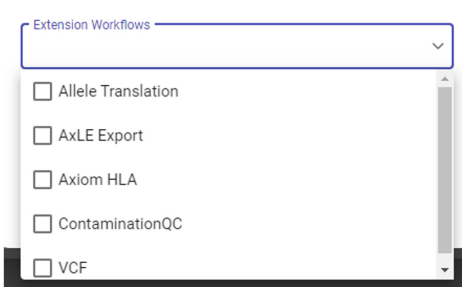
Figure 42 Add Group window



3. In the **Name** field, enter a batch name.
4. **Number of Plates:** Click arrows buttons to select the number of plates (1-50) or enter a number.

5. **Array Type:** Click the drop-down to select the array type. By default, array types are listed alphabetically.
6. **Analysis Workflow:** Click the drop-down to select a workflow.
7. **Extension Workflows:** (Optional and if available) Click the drop-down to display the available extension(s) (Figure 43), then click on its check box.

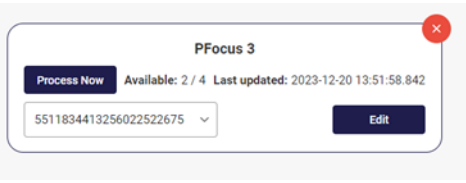
Figure 43 Extension Workflows



8. **Barcodes:** Use this field to paste (Ctrl+v) barcode(s), then click **Save**.

   If all plates have been uploaded, the analysis starts immediately. If there are any plates within the multiplate still in process on the GeneTitan, a multiplate card appears. (Figure 44)

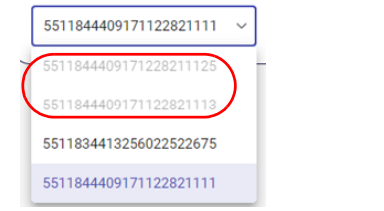
Figure 44 New multiplate card



1. Click the Barcode drop-down to view available barcodes.

   Note: A grayed-out barcode (Figure 45) indicates its upload to the server is still in progress.

Figure 45 Barcode drop-down



2. To run the plates shown in **BOLD**, click the Multiplate plate's **Process Now** button.

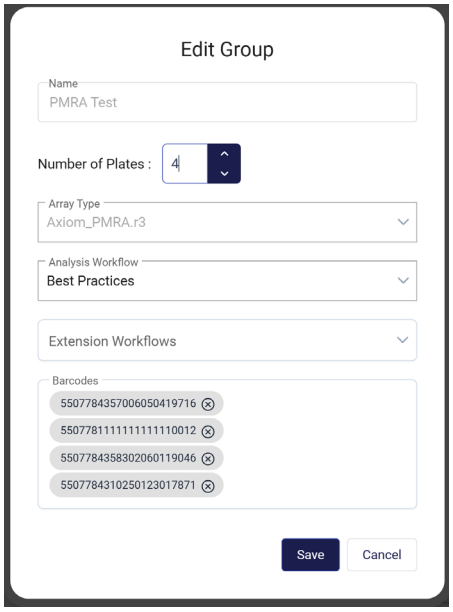
   A Confirmation pop up window appears.
3. Click **OK** to start processing the available plates.

Edit a multiplate group

1. Click the **Edit** button.

   The Edit Group window appears. (Figure 46)

Figure 46 Edit Group window



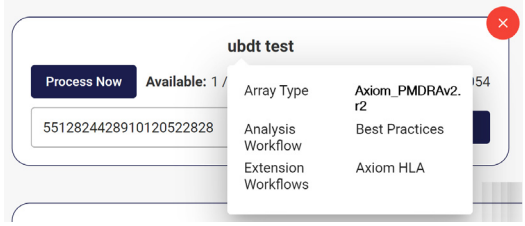
Use **Edit Group** to change:

- Number of plates
- Analysis Workflow
- Extension Workflow
- Barcodes: Paste any new barcodes here. To remove a barcode, click on its **X**.
- Click **Cancel** to cancel edits.
- Click **Save** to save all changes.

Multiplate quick view

1. Place the mouse pointer on a multiplate group card name to view its information. (Figure 47)

Figure 47 Quick view

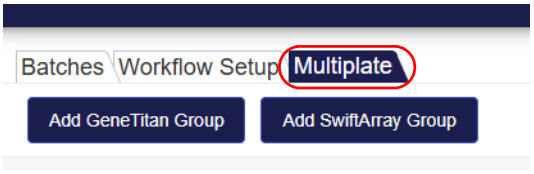


2. To remove the multiplate group card from the Multiplate window, click its **X** button.

SwiftArrayStudio multiplate

1. Click the **Multiplate** tab. (Figure 48)

Figure 48 Workflow Setup tab

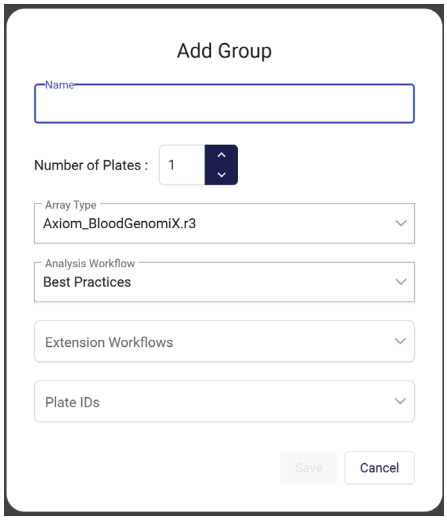


The **Multiplate** window tab appears.

2. Click the **Add SwiftArray Group**.

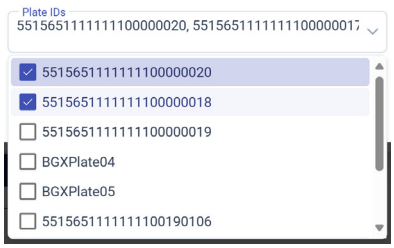
The **Add Group** window appears. (Figure 49)

Figure 49 Add Group window



1. Click the **Plate IDs** drop-down to view the available list, then click to check or uncheck the listed **Plate IDs,** as shown in Figure 50.

Figure 50 Plate ID drop-down



2. Click the Multiplate plate's **Process Now** button.

   A Confirmation pop up window appears.

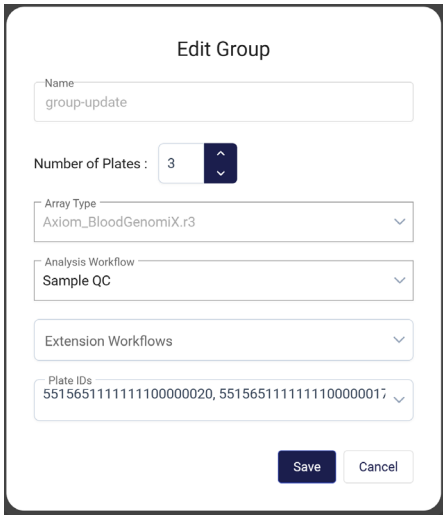
3. Click **OK** to start processing the available plates.

Edit a multiplate group

1. Click the **Edit** button.

   The Edit window appears. (Figure 51)

Figure 51 Edit Group window



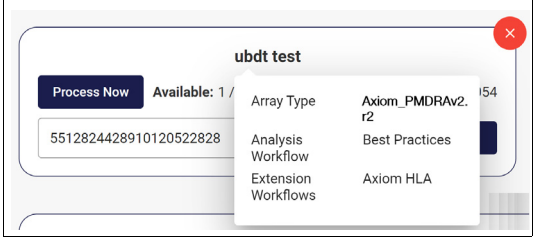
Use **Edit Group** to change:

- Number of plates
- Analysis Workflow
- Extension Workflow
- Plate IDs.
- Click **Cancel** to cancel edits.
- Click **Save** to save all changes.

