BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.12680

November 21, 2016

FOR RESEARCH USE ONLY

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

- Genome Equivalence support

  Variants in BaseSpace Knowledge Network are now stored universally, regardless of genome build. This allows users to easily match variants and associations seamlessly for different samples, as multiple samples uploaded to Variant Interpreter can be from different genome builds. Specifically, users now have the ability to add associations for one genome build (eg GRCh37), process samples for a second genome build (eg GRCh38), and match variants to associations for either.

- Import Associations from Knowledge Network to a private database

  The BaseSpace Knowledge Network contains associations from several sources. These associations are publicly available and can include evidence to support pathogenicity or direction calls for variants. Members of a workgroup can now import these associations, along with all the evidence, to a private knowledge base. Users who are logged into the workgroup Knowledge Base can edit, expand, and review the imported associations.

- Other updates
  - Users are able to only apply saved filters that had been saved in the same genome build.
  - Clinvar associations are expandable to show details of Clinvar association data that exists in BaseSpace Knowledge Network.
  - The Case metadata look and feel has been improved.
  - Privileged users are now able to delete cases that are in a pending processing state.
  - Workgroup owners can export workgroup-level audit logs.

Resolved Issues

Issue Key	Issue Category	Description
BSVI-15768	Variant Grid	For variants with multiple dbSNP IDs, only 2 of the IDs are displayed in the variant grid.
BSVI-16384	Variant Details	ClinVar data for the same variants can appear to differ between the variant summary in the grid (which reports both SCV and RCV entries) and the variant detail page (which reports only RCV).
BSVI-16523	Filters	A saved hg38 region-based filter can be incorrectly viewed in the saved filter drop-down menu, when analyzing a hg19 sample.

KNOWN 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-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-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.
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.