BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v.1.0.14577

February 2, 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

- **Interpretation**

  New optional fields are provided to capture information during variant interpretation for both Germline and Oncology cases.

  - For Germline cases, the contribution of a variant to subject phenotypes can be recorded. The contribution may be recorded as Full, None, or as a specific list of phenotypes from the subject. Contribution information is included when a variant is added to the draft and PDF report. A warning is displayed if a user attempts to remove a subject phenotype that is used to indicate partial contribution for a variant.

  - For Oncology cases, the association type (eg, "Therapy Predictive", "Prognostic", or "Diagnostic") for a variant can be captured during interpretation. Association type is displayed on the draft and PDF reports for a reportable variant.

- **Higher Order Curation**

  Evidence-based associations can have interpretations at several levels when considering Single Nucleotide Variants (SNVs) and Multiple Nucelotide Variants (MNVs). BaseSpace Knowledge Network can now support biomarkers at 3 curation levels:

  - **Amino Acid** curation enables biomarker content at the protein level. These alterations, frequently referred to in publications and observed in disease conditions, can support a wide range of nucleotide changes.

  - **Codon** curation provides an opportunity to add content from public sources on cohort analysis, grouping clinical observations at a specific protein location.

  - **Exon** curation, in combination with mutation classes, provides robust content which matches functional protein alterations.

RESOLVED ISSUES

Issue Key	Issue Category	Description
BSVI–19408	Audit Log	When archiving an interpretation, archived status is not saved in audit logs.
BSKN-3868	Curation Portal	Gene-level curation associations should not include fields for exon, amino acid or consequence data.

KNOWN ISSUES

Issue Key	Issue Category	Description
BSKN-3787	Associations	ClinVar SCV associations are not displayed as children of related RCV associations.
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).
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.