BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.7814

April 28, 2016

Introduction

These Release Notes detail known 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.

I. Import (VCF/gVCF Upload)

NEW FEATURES:

- BaseSpace Sequence Hub Import
    - Seamlessly transfer files from BaseSpace Sequence Hub into BaseSpace Variant Interpreter Beta, individually or via batch upload.
- Local File Import
    - Upload files from a local directory, individually or via batch upload.
- Metadata Sheet
    - Upload sample metadata.
- VCF Input Requirements
    - The software supports hg19 and GRCh37 genome builds, not GRC37 and GRCh38 as published in the online help.

II. Annotation

NEW FEATURES:

- Secondary Annotation
    - Annotation of variants using multiple sources.
    - External links to sources.
    - Auto-classification of pathogenicity.
        - Aggregate annotation to determine pathogenicity.
- Custom Annotation Files
    - Define private interpretations to add to public annotations.
- Variant and Annotation Display
    - Variant review table
        - Display variants and annotations in a single interface.
        - Display affected transcripts.
    - Select variants of interest and add interpretations.

III. Filtering

NEW FEATURES:

- Variant Filters
  - Restrict view to variants matching specific annotation criteria.
  - Pathogenicity and allele frequency.
- Gene List Filter
  - Restrict view to variants appearing in specific genes.
  - Customizable list of genes.
- Genomic Regions Filter
  - Restrict view to variants appearing in a specific genomic region.
  - Customizable region list in BED file format.

IV. Classification & Interpretation

NEW FEATURES:

- Family Genotype and Filtering
  - Family pedigree
    - Analyze mother, father, and siblings.
    - Analyze, singleton, duo, trio, or whole family.
  - Inheritance mode filtering
    - Restrict view to variants showing a specific inheritance pattern.
    - Filter by autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive, and *de novo*.
    - Strict mode available to reduce Mendelian errors.

V. Reporting

NEW FEATURES:

- Incorporates selected variants and interpretations.
- Text fields to add commentary.

VI. Audit Log and Change Tracking

NEW FEATURES:

- Full audit log.
- Tracks access and changes to case data.

RESOLVED ISSUES

Issue Key	Issue Category	Description
HAD-13054	Audit Log	Filter tab creation, modification, and deletion events do not trigger an audit log event.
HAD-13508	Classification (Family-Based Analysis)	Proband is not annotated correctly with parent associated sample information for certain variants.
HAD-13045	Filtering	When a user is filtering by coordinates, the coordinates do not populate the filter setting summary and the filter is not applied.
HAD-13421	Filtering	Null affected status is displaying as **unknown**.
HAD-9920	Import	Links to Ensembl Genome Browser direct users to coordinates in GRCh38 instead of GRCh37.
HAD-13071	Report	An associated condition can be only 250 characters long. Otherwise, it overlaps another column in the report.

KNOWN ISSUES

Issue Key	Issue Category	Description
HAD-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.
HAD-9300	Annotation	BaseSpace Variant Interpreter Beta does not check whether the search criteria of a chromosome a user is inputting is an actual position. The filter does not apply to invalid positions.
HAD-12497	Classification (Family-Based Analysis)	A user must set the sample class for a parent subject before applying the parent analysis to the proband.
HAD-10013	Classification (Family-Based Analysis)	Variants without a canonical transcript are sorted out of prediction order.
HAD-13804	Classification (Family-Based Analysis)	A user cannot add mother and father subjects to a proband case unless the sex of each subject is defined. Using the software interface, the user can manually define the sex to populate the case.
HAD-11250	Filtering	In the Variants grid, users cannot sort the following columns: • HGVS cDNA Changes • dbSnp • OMIM id • COSMIC id • Consequence • SIFT • PolyPhen • Exons • ClinVar Significance

Issue Key	Issue Category	Description
HAD-12422	Import	If a user tries to upload a VCF file that is not properly formatted, the upload fails with a cryptic error message. The file has a status of fail and the user can then delete it. Consequently, the Variants grid does not appear.
HAD-13094	Sample Metadata Sheet	If either or both parents are associated with a proband, the association must be added to the proband metadata. Otherwise, an error message appears.
HAD-11278	Registry	If 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.
HAD-13331	Registry	When using the sample metadata sheet to simultaneously upload 40 or more cases, the software might freeze.
HAD-13360	Registry	An indication entered when creating a new case and adding the subject and sample cannot be deleted.
HAD-11052	Report	The inheritance mode content extends off the report page.
HAD-13790	Report	Refreshing an open report might cause the browser to generate a 404 error.
HAD-13651	Report	Variant Read Frequency (VRF) field rounding is different in the report preview (rounded to 3 decimal places) than for the PDF Report (rounded to 8 decimal places).
HAD-13539	UI	Resizing the Home page to a smaller size can cause the content to overlap with the footer.

ERRATA

- ### TIMESTAMP IN PDF REPORT
    - The time-stamp shows the ingestion time, not the report generation time. It also shows UTC rather than local time.
- ### SUPPORTED FILE TYPES
    - The software only supports files with a.vcf and .vcf.gz extension.