BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.16980

April 13, 2017

Introduction

These Release Notes detail new features, known issues, 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.
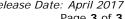
NEW FEATURES

No new features in this release.

RESOLVED ISSUES

Issue Key	Issue Category	Description
BSVI-24190	Import	When importing a WGS Tumor/Normal App Result from BaseSpace Sequence Hub, BaseSpace Variant Interpreter does not ingest all the supplemental files.

KNOWN ISSUES

Issue Key	Issue Category	Description
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.
BSKN-3475	Associations	Importing associations from BSKN for retired ontology terms require users to select a current ontology term.
BSVI-23156	Case Management	A case cannot be closed without first viewing the draft report.
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-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.
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.

Issue Key	Issue Category	Description
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.
BSVI-22331	Interpretation	A limited list of genes are displayed for Copy Number and Structural Variant interpretation
BSVI-23555	Variant Details	The "Back to Case" button does not take the user back to the same tab on the variant grid.
BSVI-23521	Variant Grid	Mode of inheritance calculation is occasionally inaccurate if family members are removed from original pedigree.
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.
ON-516	Variant Grid	Results in autocomplete pop-up might be sorted inconveniently (subjective).
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.