Variant Reporter™ Software v1.0

Finds Variants Quickly, Easily, and Accurately

- Handles greater than 10× more sample files than competing software products
- Accelerated data review reduces labor and project cost
- Fast, accurate heterozygote calling improves results
- Simple to learn—requires less than one hour
- Robust data filtering with new, more effective quality metrics

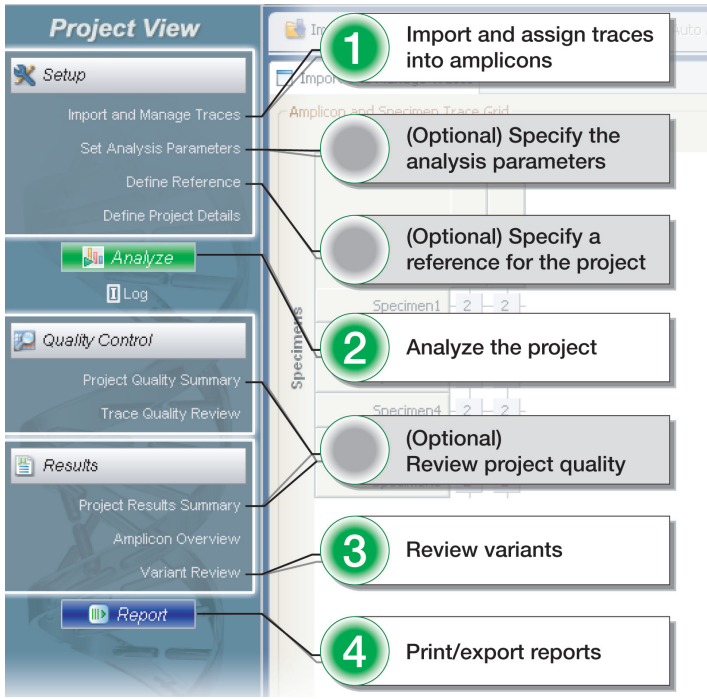

Figure 1. Simple, task-based workflow

Introduction

Manually reviewing your resequencing results requires valuable time and reduces productivity. Applied Biosystems' new Variant Reporter™ Software v1.0 does it all for you, quickly, efficiently, and accurately. Designed for basic and clinical researchers engaged in genotyping projects, as well as core resequencing labs whose focus is secondary analysis, this new variant detection software solves the workflow bottleneck caused by the 3–8 hours often required by other software products for data analysis and review. And with your resequencing workflow fully automated, you are free to concentrate on other aspects of your project.

Researchers who are new to the lab will find Variant Reporter Software simple, straightforward, and intuitive with a very gentle learning curve. The software also provides robust, high-quality trace, consensus, and variant metrics. Additionally, it is compatible with Windows® 2000, XP and Vista operating systems.

Simple Workflow

Because you don't have all day to analyze results and manage a complex workflow, Applied Biosystems designed its new Variant Reporter Software to be as fast and simple as possible. The result is an accelerated procedure that involves only a few simple steps (Figure 1).

Improved Accuracy

Variant Reporter Software includes improved algorithms that have been trained on large data sets to yield the most accurate and sensitive variant discovery, which is ideal for rare mutant detection (Figure 2).

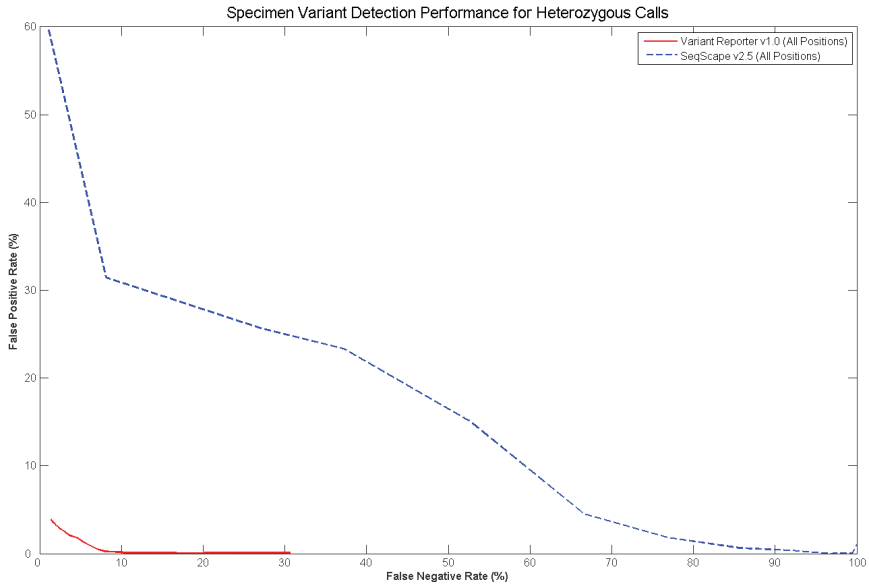

Figure 2. Improved variant detection performance as demonstrated on over 4,114,620 million base calls

Fastest Resequencing Improved Accuracy

Variant Reporter Software includes improved algorithms that have been trained on large data sets to yield the most accurate and sensitive variant discovery, which is ideal for rare mutant detection.

