

BaseSpace Variant Interpreter Beta Release Notes

BaseSpace Variant Interpreter Beta v1.0.13700

December 15, 2016

FOR RESEARCH USE ONLY

© 2016 Illumina, Inc. All rights reserved.

Illumina, 24sure, BaseSpace, BeadArray, BlueFish, BlueFuse, BlueGnome, cBot, CPro, CytoChip, DesignStudio, Epicentre, GAllx, Genetic Energy, Genome Analyzer, GenomeStudio, GoldenGate, HiScan, HiSeq, HiSeq X, Infinium, iScan, iSelect, ForenSeq, MiSeq, MiSeqDx, MiSeq FGx, NeoPrep, Nextera, NextBio, NextSeq, Powered by Illumina, SeqMonitor, SureMDA, TruGenome, TruSeq, TruSight, Understand Your Genome, UYG, VeraCode, verifi, VeriSeq, the pumpkin orange color, and the streaming bases design are trademarks of Illumina, Inc. and/or its affiliate(s) in the U.S. and/or other countries. All other names, logos, and other trademarks are the property of their respective owners.

Introduction

These Release Notes detail new features, known issues, and recently resolved issues for the BaseSpace Variant Interpreter Beta Testing software. For details on how to operate BaseSpace Variant Interpreter Beta Testing software, see the online help, which is available from the Help icon in the application.

NEW FEATURES

No new features in this release.

RESOLVED ISSUES

Issue Key	Issue Category	Description
BSVI-20062	User Interface	The "Setting" dropdown on the main page includes a "Patterns" option which should not be visible.
BSVI-20065	User Interface	The "More" dropdown on the Case Details page includes a "Export" option which should not be visible.

KNOWN ISSUES

Issue Key	Issue Category	Description
BSKN-3787	Associations	ClinVar SCV associations are not displayed as children of related RCV associations.
BSVI-19408	Audit Log	When archiving an interpretation, archived status is not saved in audit logs.
BSKN-3868	Curation Portal	Gene-level curation associations should not include fields for exon, amino acid or consequence data.
BSVI-12422	Import	If a user tries to upload a VCF file that is not properly formatted, the upload fails with an ambiguous error message. The file shows a status of fail, and the user can then delete it. Consequently, the variant grid does not appear.
BSVI-16408	Import	Uploading of multiple Grch38 .vcf files occasionally results in an error. The error can be cleared by deleting the failed upload and retrying.
ON-345	Variant Grid	Autocomplete box shows results marked as 'obsolete' by nomenclature authority.
ON-432	Variant Grid	Expansion to related phenotypes sometimes yields results that are too distant.
ON-516	Variant Grid	Results in autocomplete pop-up might be sorted inconveniently (subjective).

Issue Key	Issue Category	Description
WAC-546	Workgroups	Invited users who are not currently registered are sent an invitation email and marked as pending, but their status is not visible in the user interface. To confirm that the invitation has been received, users can contact the invitee directly, independent of BaseSpace Variant Interpreter (Beta).
WAC-730	Workgroups	In some instances, the bulk edit of user permissions checkboxes does not work. Log out/log in to resolve this, or edit users individually.
WAC-767	Workgroups	Workgroup Administrators cannot revoke a pending invitation at this time, however Workgroup Administrators can still remove members from the workgroup.