Multiplate quick view

1. Place the mouse pointer on a multiplate group card name to view its information. (Figure 52)

Figure 52 Quick view



2. To remove the multiplate group card from the Multiplate window, click its **X** button.

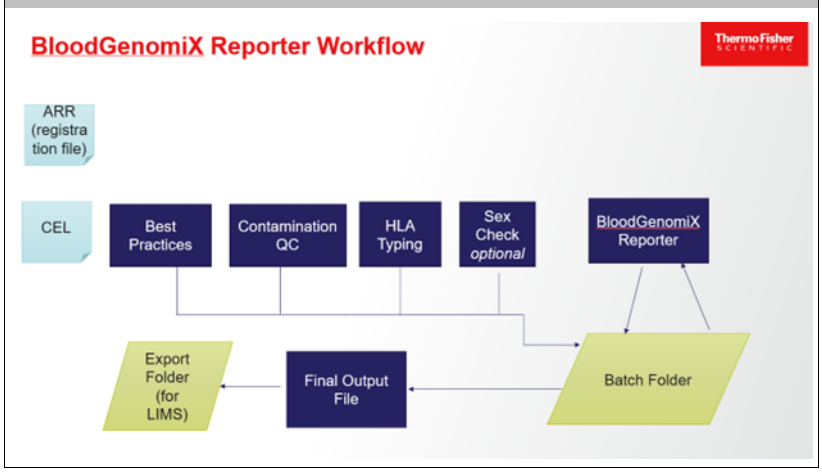
A BloodGenomiX Reporter

IMPORTANT! Reports must be pre-purchased before using the BloodGenomiX Reporter feature. See "Purchase reports" on page 56.

Overview and workflow

The BloodGenomiX Reporter workflow consists of the following analyses and quality controls steps:

Figure 53 BloodGenomiX Reporter workflow



- Best Practices
- ContaminationQC
- Axiom HLA
- Sex Check (Optional)
- Blood typing for research

Best Practices is the standard Axiom Best Practices workflow in which only the samples that pass the preset QC thresholds are genotyped and progressed further in the workflow. Best Practices includes copy number aware genotyping. For more information, see the *Axiom Genotyping Data Solutions Guide* (P/N MAN0018363).

The next step in the workflow is ContaminationQC. Contamination is estimated by the slope of a linear regression of intensity ratio of A or B allele for homozygous BB or AA calls for a given sample versus the plate frequency of the A or B allele. Based on predefined threshold values, samples are given a Yes or No result for contamination.

Samples that pass Best Practices and ContaminationQC are then Axiom HLA and blood typed. The Axiom HLA output is limited to the Class I loci (A, B, C). Extended blood phenotyping is based on the array and library file package.

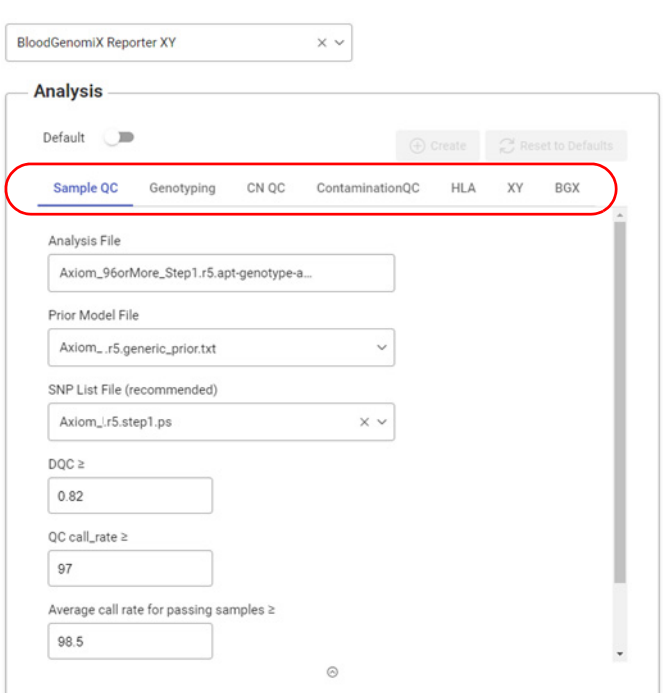
If the BloodGenomiX Reporter XY workflow is selected, computed sex is compared to the supplied sex, then either a PASS or FAIL will be reported. To generate a report *without* Sex Check, select **BloodGenomiX Reporter**.

For more information about other available Outputs and their settings, see Chapter 3.

BloodGenomiX analysis

The Analysis window contains a row of tabs. (Figure 54) Each tab is an analysis type. Click the **Analysis** tab to select it and view its available fields and values.

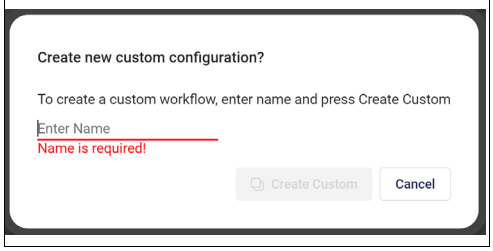
Figure 54 Analysis window - BloodGenomiX Reporter XY (includes Sex check) example



Note: Once a factory default workflow parameter is changed, a **Create** button appears. If changing a parameter within an existing custom workflow, a **Save** button appears.

After clicking **Create**, a message window appears. (Figure 55)

Figure 55 Message window



1. Enter a workflow name, then click **Create Custom.**
   The new value is displayed.

BloodGenomiX Analysis window tabs

Sample QC (fields and descriptions)

Analysis File	Displays the selected array.
Prior Model File	Defines prior knowledge of SNP cluster locations. This file has the same format as a posteriors file, which is generated by the genotyping step. This means that you can "train" on a custom data set, and use the updated knowledge of cluster locations as a "seed" to possibly improve future genotyping batches. This file must contain two row entries for the GENERIC and GENERIC:1 probesets (if there are any probesets to be genotyped that are not listed in this file).
SNP List File (recommended)	A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.
DQC	A sample's dish QC value must be greater than or equal to DQC to pass sample QC.
QC call_rate	A sample's call rate value must be greater than or equal to 'QC call_rate' to pass genotyping QC.
Average call rate for passing samples	A plate's average QC call rate of passing samples must be greater than or equal to this number to pass plate QC.
Percent of passing samples	Displays the Percent of passing samples value associated with the selected array. **Note:** If a plate's percent of passing samples is smaller than this number, all samples on the plate will display a warning in the Sample Table.

Genotyping (fields and descriptions)

Multiplate Normalization	Multiplate normalization is recommended (checked by default) for multiple plate genotyping analysis when individual plates are expected to have similar allele frequencies, i.e. a similar mix of genetic ancestry with no (or the same) enrichment for specific blood types.
Analysis File	Displays the selected array.
Prior Model File	Defines prior knowledge of SNP cluster locations. This file has the same format as a posteriors file, which is generated by the genotyping step. This means that you can "train" on a custom data set, and use the updated knowledge of cluster locations as a "seed" to possibly improve future genotyping batches. This file must contain two row entries for the GENERIC and GENERIC:1 probesets (if there are any probesets to be genotyped that are not listed in this file).
SNP List File (recommended)	A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.
CN Reference Model File	Contains reference information for CN analysis step.
CN Priors File	Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
ps2snp File	If multiple probeset designs exist on the array for a given SNP (for example, one forward and one reverse strand design), then the ps2snp file is used by the SNP classification step to identify the best performing probeset for the SNP.
Genotype Frequency File	If the library package supports a check for unexpectedly high call frequency for specific genotypes, this optional file specifies the maximum expected frequency for reviewed genotypes.
species-type	Species type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.
cr-cutoff	Minimum acceptable call rate.
fld-cutoff	For autosomal probesets, minimum acceptable FLD value for cluster separation.
het-so-cutoff	Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.
hom-ro1-cutoff	Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 1 genotype cluster.
hom-ro2-cutoff	Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.
hom-ro-3-cutoff	Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.
recommended	ProbeSets having ConversionTypes in this recommended set will be included in the recommended probeset list.

