Release Notes

BaseSpace Variant Interpreter v2.8.0

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Page 2 of 3

INTRODUCTION

These Release Notes detail the latest release of BaseSpace Variant Interpreter, including known issues.

BaseSpace Variant Interpreter provides an interface for users to annotate, curate, interpret and report on the results from the sequencing pipeline.

FEATURES

The following features are now available:
- Updates to the Registry content and functionality:
  - New filters on the Genome Build and Status columns.
  - New column to display Project name.
  - Project column content is included in the text search.
  - Bulk search on subject IDs in the text search box using the pattern. SUBJECTIDS=subjectID1;subjectID2;…subjectIDn. Note that this search does not work for any subject ID that contains a semicolon. Only exact matches to subject IDs are returned.
  - The length of the search field is limited to 255 characters unless it begins with SUBJECTIDS=.
- GATK haplotype caller and Dragen VCFs may contain variants with ALT alleles such as "*" or "<NON_REF>". If the GT (genotype) field of the VCF references these ALT alleles, the entire VCF line is not ingested; instead the skipped variants are placed into an "ignored lines" file that is accessible from the Case Panel.

RESOLVED ISSUES

- Resolved improper authorization when downloading files.
- Ingestion fails for platypus and pisces VCFs with triallelic sites.
- The subject input field in case metadata panel accepts up to 255 characters.
- The sample input field in case metadata panel accepts up to 255 characters.
- Failed samples counter is now updated when VCF ingestion fails.
- Population frequency filtering on GnomAD and GnomAD-exome now behaves as 1000Genomes filtering: selecting multiple subpopulations requires that all subpopulations meet their specified frequency filters.

KNOWN ISSUES

- Cases will not appear in the Case Registry if they are imported from BaseSpace Sequence Hub and ingestion fails before the genome build is associated with the case.
- ClinVar filters applied to cases with sample type germline become invalid if the sample type is switched to tumour-only. To resolve this, change the sample type back to germline and remove the ClinVar filter before enabling the tumour-only sample type.
- Using tumour types with the ontology ILLUMINA-CUSTOM can prevent associations being saved. Use SNOMED, HPO, or OMIM to describe phenotypes, as these are not affected by this issue.

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- When adding an interpretation for a variant in a rare disease case, the interpretation is not saved if the Mode of Inheritance selection is Unknown. All other Modes of inheritance: autosomal recessive, de novo etc are saved correctly.
- Mitochondrial genes coming from phenotype search are declined by Gene List Manager when saving.
- BaseSpace Variant Interpreter fails to process manifests if the reference header in VCF is hg19 and the Assembly column in the manifest file is GRCh37.
- Users who do not have BSKN Curator permissions can select the approval button although approve will fail.
- Gene lists containing deprecated gene symbols do not return a result when filtering.
- Dragen VCFs are not fully supported.
- Multi-sample germline VCFs are not supported.
- BaseSpace Variant Interpreter shows ClinVar status as "Enabled" for nested annotation.
- Count of Analysis Result is missing from Subjects list page.
- If filter results in 0 variants, count is shown as "0 of 0 variants".
- Variant grid is misaligned after removing an invalid filter. Refreshing the page will resolve this problem.
- BaseSpace Variant Interpreter does not load small variants from manta.
- Gene List creator becomes unresponsive in workgroups with many custom annotation files.
- Case history is slow to load.
- Extend user session is not working.
- Partially overlapping genes are not displayed in order of pLI score (popup).
- In the annotation upgrade to Nirvana 3.1.1 in BaseSpace Variant Interpreter v2.7, some gene symbols became deprecated. Gene lists containing deprecated gene symbols do not provide a filtering result. For reference, the gene lists visible in the Gene List Manager contain the original gene symbols that were submitted. These can be downloaded and resubmitted to update synonyms to the latest symbol. Gene lists that are no longer needed can also be deleted in the Gene List Manager.
- All filters on CNV consequence based on Nirvana 1.6.2 (BaseSpace Variant Interpreter 2.6 and earlier) are now obsolete. Users will be prompted to remove those filters on each case and can then apply fresh filters.
- Filters on Exac (BaseSpace Variant Interpreter 2.6 and earlier) are now obsolete. Users will be prompted to remove those filters on each case and can then apply fresh filters.