

# BaseSpace Variant Interpreter Beta Release Notes

**BaseSpace Variant Interpreter Beta v1.0.17488**

**May 1, 2017**

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## 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

- **Structural Variant Support**

For Tumor-Normal cases, the variant grid now supports displaying normal paired and split-read support in a pop-up. For all cases, filtering by paired and split-read support is now enabled.

- **Custom Annotations**

Variant Interpreter now supports multiple custom annotations within a category associated to a variant, layered custom annotations with associated subannotations, and support for BBCode format URL linking.

- **Penetrance and Tracking of Validation Status**

Improved options for recording variant penetrance information for a variant; available selections are Complete, Incomplete, or None. Increased the number of method options for recording validation methods.

- **Case Workflow Management**

Users can now assign multiple cases to one owner, saving time for case management workflows.

## RESOLVED ISSUES

Issue Key	Issue Category	Description
<b>BSVI-23156</b>	Case Management	A case cannot be closed without first viewing the draft report.
<b>BSVI-22331</b>	Interpretation	A limited list of genes is displayed for Copy Number and Structural Variant interpretation
<b>BSVI-21304</b>	Variant Grid	Predicted pathogenicity pop-up may show null transcript.
<b>ICL-719</b>	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.
<b>ICL-723</b>	Allele Frequencies	GRCh38 PAR regions are not handled correctly when computing allele frequencies. GRCh37 PAR regions are handled correctly.
<b>WAC-546</b>	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).

## KNOWN ISSUES

Issue Key	Issue Category	Description
<b>BSKN-3475</b>	Associations	Importing associations from BSKN for retired ontology terms require users to select a current ontology term.
<b>BSKN-24939</b>	Audit Log	Validation status is not currently stored in the audit log.
<b>BSVI-24043</b>	Case Management	When a case is in a nonactionable state (Failed, Action Required, etc), the case needs to be reanalyzed or reuploaded prior to updating the case status or owner.
<b>BSVI-22349</b>	Case Registry	Users may have recently experienced failed VCF uploads that require reanalysis or reupload. Contact support if reuploading the VCF files is not successful.
<b>BSVI-24943</b>	Exports	Exported population frequencies for 1000 Genomes data are only for all populations.
<b>BSVI-24942</b>	Exports	Exported Clinvar Variants included non allele-specific RCVs.
<b>ON-636</b>	Gene lookup	API should give back an appropriate error message when a given parameter value does not exist.
<b>ON-639</b>	Gene lookup	Gene autocomplete API has inconsistent sorting behavior for returned lists of matching genes.
<b>BSVI-12422</b>	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.
<b>BSVI-16408</b>	Import	Uploading of multiple Grch38 .vcf files occasionally results in an error. The error can be cleared by deleting the failed upload and retrying.
<b>BSVI-23555</b>	Variant Details	The "Back to Case" button does not take the user back to the same tab on the variant grid.
<b>BSVI-23521</b>	Variant Grid	Mode of inheritance calculation is occasionally inaccurate if family members are removed from original pedigree.
<b>ON-345</b>	Variant Grid	Autocomplete box shows results marked as 'obsolete' by nomenclature authority.
<b>ON-432</b>	Variant Grid	Expansion to related phenotypes sometimes yields results that are too distant.
<b>ON-516</b>	Variant Grid	Results in autocomplete pop-up might be sorted inconveniently (subjective).
<b>BSVI-21855</b>	Variant Grid	VCF name truncation to analysis result name may not match expectation (subjective).
<b>BSVI-24935</b>	Variant Grid	Applied Structural Variant filters do not show in the filter summary. View the filter dialog instead to verify.

Issue Key	Issue Category	Description
<b>BSVI-24136</b>	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.
<b>BSVI-24137</b>	Visualization	The dynamic visualization is located separate from the visualization panel, which may be misleading to users.
<b>WAC-730</b>	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.
<b>WAC-767</b>	Workgroups	Workgroup Administrators cannot revoke a pending invitation at this time, however Workgroup Administrators can still remove members from the workgroup.
<b>HAD-24655</b>	Zygoty	Zygoty text for variants on sex chromosomes does not account for pseudoautosomal regions
<b>HAD-24656</b>	Zygoty	Pedigree diagrams reflect familial inheritance only for "PASS" variants