CN QC (fields and descriptions)

MAPD	For arrays that support copy number (CN) analysis, the Median Absolute Pairwise Difference of log2ratio signals of adjacent copy number (CN) probesets must be less than this value to make CN calls.
Waviness SD	For arrays that support copy number (CN) analysis, the Waviness Standard Deviation of log2 ratio signals of copy number (CN) probesets must be less than this value to make CN calls.

ContaminationQC (fields and descriptions)

ProbeSet ID	ProbeSet in this list will be used to determination contamination.
Genotype Clusters	Clusters that will be used to generate the linear model used to determine cross contamination.
Threshold p-value	Threshold for adjusted p-value, samples with adjusted p-value greater than threshold will have contaminated set to **No** in the output.
Threshold estimated contamination	Samples with a value less than threshold will have contaminated set to **No** in the output.

HLA (fields and descriptions)

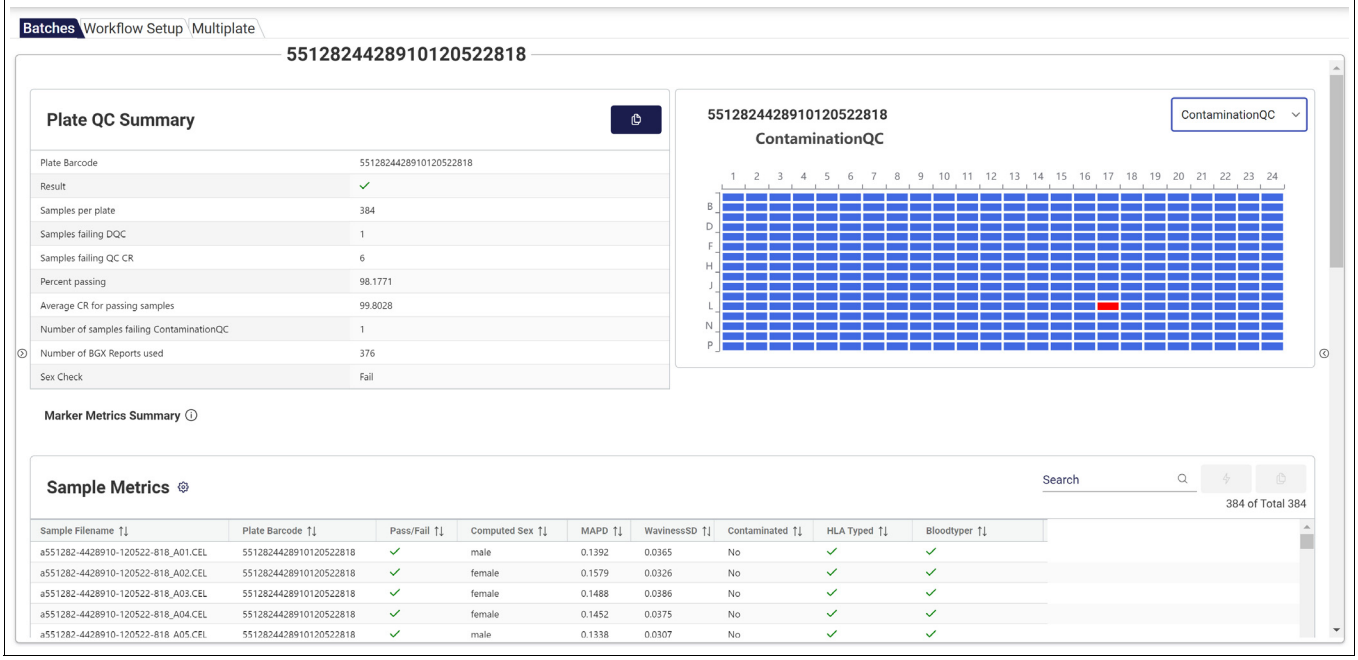
Loci Selection (default All)	Loci A, B, C are options for HLA typing.
Output Folder	Location and name of output folder.
Output Name	Name of the output which includes 2 or 4 digit as well as loci. Example: (**AxiomHLA_2_A_Results**)

BGX (fields and descriptions)

Antigen list	List of antigens and applicable alleles that will be typed. A two column text file with the headers: **test_group_name** and **test_name**. Test group name is **Blood Group System** and test name is **antigen/allele name**.
Output Folder	Name of the Results folder.

View a BloodGenomiX batch

Figure 56 Batches window tab - BloodGenomiX Reporter example



The BloodGenomiX Reporter creates two output folders:

- The **BGXPrimary** folder contains the initial files and folders that are generated from the BloodGenomiX workflow. This output is then merged into the BloodGenomix Report.
- The **BGX** folder contains the report file in text format. The results from the primary output files along with the QC results from the Best Practices analysis are collated into a BGX Report file.

BGXPrimary folder contents

The BGXPrimary folder contains the following files:

apt2-bloodtyper

This text file contains samples and corresponding blood types.

cel_file	Sample file
test_group_name	Defines the Blood group system: HEA (Human Erythrocyte Antigen, HPA (Human Platelet Antigen) or, HNA (Human Neutrophil Antigen).
test_name	Blood type that is dependent on array and antigen group file that was used for analysis.
test_description	Antigen or Allele

result_value	**Positive**: Selected test_name is detected in the sample, **Negative**: Selected test_name is not detected in the sample, **NT**: Not Typed, selected test_name antigen was not analyzed **Not_present**: Selected test_name allele is not detected in the sample. **HEM**: Test_name allele is present as hemizygous **HET**: Test_name allele is present as heterozygous **HOM**: Test_name allele is present as homozygous **PARTIAL**: Phenotype is partially expressed **WEAK**: Reduced phenotype expression
result_status	**PASS**: Sample is found to be either positive or negative for the corresponding test_name. **FAIL**: Sample was not typed, no result was found

AxiomBT.bloodtyper_summary

This text file contains the frequency of antigen or alleles in the batch.

Test type	Defines test; antigen or allele
Antigen or Allele Name	Test_name
Pos/Hom Samples	Number of samples that are positive and homozygous for antigen or allele.
Neg/Not Present Samples	Number of samples that are negative and not present for antigen or allele.
Weak/Het Samples	Number of samples that are weak and heterozygous for antigen or allele.
Partial/Hem Samples	Number of samples that are partial and hemizygous for antigen or allele.
NT Samples	Number of samples that are not typed for antigen.
Percent Pass	Percentage of samples that pass for given antigen or allele.

AxiomBT.marker_calls

This text file contains the test_name and the associated genotype or copy number call in cname format.

cel_file	Sample file
test_group_name	Blood group system: HEA, HPA or HNA, and antigen or allele type.
test_name	HGVS cname
result_value	Genotype calls in forward strand format or copy number call.
result_status	PASS or FAIL

blood-sample-list

This single column text file contains the samples that were typed.

collected-attributes

This text file contains the collated attributes of the samples in the batch. Note: SwiftArrayStuidio batches

BGXPrimary subfolders

Contamination

This folder contains the text file that shows contamination status of the samples in the batch.

cel_file	Sample file
alpha	Estimated contamination threshold, proportional to the level of contamination, value is expected to be centered on 0.0, default: 0.02.
p-value	The p-value of the linear regression fit. Null hypothesis is that the slope (alpha) is zero. The default p-value threshold is 0.005.
contaminated	Yes or No, depending on the alpha and p-value of the sample.

