BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.11175

September 13, 2016

Introduction

These Release Notes detail new features, known and recently resolved issues for the BaseSpace Variant Interpreter Beta Testing software. For details on how to operate BaseSpace Variant Interpreter Beta Testing software, see the online help, which is available from the Help icon in the application.

NEW FEATURES:

- Variant grid improvements:
  - "Algorithms" is now called "In Silico Predictions" in the variant grid.
  - "Add Interpretation" is now called "Case Interpretation" in the variant grid.
  - The "Add to report" button is no longer active until the variant has been assigned an interpretation.
  - Gene names shown in the germline sample variant grid are now hyperlinked to the OMIM gene symbol search page (http://omim.org).
- Metadata improvements:
  - Mandatory metadata fields are now indicated with a red asterisk (*).
  - A specific error message is now shown if the Sex field is left blank.
  - The metadata panel can now be temporarily minimized when looking at the variant grid.
  - "Add Subject" is now called "Edit Subject/Sample" in the sample metadata form.
- Filtering improvements:
  - Variant consequence filters can now be selected with a single click per group.
  - The variant grid now contains a link to pop up a list of active filters applied to the current view.
  - Zygosity and Prediction variant filter options are no longer shown for tumor samples.
- Reporting improvements:
  - The Illumina logo is no longer present on the PDF report.
  - Report comments can now be edited only by the original creator of the comment.
- General user interface updates:
  - "Add Analysis Result" is now called "Import Analysis File (.VCF)".
  - "Import from BaseSpace" is now called "Import from BaseSpace Sequence Hub".
  - The Workgroup Administration link now opens in a new tab.
  - The BaseSpace Sequence Hub link now opens in a new tab.

- The system now provides an explanation for required reanalysis of samples following a forced upgrade or new custom annotation.
- The Settings page now indicates which workgroup context a user is operating in.
- During assignment of interpretation for somatic variants, "L1: FDA Guidance" is now known as "L1: National Guidance".
- The online user guide now contains a direct link to contact Illumina Technical Support.
- Release notes are now available directly in the application.

Resolved Issues:

Issue Key	Issue Category	Description
BSVI-16985	Import	Sample manifest now points to the appropriate URL for sample import.
BSVI-16777	Import	Resolved an error that intermittently prevented large VCF files from being imported.
BSVI-16722	Variant Grid	Cosmic links now show consistently.
BSVI-16691	Login	Users are now redirected to the Terms of Use if they navigate to the End User License Agreement link.
BSVI-16520	Variant Grid	Updated germline prediction field to show only a single transcript.
BSVI-16401	Login	The system no longer crashes when multiple browser types are open (such as Firefox and Chrome) at the same time.
BSVI-16364	Variant Grid	The autocomplete field for phenotype terms now shows a scrolling result of up to 50 results.
BSVI-16144	Import	After manifest upload, auto-reanalyze no longer leaves analysis results in an error mode.
BSVI-16025	Report	Somatic variant frequency values on the draft report now match exactly to the four decimal places shown in the variant grid.
BSVI-15920	Import	VCF files with mixed chr format are no longer ingested without errors.

Known Issues

Issue Key	Issue Category	Description
BSVI-17165	Walkme	Walkme tutorial guide is currently disabled.

Issue Key	Issue Category	Description
BSVI-16523	Filters	A saved hg38 region-based filter can be incorrectly viewed in the saved filter drop down menu, when analyzing a hg19 sample.
BSVI-9300	Annotation	BaseSpace Variant Interpreter Beta does not validate the search criteria of a chromosome as a user enters an actual position.
BSVI-11929	Annotation	If a user tries to upload a gene, annotation, or region list with a file name longer than 50 characters, the Enter Annotation Label field appears red. BaseSpace Variant Interpreter Beta requires that these file names be ≤50 characters long.
BSVI-10013	Classification (Family-Based Analysis)	Variants without a canonical transcript are sorted out of order for prediction.
BSKN-2898	Add New Association	Clinical Trial open and close dates can appear one day earlier depending on which time zone the association was created in.
BSKN-2815	Add New Association	Clinical Trial curator summary is duplicated from the evidence summary section.
BSKN-2194	Add New Association	Publication Id allows non-integers to be used and creates an erroneous link to the PubMed website.
BSVI-12422	Import	If a user tries to upload a VCF file that is not properly formatted, the upload fails with an ambiguous error message. The file shows a status of fail, and the user can then delete it. Consequently, the variant grid does not appear.
BSVI-16408	Import	Upload of Grch38 .vcf files will occassionally result in error. Deleting the failed upload and retrying will clear the error.
BSVI-16384	Variant Details	ClinVar data for the same variants can appear to differ between the variant summary in the grid (which reports both SCV and RCV entries) and the variant detail page (which reports only RCV).
BSVI-11278	Registry	If user enters a partial name and then changes it, both names become part of autopopulate. A user must completely clear the text box to see both autocomplete options.
BSVI-13331	Registry	When using the sample metadata sheet to simultaneously upload 40 or more cases, the software might freeze.
BSVI-11052	Report	The inheritance mode content extends off the report page.
BSVI-15768	Variant Grid	For variants with multiple dbSNP IDs, only two will be displayed in the variant grid.
ON-345	Variant Grid	Autocomplete box shows results marked as 'obsolete' by nomenclature authority.

Issue Key	Issue Category	Description
ON-432	Variant Grid	Expansion to related phenotypes sometimes yields results that are too distant.
ON-516	Variant Grid	Results in autocomplete pop-up might be sorted inconveniently (subjective)
WAC-546	Workgroups	Inviting a user who is not currently registered will send an invitation email and is user marked as pending. This is not visible in the user interface. If users would like to guarantee that a workgroup member has been notified, they can contact the invitee directly independent of Variant Interpreter (Beta).