VariantStudio Software Release Notes

Version 3.0.12

Introduction

These Release Notes detail the key changes to software components for VariantStudio since the package containing 2.2.3.

Refer to the user guide for more details on how to operate VariantStudio:

http://support.illumina.com/downloads/variantstudio_userguide.html

The FAQs for the application are available at:

http://support.illumina.com/sequencing/sequencing_software/variantstudio/questions.html

The software components include:
- Illumina VariantStudio v3.0.12
- BaseSpace Annotation Engine Database v1.4.2.60

I. Import

New Features:

- VariantStudio v2.x Projects are not supported. Samples from these projects must be reimported and annotated using VariantStudio v3.0.

Defect Repairs:

- Previously, restricting import of variants to only those that overlap genes in a user-specified gene list resulted in failure to import variants that overlap any specified genes represented as lower case or mix of lower case like C10orf2. Now variants are properly imported, regardless of whether the genes they overlap are represented as upper or lower case.

Known Issues:

- When loading extremely large vcf.gz files, the import progress bar may not initially update.

II. Annotation

New Features:

- Variant annotation is performed locally using BaseSpace Annotation Engine and no longer requires internet connection. For information on installing the annotation database, refer to the User Guide.
- Annotation source database versions:
    - VEP, Version: 84, Release Date: April 29, 2016
    - UCSC phyloP, Version: hg19, Release Date: November 10, 2009
    - dbSNP, Version: 147, Release Date: June 01, 2016
    - COSMIC, Version: 77, Release Date: April 07, 2016
    - 1000 Genomes Project, Version: Phase 3 v5a, Release Date: May 27, 2013
    - EVS, Version: 2, Release Date: November 13, 2013
    - ExAC, Version: 0.3.1, Release Date: March 16, 2016
    - ClinVar, Version: unknown, Release Date: July 05, 2016

- 1000 Genomes Asian population allele frequency has been separated into 2 columns: East Asian and South Asian
- Conservation score column displays actual PhyloP score instead of "yes/no" value. PhyloP scores measure evolutionary conservation at individual alignment sites. The scores are interpreted as compared to the evolution expected under neutral drift.
    - Positive scores—Measure conservation, which is slower evolution than expected, at sites that are predicted to be conserved.
    - Negative scores—Measure acceleration, which is faster evolution than expected, at sites that are predicted to be fast-evolving.
- Updated link for PubMed lookup

DEFECT REPAIRS:

- None

KNOWN ISSUES:

- For single nucleotide deletions, the annotation data for rsID, read depth, and sample coverage used with EVS comes from the following genomic position (variant position+1) as opposed to the variant position. Because everything is encoded in "VCF coordinates", the actual deletion takes place at the following base.
- A few rsIDs were inadvertently removed due to the way entries are matched that likely contain assembly artifacts. rsID entries overlapping genes with the phrase "CFL" contained in them are affected.
- The Load Default Transcripts feature does not allow the user to apply as default any transcript that is not in the predefined list of known gene-transcript sets in the software (as listed in the Load Default Transcripts window). If an unknown transcript is imported and set as default, the transcript is ignored.

III. Filtering

NEW FEATURES:

- Variant filtering based on:
    - PhyloP scores
    - ExAC population allele frequency

DEFECT REPAIRS:

- None

KNOWN ISSUES:

- When Creating or Modifying an Advanced Filter through the text expression form of the filter, using the "!" character next to expressions enclosed in parentheses has the effect of negation, but this effect is not reflected in the graphical tree representation of the filter. Use the "not equals" operator in these situations. Using a "!" character outside of the square brackets, however, does not have the effect of negation; when set, it can only be cleared by using the Clear Filters button in the main Filters panel.
- The filter history shows the filter name for any saved filters. However, the name is displayed in the filter history only when an existing filter is reused. The saved name is not displayed in the "Filter Name" column when the filter is initially created and saved, only when it is reused.

IV. Classification

NEW FEATURES:

- None

DEFECT REPAIRS:

- None

KNOWN ISSUES:

- None

V. Reporting

NEW FEATURES:

- Variants occurring in multiple samples within the project can be exported to a TSV file by clicking the Shared Variants button under the Reports menu.
- Ability to select a sample in the Sample Report Generator.

DEFECT REPAIRS:

- None

KNOWN ISSUES:

- None

VI. Outputs

NEW FEATURES:

- None

DEFECT REPAIRS:

- None.

KNOWN ISSUES:

- The following report-exporting processes cannot be cancelled: Filtered Variants, All Transcripts for Variants, Filter History, Sample Report in PDF format, and Sample Report in RTF format. Genes table and the No-call Regions table report exporting can be cancelled. Non-cancellable exporting processes do not show a progress bar for the process.

VII. Additional Features

NEW FEATURES:

- The View All Samples button displays data for all samples in the project.
- Mouseover on variant grid columns displays description of column content.
- Display scaling is checked on application startup, with an informational notice to set scaling factor to 100% for optimal screen display.

DEFECT REPAIRS:

- None

KNOWN ISSUES:

- If the user zooms too far when zooming in or out of the Gene View, an error is thrown and the Gene View becomes disabled. Under normal usage, the zooming limit is not expected to be reached. The loss of the Gene View has no impact on continued use of the rest of the application.
- Transcript column counts all transcripts present in the User Interface. Transcripts that are annotated as either upstream or downstream gene variants are included in this count.
- The “Add Variants to Sample” import option does not limit importing additional variants from the sample multiple times. It is possible to import the same variants multiple times and have duplicate entries in the User Interface.
- If a workspace is overwritten, the existing *.vbp file is deleted, but all other files in the subfolder remain.
- During application startup, if the user locks the screen or switches users, VariantStudio may throw an exception and close.
- The Distance column (hidden by default) sorts by the string value of the field, rather than as a number.
- When loading a VCF with " at the beginning or end of the Alt value, the variant is loaded as-is, with the Type listed as "complex".