HLA

The analysis sub-folder within this HLA folder contains the intermediary and results files for the A, B, and C loci.

BGX folder contents

The BGX folder contains the Report file that is in a text (TXT) format. The results from the primary output files along with the QC results from the Best Practices analysis are collated into the BGX Report. The Report file displays each sample result in a single row.

Each BGX Report displays these same default column headers:

Note: The sample name (that is associated with the attribute file) MUST BE alphanumeric only with no special characters or symbols. (Example: 22IETAW3RUTG)

sample_identifier	Sample name; derived from the ARR file **Note:** For more information on ARR, see the GeneChip Command Console (GCC) User Guide (P/N MAN0027771).
cel_file	.cel file
array_barcode	Array barcode
run_identifier	Automated Axiom batch ID
method	Array name
result_date	ISO8601 date time stamp of the analysis

The remaining columns: **software**, **test_group_name**, **test_name**, **test_description**, **result_value**, **result_unit**, and **result_status** are specific for each primary BGX report file. These columns are detailed in the following filename tables:

Collected-attribute

The BloodGenomiX Reporter requires a customized batch registration file. To obtain one or for more information, contact your local field support person.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
ARR	Attributes	Well location	Sample Attributes	well	string	PASS/FAIL
		Source plate barcode		barcode	string	PASS/FAIL
		Source plate well location		well	string	PASS/FAIL
		sample type		source of DNA	string	PASS/FAIL
		Customer supplied sex		Female or Male	string	PASS/FAIL
		Date received		date	datetime	PASS/FAIL
		Reagent Lot		Lot number	string	PASS/FAIL
		Project name		Name	string	PASS/FAIL

Output from the AxiomGT1.contamination

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
apt2-contamination-qc version #	ContaminationQC	p-value	ContaminationQC	Example: 0.476659	integer	PASS/FAIL
		Estimated contamination		Example: 0.00242784	integer	PASS/FAIL
		Contaminated		No	string	PASS/FAIL

Output from HLA

The HLA section of the BGX Report contains the output from Loci A, B and C.

X and Y are designations for allele 1 and allele 2.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
apt2-HLA	HLA Locus Genotype	HLA-locus-x	type	HLA-four-field	four digit	PASS/FAIL
		HLA-locus-y	type	HLA-four-field	four digit	PASS/FAIL
		HLA-locus-p1	probability	probability to 2 decimals	unit interval	PASS/FAIL
		HLA-locus-p2	probability	probability to 2 decimals	unit interval	PASS/FAIL

QC Metrics

The sample QC metrics are ported from the Best Practices analysis.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
AutoAx	QC	DQC	Sample QC Metrics	example: 0.98617	float	PASS
		QC Call Rate		example: 99.84586	float	PASS
		MAPD		example: 0.136106	float	PASS/FAIL
		WavinessSD		example: 0.036856	float	PASS/FAIL

Antigen Genotype

From the **AxiomBT.marker_calls** file, the BGX Report collates the batch samples' genotype or copy number calls per test name in HGVS (cname) nomenclature.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
AutoAx	Blood group name	HGVS cname or CN Region	genotype	Forward strand or Copy Number Call	genotype	PASS/FAIL

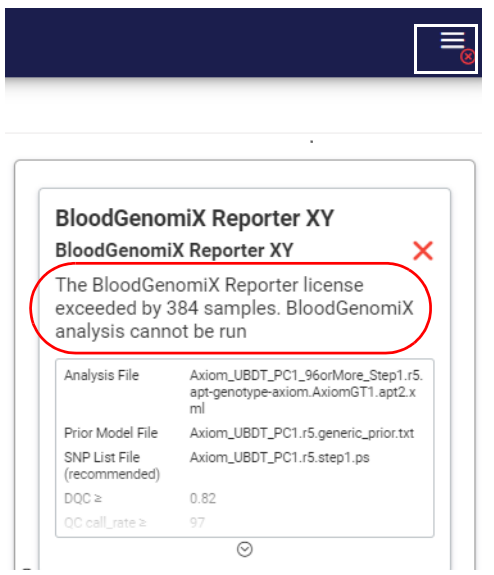
Extended Blood Typing

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
apt2-bloodtyper, version #	Blood group name	Blood Type	Antigen or Allele	POS NEG NT NOT_PRESENT HEM HET HOM PARTIAL WEAK	antigen type	PASS/FAIL

Purchase reports

An alert (Figure 57) appears stating reports must be purchased in order to unlock and use AutoAx's BloodGenomiX reporting feature.

Figure 57 BloodGenomiX Reporter Reports remaining alert/message

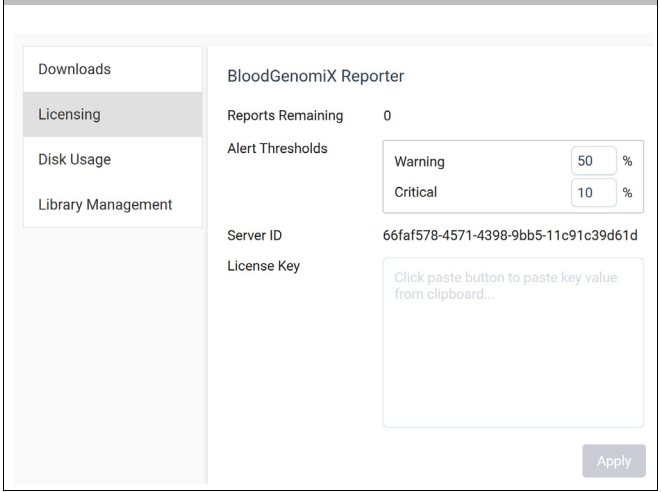


1. Contact your Thermo Fisher Sales representative for a BloodGenomiX Reporter quote.
2. Click on (far right), then click **Configurations**.

   The **Batch Download Options** window appears.
3. Click **Licensing**.

   The **BloodGenomiX Reporter** window appears. (Figure 58)

Figure 58 BloodGenomiX Reporter window

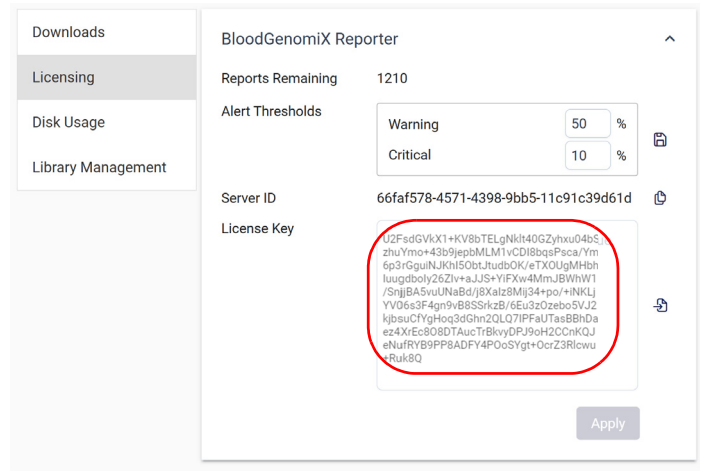


4. Click the **Server ID** field's **Copy to Clipboard** button.
5. Open a new email, then paste the Server ID into it.
6. Enter your Purchase Order number to the email, then send it your Thermo Fisher Sales Representative.

   Note: Allow 48 hours for your order to process. After processing, you will receive an email containing a license key.

7. Copy the license key string, then paste it into the **License Key** field, as shown in Figure 59.

Figure 59 License Key string field



The **Apply** button is enabled.

8. Click **Apply**.

   The **Reports Remaining** count updates.

Customize thresholds (Optional)

After clicking **Apply**, you can change these two default **Threshold** values:

- **Warning:** This threshold's default is 50%. Meaning: When 50% of the reports have been used, a yellow triangle icon will appear on the hamburger-shaped icon (top right), as shown below.

