Release Notes

BaseSpace Variant Interpreter v2.11

August 2020

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

Support for DRAGEN Enrichment v3.5.7 and v.3.6.3 output:

- BSVI now supports the loading of output from DRAGEN Enrichment by import from BaseSpace Sequence Hub (BSSH) or by manifest. Variants from all three variant callers provided by DRAGEN are displayed in the variant grid (single nucleotide variants/indels, structural variants, copy number variants).
- For an analysis result to load all types of variants into one case, the small variant VCF must be present in the DRAGEN output files. Users are advised to use the “hard-filtered” VCF for ingestion.
- For a DRAGEN Enrichment case that has been loaded from BSSH, if the bam file is present alongside the VCFs in BSSH, the reads supporting a variant will be visible when using “ViewInIGV”.
- The Dragen variant caller version is extracted from the VCF header and displayed in the Case Panel.
- Earlier versions of the DRAGEN Enrichment workflow are not fully supported.

DRAGEN Somatic v3.6.3 is supported.

RESOLVED ISSUES

- Previously, if a filter resulted in 0 variants, count is shown as "0 of 0 variants". The correct total number of variants is now shown.

KNOWN ISSUES

- When importing from BSSH, users are advised not to add information to the Project column of the manifest as this may result in a duplicated view of the case in the registry. The case itself is not duplicated.
- Other DRAGEN workflows, versions and VCFs are not fully supported.
- Copy number variants with LOH (loss of heterozygosity) will have two near-identical entries in the variant grid, one representing the “deletion” part of this event and another for the “duplication”
- Importing associations from BSKN may fail if they were created using an ILLUMINA-CUSTOM tumour type
- Hyperlinks to variants within cases will take the user to that variant only if they are already logged in and in the appropriate workgroup. Otherwise, following a hyperlink to a variant will take the user to the Case Registry of their current workgroup, if logged in, or to the BSVI login page, if not logged in.

- 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.
- 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.
- BaseSpace Variant Interpreter does not load small variants from the structural variant caller manta.
- Case history is slow to load.
- Extend user session is not working.
- Partially overlapping genes are not displayed in order of pLI score (popup).
- The case registry displays the owner as “Unassigned” for inactive cases.
- In the registry, a case owner cannot be assigned to inactive cases
- After reanalysis, there is a lag before the case owner and status is displayed