VariantStudio Software Release Notes

Version 2.2.3

September 18, 2015

Introduction

These Release Notes detail the key changes to software components for VariantStudio since the package containing version 2.2.2.

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 can be found at this location:

support.illumina.com/sequencing/sequencing_software/variantstudio/questions.ilmn

The software package includes:
- Illumina VariantStudio v2.2.3

I. Import

NEW FEATURES:

- Improved the .ant loading process bar to give a more accurate representation of progress.

DEFECT REPAIRS:

- Fixed issues related to prematurely terminating import of .ant files.

- Previously, loading a VCF with " at the beginning or end of the Alt value, the variant is loaded as-is, with "Type" of "complex". Now variants containing quotes are ignored and not imported into VariantStudio

KNOWN ISSUES:

- If the sample import process is canceled during a multi-select import or a folder import, the samples that were successfully imported before cancelation may not immediately appear in the Variants table and the Current Sample menu. If so, then the recommendation is to save, close, and reopen the project. The samples should then be visible. Alternatively, the user may add or remove a sample to produce the same effect.

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

II. Annotation

NEW FEATURES:

- None

DEFECT REPAIRS:

- Repaired a known bug where variants were assigned as “Polyphen Damaging” and “SIFT Deleterious” when the Polyphen and SIFT columns are blank.

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 pre-defined list of known gene-transcript sets built in to the software (those variants that show up in the table in the Load Default Transcripts window). If a user tries to change the default transcript to one that is not in this list by using the Import feature, the unknown transcript is ignored.

III. Filtering

NEW FEATURES:

- None

DEFECT REPAIRS:

- Modified the filtering behavior so that gene name filtering can be performed without first annotating the VCF.

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, and when it is set in place, the character can only be cleared by using the Clear Filters button in the main Filters panel.

IV. Outputs

NEW FEATURES:

- None

DEFECT REPAIRS:

- Transcript export files are now deleted if they were generated after cancelling transcript export

KNOWN ISSUES:

- None

V. Additional Features

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 limits should not 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 counted 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.

- During application startup, if the user locks the screen or switches users, VariantStudio may throw and exception and close.