Software Available

Variant Reporter Software effortlessly analyzes up to ten times more trace files than competitive products, a unique feature that is highly valued by researchers with large projects. Through the use of advanced algorithms and quality metrics, project review time can be slashed by up to 97%. And because the software organizes projects by amplicons, the time required for computing alignment and variant detection is dramatically reduced. Thus, the days of having to break large projects into smaller ones because the software cannot accommodate complexity are now over.

Resequencing Applications

Resequencing—also known as comparative, direct, medical, or PCR sequencing—is useful for discovering variants, including HIMs, insertions, deletions, SNPs, and genotypes. The process begins when a researcher chooses a gene of interest, isolates DNA from the gene, amplifies one or more amplicons for each exon, and then performs cycle sequencing using the BigDye® Terminator Cycle Sequencing Kit, followed by cleanup with the BigDye® XTerminator™ Purification Kit. The sample is then run on an Applied Biosystems capillary electrophoresis instrument. During the final step, Variant Reporter Software discovers the variants, determines the genotypes, and creates a report. That's all there is to it.

Simple Workflow, Accurate Results

The workflow for Variant Reporter has been designed to be amenable to a range of project sizes and types.

1. Import and Assign Traces

To set up a project, simply import and group traces into amplicons and specimens. The analysis parameters can be adjusted and a reference defined, although the data can be analyzed with or without a reference. The software automatically groups traces into specimens and amplicons based on sample file name parsing (Figure 3).

Analysis Parameters (Optional)

Variant Reporter Software is also highly flexible. Researchers who choose not to use the recommended analysis parameters have the option to customize the settings as desired.

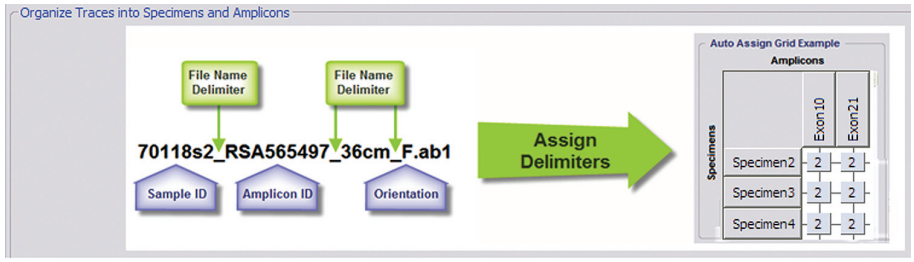

Figure 3. Easy project set up

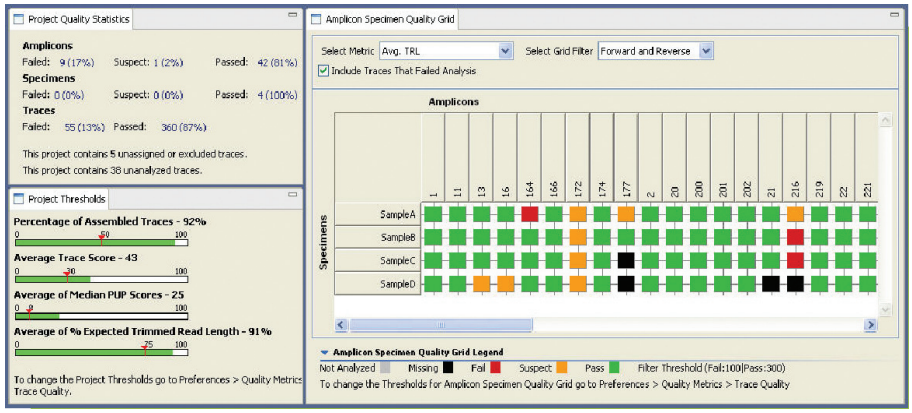

Figure 4. Dashboard of project quality metrics

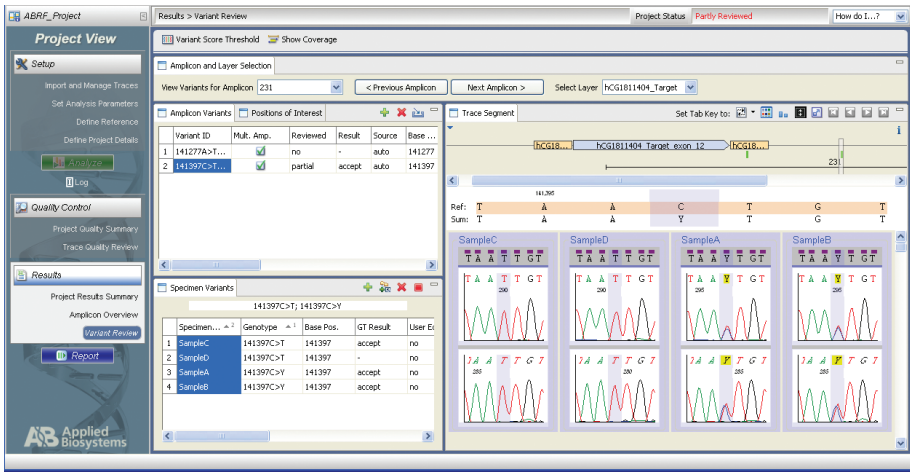

Figure 5. Single interface for reviewing variants with targeted electropherogram view