- **Critical**: This threshold's default is 10%. Meaning: When 10% of the reports are remaining, a red circle with **X** icon will appear on the hamburger-shaped icon (top right), as shown below.

  - To change these default values, click inside each percentage field to enter a different value, then click the **Apply** button. Click the **Batches** window tab to close the Licensing window.

IMPORTANT! The number of reports must be **equal to or greater** than the number of samples in the batch that are to be processed. If not, BloodGenomiX Reporter will not run.

B Archiving

GCC Uploader information

The **GCC File Uploader**, uses APIs to upload CEL and ARR files to the server from a GeneTitan MC Workstation.

The GCC Uploader uses APIs to notify the AutoAx server when a plate is complete. A plate complete notification triggers a batch to be created, its workflow steps queued, and when able, it is processed.

SwiftArrayStudio information

SwiftArrayStudio™ Data Collection Software is an integrated web application enabling users to:

- Manage 1-4 instruments
- Monitor plate runs remotely by viewing progress on the application dashboard
- Sample and SwiftArray plate registration
- View individual plates post image processing
- Submit service and support tickets using SmartHelp™

How to archive AutoAx

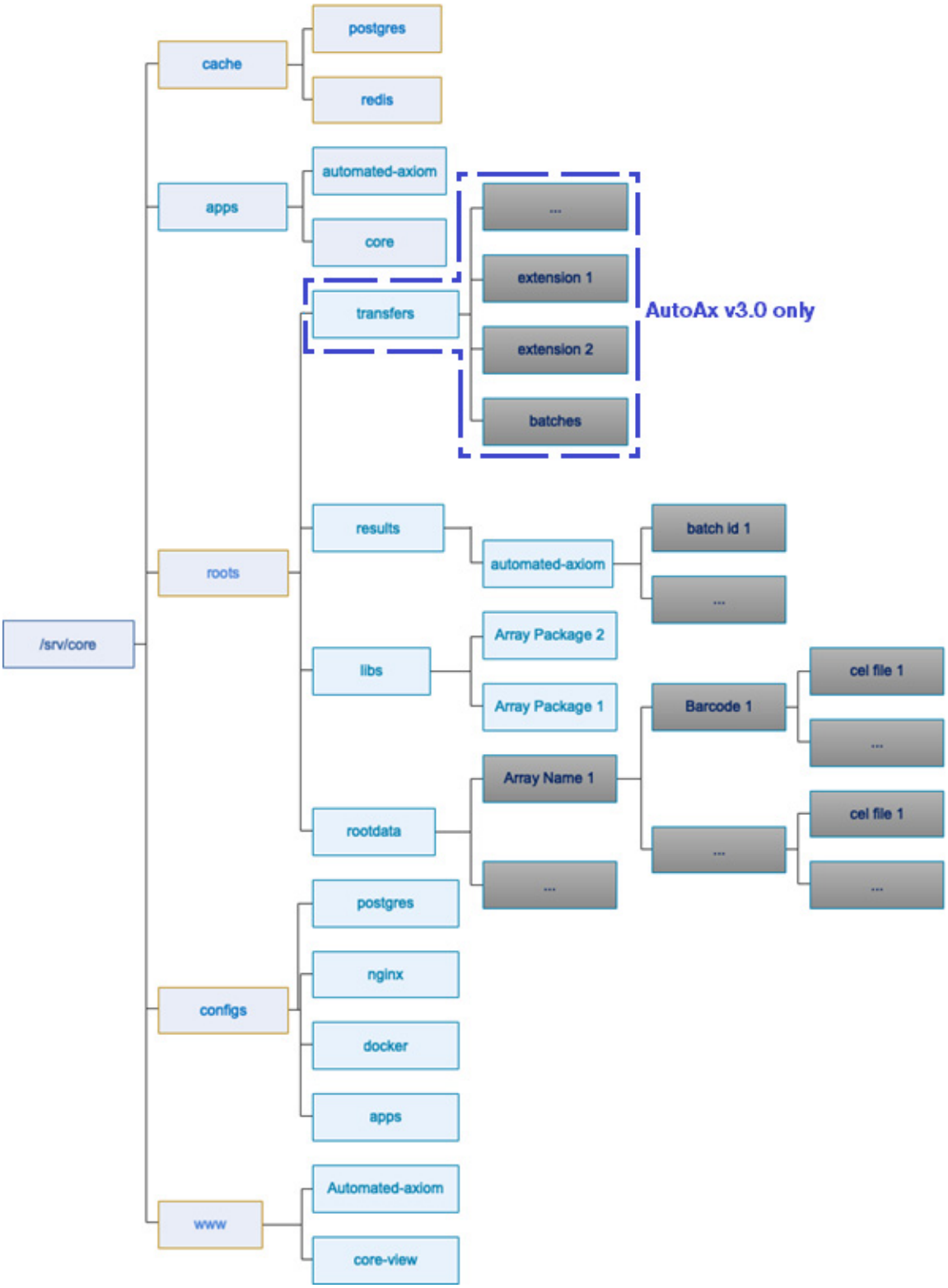
- See "Archive v3.0 and v3.1" on page 61.

AutoAx directory (workflow) tree

- Plate data is stored by its array type name, then by sub-directories named using the plate's barcode. This plate data location is at: **/srv/core/roots/rootdata**

  Example: **/srv/core/roots/rootdata/Axiom_BovMDv3/5510894388058030721881**
- Results are here: **/srv/core/roots/results/automated-axiom**
- Each batch is stored in a sub-directory by its **ID** as seen on the AutoAx application's dashboard. **Note:** Boxes shown in gray (Figure 60) can be archived.

Figure 60 AutoAx directory (workflow) tree - **Directories in gray are archiveable**



Archive v3.0 and v3.1

IMPORTANT! All archiving procedures listed below are intended for script writers, command line users, and those with HTTP and Linux experience only.

Note: All APIs in AutoAx v3.0 and v3.1 for results (batch information) use **RESTful api** endpoints. To make API endpoint calls, tools such as **curl**, **mget**, or **postman** are required. Specific endpoints are required to be called to help ensure the system stays in sync with any data that may have been archived.

Archiving of **roots/rootdata** does not require synchronization of the system, however *Results* (such as batches), do require the system to be synchronized.

Retrieve and sort batches

Before you can view the **batchId**, the batch information must be retrieved from the system. The screen examples that follow (Figure 61) show how to retrieve the batchId based on the ID and the display name, which is at the top of the batch information card.

Note: If needed, change or remove **sortby** and/or **sortorder**. The barcode ending in **2828** is used in the screen examples that follow.

If searching for the ID, replace 2828 with the value of the ID. The results are a collection of batches that match the filter value in any field of the batch. It is very important to review the results before using the **batchId** command. Doing so will help ensure the batch you want to remove is the correct one.

The following example screens within Figure 61 show how to retrieve the batchId based on the ID and the display name, which is at the top of the batch information card.

