

# Release Notes

## BaseSpace Variant Interpreter v2.9.1

February 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

**Case hyperlinks:** Specific cases can be reached in BSVI using a hyperlink. If a user is logged into BSVI in a different workgroup, or was in a different workgroup when they last logged in, the hyperlink would not link to the case, but to their previous Case Registry view. In this release, following the hyperlink will take the user through authentication if required, switch the workgroup that the user is in, provided they have access, and open the case indicated in the hyperlink. Please note that simultaneous access to different cases across different workgroups, even if in different browser instances, is unsupported in any browser.

## KNOWN ISSUES

- 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.
- 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.