BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v.1.0.16757

April 6, 2017

INTRODUCTION

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

NEW FEATURES

- **Knowledge Network - Curation Portal**

  The curation portal provides a way to view all content from your private workgroup. In addition, you can search for relevant associations by gene name, transcript ID (including transcript equivalence), or variant ID (including genome equivalence) in both your private workgroup and Knowledge Network.

  - A status board enables you to quickly access associations in various stages of the curation process.
  - The Curation Manager can track curations that are In Progress, Ready for Review, Enabled, or Disabled.
  - You can access any workgroup in which you are a member.
  - The curation portal can be accessed at https://knowledgenetwork.kn.basespace.illumina.com/bskn

- **Platform Ontology Service**

  Our Ontology Service now includes functionality to look up genes by symbols and full names (autocomplete, type-ahead) and to retrieve positional information on transcripts and exons.

  - Gene name type-ahead for HGNC genes.
  - Support for RefSeq and Ensembl gene and transcript models.
  - Support for gene/transcript/exon positions for GRCh37 and 38.
  - Integration with Illumina Annotation Engine.

- **Variant Inheritance**

  The variant grid for SNPs and InDels has been enhanced by the introduction of a visual representation of variant inheritance enabling rapid triage of variants. The visual inheritance representation is only visible for RUGD cases with SNPs and InDels in noncompact mode.

- **Compact Variant Grid**

  The Compact Variant View provides functionality to view variants in a compact form by hiding detailed information while displaying general information useful for performing most preliminary variant filtering. Individual variants can be expanded or collapsed to provide additional detail or this action can be performed on all variants within a tab.

- **Report Templates**

  Variant Interpreter now supports the creation and application of Report Templates allowing users to customize templates for RUGD and Oncology reports. Reports now support:

  - Renaming sections within a report template.
  - Saving fixed text to sections within a report template.
  - Automatic omission of empty fields/sections for a cleaner PDF report.

- **Mutational Signature Plot**

  For Oncology cases, a new mutation signature plot displays the predicted mutational signatures defined by COSMIC for a particular tumor sample. This plot includes the mutational context profile for a particular sample showing the distribution of substitutions across the sample with a 3mer context and decomposition of signatures visualized in a stacked barplot and table with probable associations. Note: This feature is optimized for whole genome sequencing analysis workflows only, and will not work if the total number of variants called is below 200.

- **Dynamic CNV and SV Visualization**

  For cases with CNV and SV data, a new dynamic visualization displays as a component within the variant grid. Users can access the visualization through the “Show Visualization” button allowing interactive investigation of the CNV and SV data through:

  - Pan, zoom, and gene search controls
  - Interactive variant information by clicking variants within the visualization
  - Structural variant track displays arcs between structural variant breakends. Translocation breakends are shown with the source breakpoint aligning with the bottom axis and the target breakpoint aligning with the top axis.
  - Copy number coverage track displays average coverage across genomic segments with supporting datapoints shown superimposed on the background.
  - Copy number bi-alleles track displays major and minor allele differences.
  - Display of genes within an overlapping genomic region through a Gene Finder panel.

- **Export to TSV**

  The variant grid now enables some basic export of up to 1000 variants to a tab-separated value (TSV) file. This includes some details provided by the variant caller and annotation engine.

- **Case Workflow Management**
  Variant Interpreter now introduces four case statuses that can be assigned to a case along with a case owner. These statuses, in order of case progress, are: Pending Review, In Progress, Awaiting Approval, and Closed. A case can be moved backwards or forwards through these states until case closure. When a case is closed, a PDF report is generated and made available on the case history page. To reopen and begin working on a closed case, move the case from Closed back to In Progress.

Resolved Issues

Issue Key	Issue Category	Description
WAC-546	Workgroups	Invited users who are not currently registered are sent an invitation email and are marked as pending

Known Issues

Issue Key	Issue Category	Description
BSKN-3475	Associations	Importing associations from BSKN for retired ontology terms require users to select a current ontology term.
BSVI-22331	Interpretation	A limited list of genes are displayed for Copy Number and Structural Variant interpretation
BSVI-24043	Case Management	When a case is in a non-actionable state (Failed, Action Required, etc), the case needs to be re-analyzed or re-uploaded prior to updating the case status or owner.
BSVI-23156	Case Management	A case cannot be closed without first viewing the draft report.
BSVI-23521	Variant Grid	Mode of inheritance calculation is occasionally inaccurate if family members are removed from original pedigree.
BSVI-23555	Variant Details	The "Back to Case" button does not take the user back to the same tab on the variant grid.
BSVI-22349	Case Registry	Users may have recently experienced failed VCF uploads that require re-analysis or re-upload. Please contact support if re-uploading these VCFs is not successful.
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	Uploading of multiple Grch38 .vcf files occasionally results in an error. The error can be cleared by deleting the failed upload and retrying.
ICL-719	Allele frequencies	Some allele counts and total allele counts are off for sex chromosomes. For indels, these can be remediated by sending the CaseLog VID instead of the annotation engine VID.
ICL-723	Allele frequencies	GRCh38 PAR regions are not handled correctly when computing allele frequencies. GRCh37 PAR regions are handled correctly.
ON-345	Variant Grid	Autocomplete box shows results marked as 'obsolete' by nomenclature authority.
ON-432	Variant Grid	Expansion to related phenotypes sometimes yields results that are too distant.

Issue Key	Issue Category	Description
ON-516	Variant Grid	Results in autocomplete pop-up might be sorted inconveniently (subjective).
ON-636	Gene lookup	API should give back an appropriate error message when a given parameter value does not exist.
ON-639	Gene lookup	Gene auto-complete API: inconsistent sorting behavior for returned lists of matching genes.
BSVI-21304	Variant Grid	Predicted pathogenicity pop-up may show null transcript.
BSVI-21855	Variant Grid	VCF name truncation to analysis result name may not match expectation (subjective).
BSVI-24136	Visualization	The mutational signature plot is optimized for WGS, however there is no messaging to the user that analysis on any other platform may yield inaccurate findings or why a plot is not generated for any case with less than 200 SNVs.
BSVI-24137	Visualization	The dynamic visualization is located separate from the visualization panel, which may be misleading to users.
WAC-546	Workgroups	Invited users who are not currently registered are sent an invitation email and marked as pending, but their status is not visible in the user interface. To confirm that the invitation has been received, users can contact the invitee directly, independent of BaseSpace Variant Interpreter (Beta).
WAC-730	Workgroups	In some instances, the bulk edit of user permissions checkboxes does not work. Log out/log in to resolve this, or edit users individually.
WAC-767	Workgroups	Workgroup Administrators cannot revoke a pending invitation at this time, however Workgroup Administrators can still remove members from the workgroup.