Release Notes

BaseSpace Variant Interpreter v2.6.0

February 2019

INTRODUCTION

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

BSVI 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:

- The Data Type column now includes tumour type in the registry. If text is wider than the column, a user can either resize the column or hover over to view the complete text. Users can also search the tumor type content.
- The reference genome build can be entered in the optional "Assembly" column in the Manifest. This provides support for family-based analysis using VCF files that do not contain the reference genome build in their header. Note that all members of a pedigree must have the same genome build in the Manifest.
- Links are now displayed to the UCSC genome browser for SV and CNV. These were previously available only for SV/indels.

RESOLVED ISSUES

- Included support and display of sequencing metrics from platypus VCFs in family-based analysis.
- Fixed an issue where mutational signatures were not generated for samples with a very large somatic VCF.
- Fixed uploading issue for VCF files when its first variant starts on chrM.

KNOWN ISSUES

- If a user selects the "Mode of Inheritance" as "Unknown" when adding an interpretation for a variant in a rare disease case, the interpretation cannot be saved. All other modes of inheritance such as autosomal recessive, de novo, etc. are unaffected.
- Using tumour types with the ontology ILLUMINA-CUSTOM can prevent associations being saved. It is preferable to use SNOMED, HPO, or OMIM to describe phenotypes and these are not affected by this issue.