BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.13548

December 8, 2016

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

- **Higher Order Curation**

  Evidence-based associations can have interpretations at several levels when considering SNVs. Nucleotide-level curation provides a specific alteration, which is frequently observed in the disease condition or mentioned in the evidence. Gene-level curation combines a gene with a mutation class(es). Multiple mutation classes can be used to refine the association to match the evidence. BaseSpace Knowledge Network can support both nucleotide and gene level curation.

- **Flag variants for review**

  Variants of interest can now be bookmarked for later review and/or for bringing to the attention of a co-worker analyzing the same case. Every variant row now contains a bookmark icon on the interpretation column that can be checked to flag the variant. A bookmark counter on the header line maintains a live count of the total number of variants that have been flagged for review. The user can sort these variants to the top of the grid by clicking on the bookmark counter.

- **Usability enhancements for phenotypes**

  In order to provide users with a uniform and consistent user experience while entering case phenotypes, we have made the following enhancements:
  - Phenotype and Indications fields have now been merged and duplicate values entered for these fields have been removed.
  - Phenotype entry is now under ontology control (HPO and SNOMED). Matching phenotypes are suggested to the user as they begin typing in a term thereby ensuring accurate and quick data input.
  - Legacy phenotype terms have not been modified and are still available for the user and are suffixed with “(other)” to distinguish these from ontology controlled terms.
  - New phenotype terms link to the relevant ontology resource where more information about the term can be found. These links are available from both the Case Details and the Subject Details page (in non-edit mode).

- **Workgroup Administration**

  Workgroup administrators can now modify access to BaseSpace Sequence Hub. Pending invitations for applications in the BaseSpace Suite are now visible, and invitation emails can be re-sent.

Resolved Issues

Issue Key	Issue Category	Description
BSKN-2898	Associations	Clinical Trial open and close dates can appear one day earlier depending on which time zone the association was created in.
BSKN-3612	Associations	Genome equivalence maps associations between GRCh37 and GRCh38 positions. There is a known issue when a batch process fails and some mapping values are missing.
BSKN-3628	Curation Portal	In the curation portal, the “Last Updated” field is not populated when a user searches for a variant ID that is included in both private and public data.
BSVI-18409	Variant Grid	The validation message for applying Tumor-Normal specific filters to a Germline analysis result is not displaying.
BSVI-18942	Variant Grid	1000 genomes allele frequency information is temporarily unavailable, filters are not affected.

Known Issues

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

Issue Key	Issue Category	Description
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.