Figure 61 Different methods for retrieving, changing, and sorting batches

Postman Method

Curl Method

```
1. curl --location '10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id%2CbatchId%2Cname'
2.
```

Powershell Method

```
1. $response = Invoke-RestMethod '10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id,batchId,name' -Method 'GET' -Headers
$headers
2. $response | ConvertTo-Json
3.
```

Python Request Method

```
1. import requests
2. url = "10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id,batchId,name"
3. payload = {}
4. headers = {}
5. response = requests.request("GET", url, headers=headers, data=payload)
6. print(response.text)
7.
```

wget Method

```
1. wget --no-check-certificate --quiet \
2.   --method GET \
3.   --timeout=0 \
4.   --header '' \
5.    '10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id,batchId,name'
6.
```

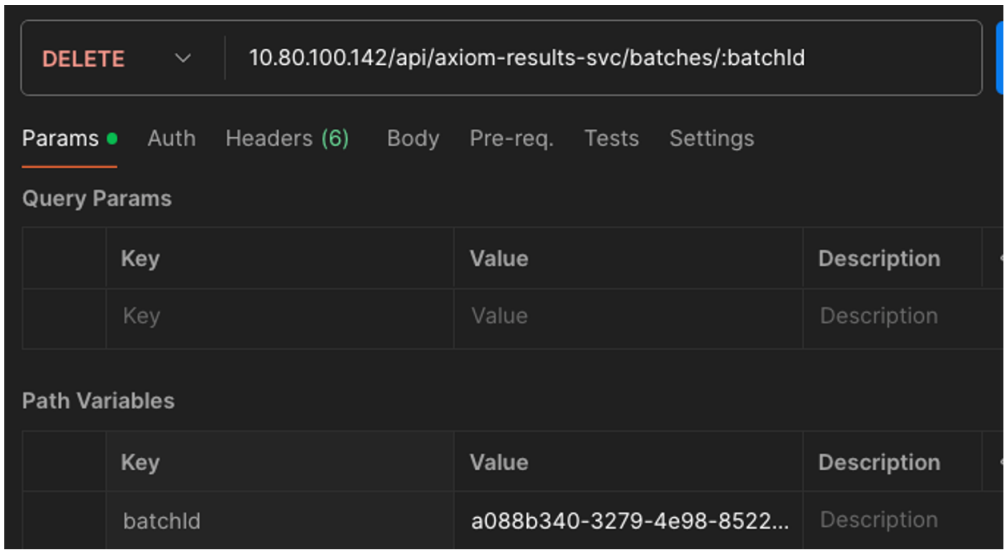
Remove Batches using API

- Internally, all batches are identified and referenced by the **batchId**, not the ID.
- The **batchId** is a **U**niversal **U**nique **I**dentifier (UUID)
- If you want to remove a batch from the system without having to ssh, delete, and refresh the cache, you can use the remove api. This is a two-step process.
- To know the batchId for any batch, the batch information must be retrieved from the system.

  After the batchId you want has been retrieved, use the remove api call using one of the methods shown in Figure 62.

Figure 62 Different methods for using the remove api call

Postman Method



Curl Method

```
1. curl --location --request DELETE '10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486'
```

PowerShell Method

```
1. $response = Invoke-RestMethod '10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486' -Method 'DELETE' -Headers $headers
2. $response | ConvertTo-Json
3.
```

Python Request Method

import requests

```
1. url = "10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486"
2.
3. payload = {}
4. headers = {}
5.
6. response = requests.request("DELETE", url, headers=headers, data=payload)
7.
```

print(response.text)

wget Method

```
1. wget --no-check-certificate --quiet \
2.   --method DELETE \
3.   --timeout=0 \
4.   --header '' \
5.    '10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486'
6.
```

Re-sync all batches

After the selected batches have been archived off the server, they can be re-synced. Below, examples for each tool will be provided for how to synchronize the system.

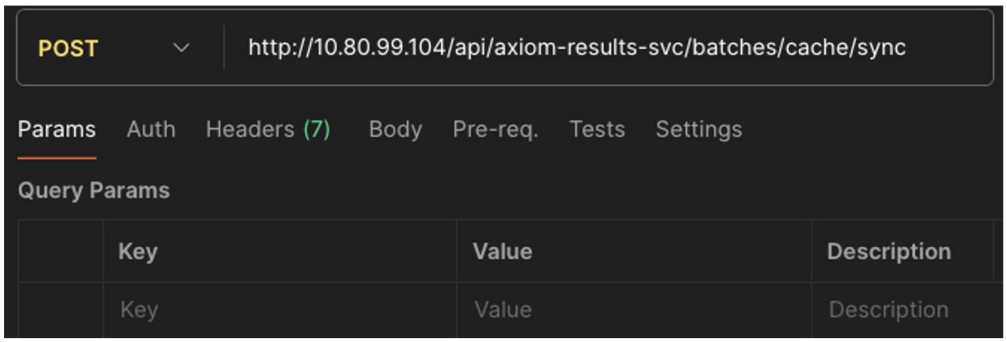
There are many tools/options for performing HTTP endpoint calling. The screen examples in Figure 63 show the most common.

The time to synchronize is based on the number of batches and the speed of the server and/or network connection.

Note: In all cases, you must change the currently displayed IP address to match the server's IP address.

Figure 63 Different methods to re-sync batches

Postman Method



Curl Method

```
1. curl --location --request POST '10.80.99.104/api/axiom-results-svc/batches/cache/sync'
2.
```

Powershell REST method

```
1. $response = Invoke-RestMethod '10.80.99.104/api/axiom-results-svc/batches/cache/sync' -Method
'POST' -Headers $headers
2. $response | ConvertTo-Json
3.
```

Python Method

```
1. import requests
2.
3. url = "10.80.99.104/api/axiom-results-svc/batches/cache/sync"
4.
5. payload = {}
6. headers = {}
7.
8. response = requests.request("POST", url, headers=headers, data=payload)
9.
```

print(response.text)

wget Method

```
1. wget --no-check-certificate --quiet \
2.   --method POST \
3.   --timeout=0 \
4.   --header '' \
5.    '10.80.99.104/api/axiom-results-svc/batches/cache/sync'
6.
```

C Extensions

How to use the extension pane

The Extension pane (right) manages features or settings available for export or analysis options beyond the Best Practices and Best Practices with Dual Workflows.

Extensions are enabled when available, as types vary depending on the array design.
Note: Not every extension is available for every array design.

1. To set a specific Extension as default with no customization, toggle **Default** on. (Figure 64) When on, the selected export file (from the drop-down) is automatically generated and placed in the **Extension** folder of the batch.

VCF extension

When set as default, a VCF export file is automatically generated and placed in the **Extension** folder of the batch.

Input tab

(Figure 64)

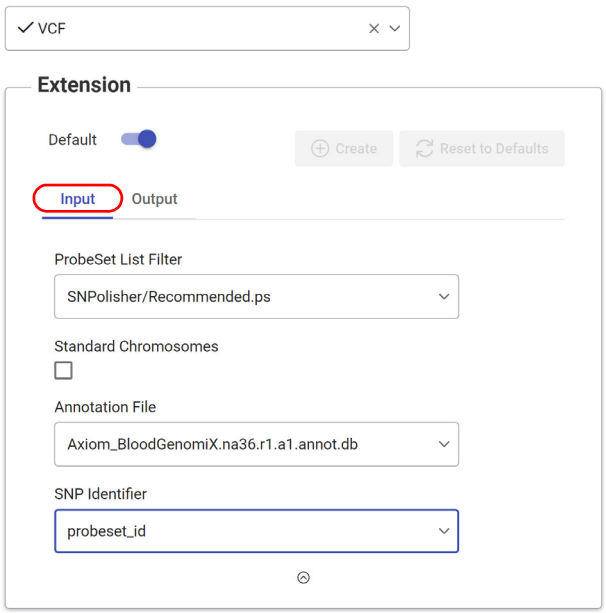
Note: Depending on what array you are running, the extension options may differ.

- Click the available drop-down to select a different file or add a new file.
- Optional: Click the **Standard Chromosomes** check box to include only probsets that are assigned to standard chromosomes or other annotated contigs.
- To add a file to the VCF Input tab entries above, see "Add a file not listed" on page 39.

Note: Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

Note: Dual workflow samples are now included in the VCF export.

