BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.10580

September 1, 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.

Note: The new release of Variant Interpreter requires reanalysis of existing samples.
Samples that have not been analyzed in the current software version are listed as Action Required in the Status column of the Cases tab. The variant data for these samples are not displayed in the variant table until the reanalysis is performed.

New Features

- Support for BaseSpace Knowledge Network, which stores genotype-phenotype association data for use during interpretation of genomic variants.
    - Features include:
        - Variant Details button in the Variant Grid provides access to BaseSpace Knowledge Network association data, relative to the variant in question.
        - Association column in variant grid provides summary of available data in BaseSpace Knowledge Network sources.
        - Users can create a knowledge base associated with a user-defined private workgroup (see *Support for Workgroups* below).
        - Users who have established a private knowledge base can add their own association data to the knowledge base. This feature is accessible within the Variant Details page.
        - Support for major ontology standards (see *Support for standard ontologies* below).
    - Content includes:
        - Associations generated by Illumina Curation Scientists.
        - Associations present in the ClinVar public repository. This content is shown when the transcript matches the expected reference (mismatches are not shown). Link: http://www.ncbi.nlm.nih.gov/clinvar/.
- Support for Grch38 human reference genome assembly has been added.

  Note: BaseSpace Knowledge Network content from Illumina is curated in the context of reference Grch37. This content is not available for *.vcf files generated using reference Grch38.
    - When a user imports a new *.vcf file, the system performs a validation to check the reference version.
    - The reference version for each case is shown on the Cases page.

    - Additional rules applied to reference genome support:
        - The system does not support the use of hg19/Gcrh37 mitochondrial representation (chrM) in customer annotations and region filter files.
        - The system ignores unplaced/unlocalized Gcrh38 contigs.
    - Support for standard ontologies.Includes the following standard ontologies: SNOMED-CT, HPO, OMIM, ICD-9.ICD-10, RxNorm.
    - Real-time suggestion (incremental search) functionality narrows search space as user enters phenotype terms.
    - Support for custom phenotype and drug ontologies.
- Support for Tumor/Normal analysis.
    - Workflow allows import of paired *.vcf files generated by the Illumina Strelka Tumor/Normal somatic variant calling pipeline.
- Support for Workgroups (multi-user entities).
    - Users can create a workgroup for collaboration; the workgroup creator is defined as the primary owner.
    - Allows access to previously established Workgroups in BaseSpace Sequence Hub.
    - Workgroups are managed through the Workgroup Administration Console. Management features include:
        - Create a workgroup.
        - Add/remove members.
        - Change access level for members. Access level determines the availability of features for each member.
- Variant grid redesign.
    - New representations of annotation and context data for each variant found in a case. Columns now include the following grouped information cells: variant, gene, consequence/allele, associations, zygosity (germline analysis), maximum allele frequency, read, custom annotation, and interpretation.
        - Single field columns are sortable (gene, zygosity, and allele frequency).
        - Columns are no longer customizable.
- Annotation engine update.
    - Refreshed annotation engine to include more recent data. Update is version 1.4.1 (previous version was 1.3.6).
- Transcript filtering.
    - A user can now apply filters to either all transcripts or canonical only.
- Internet browser support.
    - The system no longer supports Internet Explorer 10.

Resolved Issues

Issue Key	Issue Category	Description
BSVI-12497	Classification (Family-Based Analysis)	A user no longer needs to set the sample class for a parent subject before applying the parent analysis to the proband.
BSVI-13804	Classification (Family-Based Analysis)	A user is now allowed to add mother and father subjects to a proband case when the sex of each subject is undefined. The user is prompted to update the missing fields during case validation.
BSVI-13360	Registry	An indication entered when creating a new case and adding the subject and sample can now be deleted.
BSVI-13790	Report Generation	Refreshing an open report no longer should cause the browser to generate a 404 error.
BSVI-13651	Report generation	Variant Read Frequency (VRF) field rounding is now rounded to 3 decimal points in the report preview and the PDF Report.
BSVI-13094	Sample Metadata	If either or both parents are associated with a proband, the association must be added to the proband metadata. Otherwise, an error message appears.
BSVI-13539	User Interface	Resizing the Home page to a smaller size no longer causes the content to overlap with the footer.

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-11929	Annotation	If a user tries to upload a gene, annotation, or region list with a file name longer than 50 characters, the Enter Annotation Label field appears red. BaseSpace Variant Interpreter Beta requires that these file names be ≤50 characters long.
BSVI-10013	Classification (Family-Based Analysis)	Variants without a canonical transcript are sorted out of order for prediction.
BSKN-2898	Add New Association	Clinical Trial open and close dates can appear one day earlier depending on the 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 non-integers to be used and creates an erroneous link to the PubMed website.
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 Variants grid does not appear.

Issue Key	Issue Category	Description
BSVI-16408	Import	Upload of Grch38 VCF files will occassionally result in error. Deleting the failed upload and retrying will clear the error.
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-11278	Registry	If a user enters a partial name and then changes it, both names become part of autopopulate. A user must completely clear the text box to see both autocomplete options.
BSVI-13331	Registry	When using the sample metadata sheet to simultaneously upload 40 or more cases, the software might freeze.
BSVI-11052	Report	The inheritance mode content extends off the report page.
BSVI-15768	Variant Grid	For variants with multiple dbSNP IDs, only 2 of the IDs will be displayed in the variant grid.
BSVI-16383	Variant Grid	The Associations Column does not explicitly notify the user when a public or private association requires reanalysis.
ON-345	Variant Grid	Auto-complete box shows results marked as 'obsolete' by nomenclature authority.
ON-432	Variant Grid	Expansion to related phenotypes may include results that are too distant.
ON-516	Variant Grid	Results in auto-complete pop-up might be sorted inconveniently (subjective).
WAC-546	Workgroups	Inviting a user who is not currently registered will send an invitation email and mark the user as pending, however this is not visible in the user interface. To guarantee that an invited member has been notified, contact the invitee directly, independent of Variant Interpreter (Beta).