Release Notes

BaseSpace Variant Interpreter v2.9.0

December 2019

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

Updates to the Case Registry:

- Refactored Dates column now displays both the Created and the Last Updated date. The default sort of the case registry is to show the most recently updated cases first.
- Improved sort on the refactored date column, to enable users to search for cases on the Created and/or Last Updated dates.

RESOLVED ISSUES

- Resolved issue where Gene List creator becomes unresponsive in workgroups with many custom annotation files. Only the content required for each tab within the Settings page is loaded. Note that creating gene lists or loading custom annotation files may still be slow if there are many hundreds of that type of file already present.
- Resolved issue where cases that fail ingestion very early (before the Genome Build is identified by the system) are not displayed in the Case Registry.

KNOWN ISSUES

- During an import from BaseSpace Sequence Hub, the Last Updated date is unavailable, and the case will appear at the bottom of the last page in the Case Registry. This is because the default sort of the Case Registry is on the Last Updated date. Cases that fail ingestion and result in a Failed state also have no Last Updated date, so will appear at the end of the Case Registry listing.
- 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.
- 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.