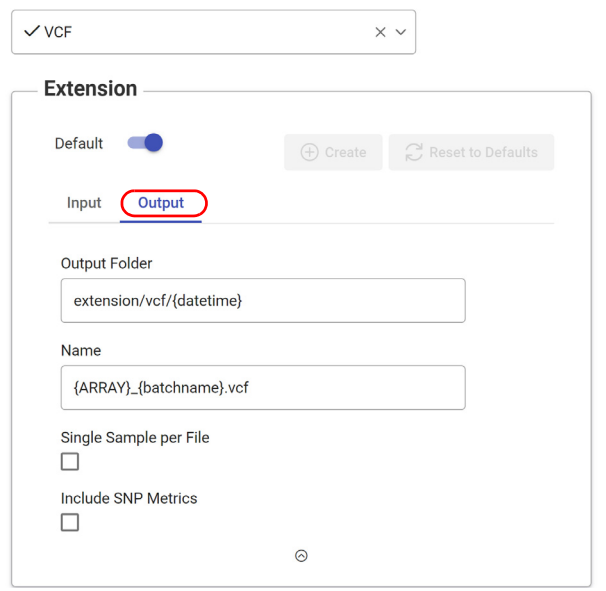
Figure 64 Extension pane - VCF Input



Output tab

(Figure 65)

Figure 65 Extension pane - VCF Output



- **Output Folder:** Displays the output folder's location.

- **Name:** Displays the selected extension's file naming convention.
- **Single Sample per File:** Click this check box to generate one file per sample.
- **Include SNP Metrics:** Click this check box to include CR, FLD, HomFLD, HomRO, and HetSo to the analysis.

  Note: Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

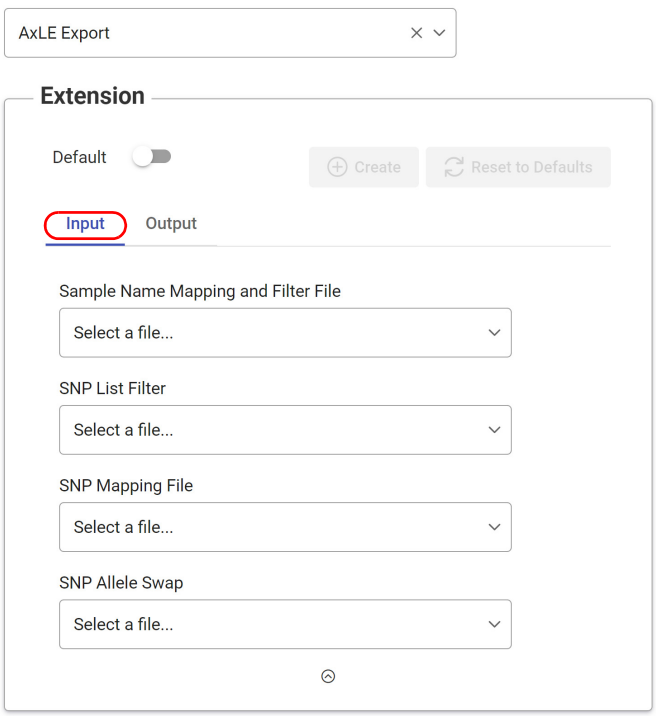
AxLE extension

Input tab

(Figure 66)

Note: To learn more about these AxLE Export inputs, see the *Axiom Long Format Export User Guide* (P/N MAN0027964).

Figure 66 Extension pane - AxLE Export



Note: Depending on what array you are running, the extension options may differ.

- Click the available drop-down to select a different file or add a new file.
- To add a file to the entries above, see "Add a file not listed" on page 39.

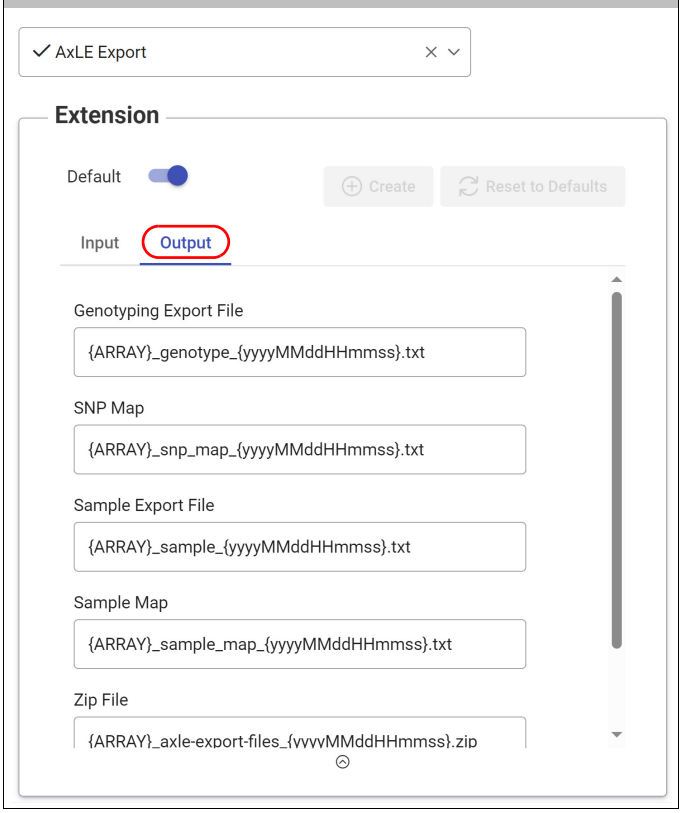
  Note: Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

Output tab

(Figure 67)

Note: To learn more about these AxLE Export outputs, see the *Axiom Long Format Export User Guide (*P/N MAN0027964*)*.

Figure 67 Extension pane - AxLE Export



- **Genotyping Export File:** Displays the file's naming convention.
- **SNP Map:** Displays the SNP Map's naming convention.
- **Sample Export File:** Displays the file's naming convention.
- **Sample Map:** Displays the Sample Map's naming convention.
- **Zip File:** Displays the file's naming convention.
- **Include SNP Metrics** (scroll down to view): Click this check box to include CR, FLD, HomFLD, HomRO, and HetSo to the analysis.

  Note: Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

Allele Translation extension

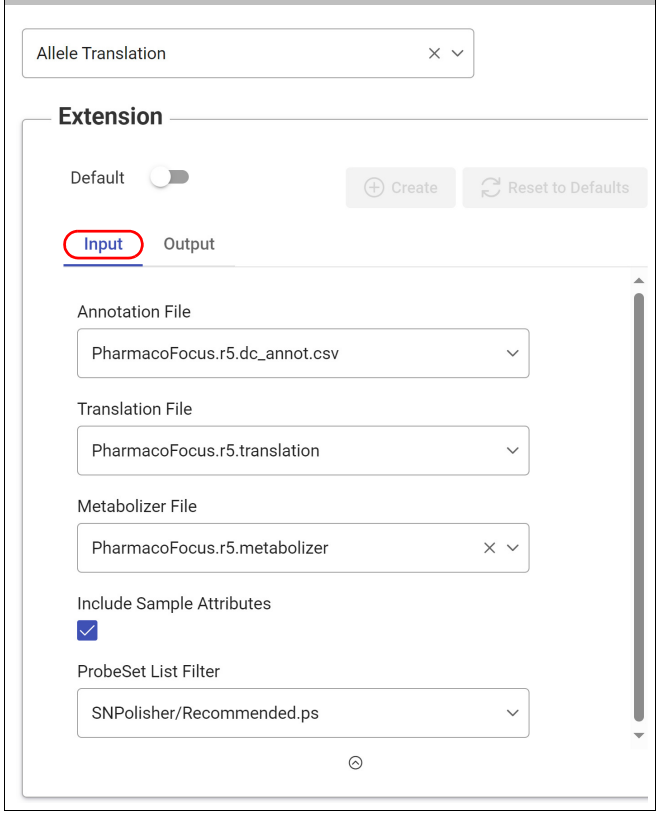
An **Allele Translation** extension appears as an extension option (Figure 68) if the arrays you are using have been enabled. See the *Axiom Analysis Suite User Guide* (P/N MAN0027928) for more information.

