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

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

<table>
<thead>
<tr>
<th>Issue Key</th>
<th>Issue Category</th>
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</table>
| 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.
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<td>ON-345</td>
<td>Variant Grid</td>
<td>Autocomplete box shows results marked as 'obsolete' by nomenclature authority.</td>
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<tr>
<td>ON-432</td>
<td>Variant Grid</td>
<td>Expansion to related phenotypes sometimes yields results that are too distant.</td>
</tr>
<tr>
<td>ON-516</td>
<td>Variant Grid</td>
<td>Results in autocomplete pop-up might be sorted inconveniently (subjective)</td>
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<tr>
<td>WAC-546</td>
<td>Workgroups</td>
<td>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).</td>
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<tr>
<td>WAC-730</td>
<td>Workgroups</td>
<td>In some instances, the bulk edit of user permissions checkboxes does not work. Log out/log in to resolve this, or edit users individually.</td>
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