TABLE 1: SCALE UP YOUR PROJECT WITHOUT A CORRESPONDING INCREASE IN ANALYSIS TIME

Speed of Analysis

Project Size	Variant Reporter Software* (minutes)
814 Traces (51 amplicons)	1:26
5000 Traces (51 amplicons)	4:42

*Pre-basecalled data

Built-in trimming and filter features enable the software to remove low-quality traces.

- Improvements in the KB™ Base-caller algorithm improve accuracy and quality value assignment for mixed base values.
- Flexible trimming enables the exclusion of amplicon primer sequences and allows various trimming parameters to be set for the 5′ and 3′ ends.
- New metrics for filtering traces include the "Peak under Peak Score" and the "Percentage of Expected Trimmed Read Length".

Set a Reference (Optional)

Set a sequence as the reference for comparison. Allowed file types include Genbank, .ab1, .seq, and fasta. For quick projects, you can analyze without specifying a reference sequence.

2. Analysis

Analyze your project.

3. Quality Control (as needed)

After analyzing the data, the software calculates the project-quality statistics and generates an overview of the project data quality (Figure 4). If samples pass QC thresholds, the software will skip this step. You can also drill down to review the quality of specimens, amplicons, or individual traces. The software contains all the viewing and reporting functionalities of Sequence Scanner Software.

4. Review and Edit Variants/Genotypes

The analysis algorithms in Variant Scanner Software allows you to set a variant score threshold for reviewing variants (Figure 5). The software presents a list of potential variants in tabular formats, one amplicon at a time.

- You can view variants, edit, and accept or reject amplicon variants or specimen variants.
- The analysis provides variant scores for variant evaluation.
- In addition to the traditional scrolling trace view, user can view traces as snippets (e.g., a variant in a 7-bp segment) or in an extended view (e.g., a variant in a 25-bp segment) to enable quick viewing of multiple genotypes per page.

Reports

You can select from the following reports and exports:

- Project Summary Report: Contains high-level project statistics, lists of variants and genotypes, and snippets of each variant.
- Quality Report: Provides a comprehensive view of trace, specimen, amplicons, and project quality.
- Specimen Report: For clinical researchers interested in reporting data one patient at a time, this report provides specimen statistics, specimen genotypes, and snippets of each variant for all specimens in a project.

Conclusion

Variant Reporter Software offers a simple workflow with easy-to-use navigation, organization, and variant confirmation. Targeted algorithms and improved quality metrics dramatically reduce the time spent on data review. The software enables researchers to study complex diseases across multiple genes and create projects of >4000 trace files. Download a demonstration copy and see results in less than an hour.

ORDERING INFORMATION

Product Description	Quantity	P/N
Variant Reporter™ Software v1.0	Initial license	4385261
Variant Reporter™ Software v1.0	1 additional license	4385262
Variant Reporter™ Software v1.0	2 additional licenses	4385263
Variant Reporter™ Software v1.0	3 additional licenses	4385264
Variant Reporter™ Software v1.0	4 additional licenses	4385265
Variant Reporter™ Software v1.0	5 additional licenses	4385267
Variant Reporter™ Software v1.0	10 additional licenses	4385268
Variant Reporter™ Software v1.0	1 additional copy (no documentation)	4385269
Variant Reporter™ Software v1.0 Demo	Demonstration CD (30 days)	4385270

Bundle and Save

When purchased along with an Applied Biosystems capillary electrophoresis instrument, Variant Reporter Software v1.0 can be bundled cost-effectively with our other sequencing software packages in the following three combinations:

1. Variant Reporter and Sequencing Analysis Software (a combination that significantly improves accuracy)
2. Variant Reporter, Sequencing Analysis, and GeneMapper® Software
3. Variant Reporter, Sequencing Analysis, and GeneMapper® *ID* Software

Alternatively, Variant Reporter Software can be purchased separately. Talk to your Applied Biosystems sales representative about which combination makes the most sense for your research focus. (Terms and conditions may apply.)

Visit **www.appliedbiosystems.com/support/software/variant_reporter/VariantReporterDemo.exe** to download a FREE, 30-day, trial copy of Variant Reporter Software and/or call your local sales representative to find out more.

Printed in the USA. 05/2007 Publication 133PB03-02

Applied Biosystems

Headquarters
850 Lincoln Centre Drive | Foster City, CA 94404 USA
Phone 650.638.5800 | Toll Free 800.345.5224
www.appliedbiosystems.com

International Sales
For our office locations please call the division headquarters or refer to our Web site at
www.appliedbiosystems.com/about/offices.cfm