Input tab

Note: Depending on what array you are running, the extension options may differ.

- Click the available drop-down to select a different file or add a new file.
- If you disagree with the phenotype interpretations, you may want to leave the Metabolizer File option blank. If left blank, a Phenotype report will not be created. If you want to include **Sample Attributes**, click its check box.

Figure 68 Extension pane - Allele Translation

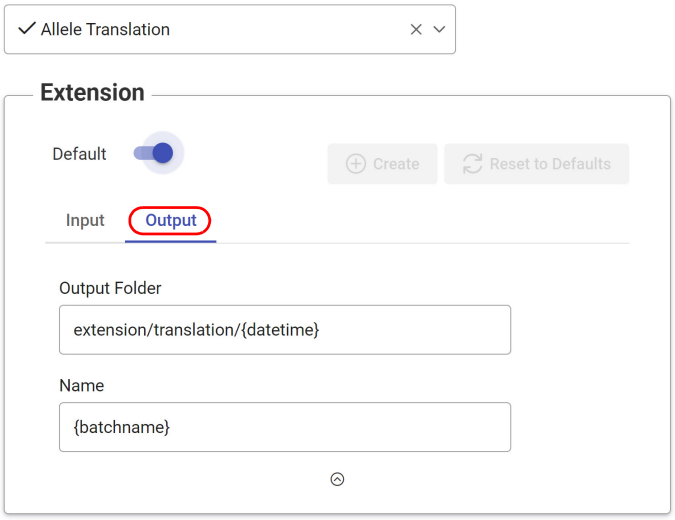


Output tab

- Click the **Extension** pane's **Output** tab to view the **Output Folder** location and naming convention. Click the **Default** toggle on to automatically run Allele Translation for arrays that are enabled. **Note:** If any options have been modified the **Create** and **Reset to Default** buttons appear.

  Note: Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

Figure 69 Extension pane - Allele Translation



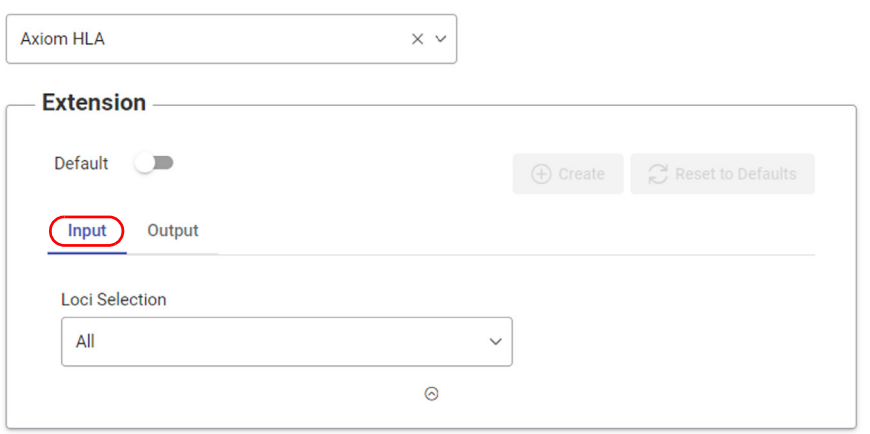
Axiom HLA extension

For arrays that are enabled, generating human leukocyte antigen calls is an extension option. Axiom HLA uses a multi-population reference panel and the HLA type imputation model HLA*IMP:021 to statistically infer the HLA types of human samples from genotype data. The loci include Class I (A, B, C) and Class II (DRB1, DRB3, DRB4, DRB5, DQA1, DQB1, DPA1, DQB1).

Input tab

(Figure 70)

Figure 70 Extension pane - Axiom HLA

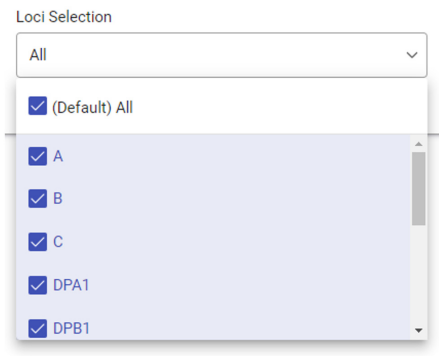


1. Click the **Loci Selection** drop-down.

   Note: Once a new Loci is selected, the **Create** and **Reset to Defaults** buttons become enabled.

2. All Locis are selected by default. (Figure 71) Click on a check box(es) to de-select a Loci from the analysis.

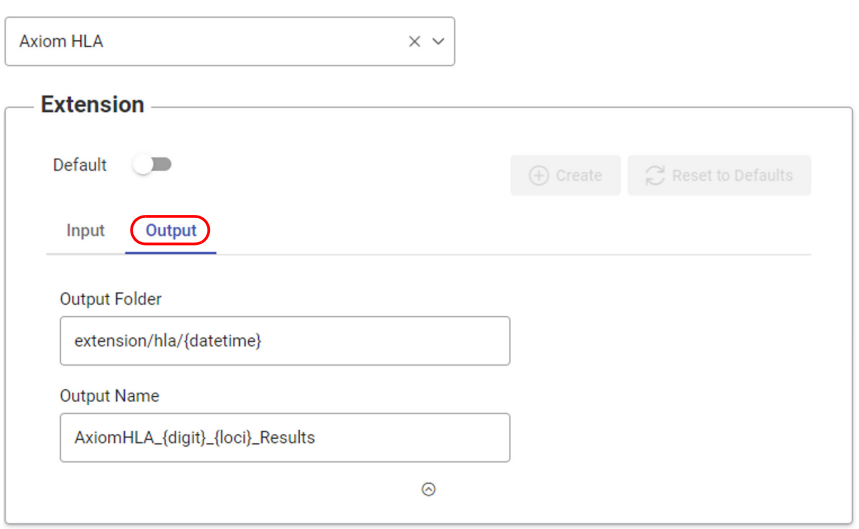
Figure 71 Loci Selection drop-down



Output tab

(Figure 72)

Figure 72 Extension pane - Axiom HLA



- **Output Folder:** Displays the output folder's location.
- **Output Name:** Displays the selected extension's file naming convention.
- (Optional) Click the **Default** toggle right (on) to apply this extension to the designated Axiom HLA array type.

  Note: Once a parameter is changed or a file is selected, the **Create** and **Reset to Defaults** buttons become enabled.

How to access extension outputs

For direct access to the BloodGenomiX Report (and any other final output), AutoAx v3.1 contains this additional folder in its directory tree: **/srv/core/roots/transfers**

- The **transfers** folder contains sub-folders for each extension.
- The BloodGenomiX Report resides here:

  /srv/core/roots/transfers/BGX

Enable access to final outputs

Direct access to final outputs is set to **on** by default. If this feature needs to be restored, go to path:

/srv/core/configs/apps/automated-axtiom/settings/autoxfer.json

1. *nano* into the file.
2. Change line #30 *transfer* property to **true** (Figure 73), then press **Enter** to save the .json file.

Figure 73 Editing the transfer property example

```
27        {
28            "name": "best_practices_for_blood",
29            "internalBatchPath": "BGX",
30            "transfer": true,
31            "destination": "transfers/BGX",
32            "deleteSrc": false,
33            "type": "xfer",
34            "imageTag": "core/core-sh:latest",
35            "taskType": "lite"
36        },
```

Note: If you modify the **.json** file with tasks still in the pipeline, those tasks will complete using the previous .json file. The saved **transfer** property edit takes effect AFTER the task pipeline clears.