BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.12010

October 13, 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:

- Family-based analysis
    - Trio filtering that results in a large number of genes (> 4096) is automatically stopped to prevent loading errors. Users are notified and directed to reapply filters.
- Metadata improvements
    - Additional sample import fields have been added to reduce redundant data entry. Sex, Sample Class, and Tumor Type can now be entered at the time of sample import.
- Workgroup improvements
    - The Workgroup Administration Console now identifies in a more visible location which workgroup the user is configuring.
    - Workgroup identification is now shown more prominently in the user profile drop-down.
- General user interface updates
    - The system now warns users (in multiple user interface locations) not to enter patient health information.
    - The system now provides an error message when a gene list name is longer than 50 characters. The gene list name must be corrected before proceeding.
    - The Audit Trail link is no longer available in the User Settings dialog. The link remains in the More dropdown within a given case.
    - The QC Filter checkbox no longer displays differently from other filter settings checkboxes.

Resolved Issues

Issue Key	Issue Category	Description
BSVI-10013	Classification (Family-Based Analysis)	Variants without a canonical transcript are now sorted in proper order for prediction.
BSVI-17206	WalkMe	WalkMe tutorial guide is now enabled.
BSVI-17350	Variant Grid	Association count numbers are now displayed properly when using the latest version of Mozilla Firefox.
BSVI-17514	Metadata	Case modification errors are all now shown in the user interface.

Issue Key	Issue Category	Description
BSVI-17631	Sample Delete	Fixed an error that prevented some samples from being deleted.
WAC-572	Workgroups	When an admin removes themselves from a given workgroup, they are now rerouted to the Workgroup List page.

KNOWN ISSUES

Issue Key	Issue Category	Description
BSVI-9300	Annotation	BaseSpace Variant Interpreter Beta does not validate the search criteria of a chromosome as a user enters an actual position.
BSVI-11052	Report	The inheritance mode content extends off the report page.
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-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-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-16523	Filters	A saved hg38 region-based filter can be incorrectly viewed in the saved filter drop-down menu, when analyzing a hg19 sample.
BSVI-17531	Registry	When using the sample metadata sheet to upload 40 or more cases, the software might freeze.
BSVI-17814	Workgroups	Users may experience issues when connecting to BaseSpace Sequence Hub for the first time. When prompted, the user should select their Personal workgroup.
BSVI-17822	Workgroups	After changing workgroups, the user may need to perform a browser refresh to update the data.
BSKN-2898	Add New Association	Clinical Trial open and close dates can appear one day earlier depending on which time zone the association was created in.
BSKN-2815	Add New Association	Clinical Trial curator summary is duplicated from the evidence summary section.
BSKN-2194	Add New Association	Publication ID allows nonintegers to be used and creates an erroneous link to the PubMed website.

Issue Key	Issue Category	Description
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.