BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v.1.0.15015

February 20, 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

- **Common Interpretation Dialog**

  Interpretation and curation process have been streamlined. To provide a consistent user experience and to provide access to curated knowledge during interpretation, the following enhancements have been made:

  o Interpretation is now an expandable section inside the Variant Details page, which is accessible by clicking the Case Interpretation button.
  o Context-sensitive content in the variant grid opens to the relevant section of the Variant Details page.
  o A Back to Case Page button is added to the Variant Details page. Selecting this button returns to the corresponding variant grid row on the Case page.

- **Past Cases**

  You can now view information from prior interpretations made for the same variant in other cases within a workgroup. Information regarding past cases where the same variant has been interpreted is shown in the Association column of the variant grid and the Past Cases section under the Variant Details pages. The Past Cases section provides links to related cases with additional information about the variant. This feature is currently only available for SNPs and InDels.

  Past Cases Features:

  o For RUGD, interpretations from past cases for a variant are shown as chiclets on the variant grid, distributed by pathogenicity call.
  o For Oncology, if past interpretations exist for a variant, they are shown as a single value (L1 to L4) corresponding to the worst interpretation.
  o Variant Prioritization:
    - For RUGD, variants are prioritized on the variant grid if they have worse pathogenicity recorded compared with the default pathogenicity prediction.
    - For Oncology, variants that have prior interpretations are prioritized over variants without prior interpretation information.
  o Clicking the Past Cases header on the variant grid links to the Variant Details page with the Past Cases section highlighted.

RESOLVED ISSUES

Issue Key	Issue Category	Description
BSVI-21634	Variant Grid	Comment editing and deletion is temporarily unavailable

KNOWN ISSUES

Issue Key	Issue Category	Description
BSKN-4211	Associations	Older curations that are missing mutation class cannot be edited into a personal Knowledge Base
BSKN-3787	Associations	ClinVar SCV associations are not displayed as children of related RCV associations.
BSVI-22349	Case Registry	Some VCF uploads may fail and 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.
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).
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.

Issue Key	Issue Category	Description
WAC-767	Workgroups	Workgroup Administrators cannot revoke a pending invitation at this time, however Workgroup Administrators can still remove members from the workgroup.