Illumina TruSight™ Software Suite Release Notes

v.2.6.3

November 2022

Introduction

TruSight™ Software Suite (TSS) is designed for translating genomic sequencing data into meaningful, interpretable results in rare disease cases. Highlights include:

COMPREHENSIVE, ULTRA-RAPID VARIANT CALLING

Use DRAGEN™ secondary analysis to call small variants, structural variants, mitochondrial variants, repeat expansions, runs of homozygosity, and SMN1/SMN2 variants.

SIMPLIFIED, CUSTOMIZABLE CASE MANAGEMENT

Manage cases from sample acquisition to report, assign cases to users, configure pipeline settings, and set quality control (QC) thresholds.

INTUITIVE, HIGH-POWERED INTERPRETATION AND REPORTING

Filter variants via gene lists, inheritance modes, custom annotations, and complex logic; flag, sort, and prioritize important variants; use customizable reporting templates.

SECURE, COMPLIANT ENVIRONMENT

TruSight Software Suite has been independently audited and certified for HIPAA compliance, ISO27001, and ISO13485. It is built to enable data privacy and compliance with the principles of GDPR.

These Release Notes detail the key features and changes to software components for the release of TruSight Software Suite v.2.6.3. This patch release ensures continued functionality and performance of existing Trusight Software Suite features including support for updated versions of the Nirvana clinical grade annotation service and CaseLog.

For information on how to use the system, see the TruSight Software Suite Online Help. TruSight Software Suite is a comprehensive solution for alignment, variant calling, variant annotation, filtering, interpretation, curation, and reporting, including features such as:

- Automatic secondary analysis with DRAGEN™ and annotation of:
  - Small Variants, CNVs, SVs, Mitochondrial variants, ROH, STRs, SMA
- Support for whole genome and whole exome sequencing; both from sequencing output file (FASTQ) and secondary analysis output (VCF)
- Sequencer and BaseSpace Sequence Hub Integration
- Case Dashboard and Test Management
- Turnaround time (TAT) management
- IGV Visualization
- Complex custom filters
- Custom flagging of variants

- Custom annotation
- SpliceAI & PrimateAI
- AI-based variant prioritization via Emedgene
- Auto-populated ACMG criteria
- Gene lists from phenotypes
- Storage of variant curation
- Visualization of aggregate data for genes or variants
- Customized report generation
- Multiple reports
- Audit logging
- Command-line interface for uploading FASTQs
- Improved API documentation

RESOLVED ISSUES

- Update code base to support new versions of Nirvana, Illumina's clinical grade annotation service. This update will ensure monthly updates of ClinVar, OMIM, ClinGen Gene Disease Validity and ClinGen Dosage Sensitivity Map.
- CaseLog functionality has been updated and customers should be able to access it without experiencing intermittent outages.
- Case ingestion will not fail when the DRAGEN SMN caller returned a "None" value.
- Resolved case ingestion failures intermittently experienced for complex cases.

KNOWN ISSUES

- Literature search result is not matched with Gene Details.
- The link to STR alignment is not working in variant details.
- Incorrect toast notification when adding association to a completed report.
- In default STR annotation threshold file - locus 'Spinocerebellar ataxia 17' is missing.
- New QC coverage BED file is ignored for "start from analysis" cases.
- labels in IGV track settings are distorted for long sample ID.
- ClinVar resource shows no genes for any phenotypes chosen in the TSS variant filter window.
- Large variants are annotated with multiple QC regions.
- 100x fastq single sample case ingestion completes great than 6hrs.
- Inconsistent behavior for the "cancel curation" button.

- Active filter tab cannot be consistent after navigating back from the case list page.
- Resume case is not working when case failed in VQS ingestion.
- Phenotype number in the subject header is not changed after switching subject in OUI.
- TAT start is not set for case posted via API.
- Different SV variant counts for the same case from FASTQ
- In a recently ingested case, clicking "Duplicate" on a filter tab does not create a new tab.
- Filter View Duplication tab is not working when filter name exceeds the character limit.
- User cannot return to the full gene list after closing gene details of a searched gene on the gene details tab
- TAT: Time elapsed increment before the case has been processed instead of staying in day 0.
- Reset filter redirects to the new case tab.
- Incorrect sex ploidy in exome workflow. The workaround is to remove the sex ploidy check from exome test definitions.
- Autosomal and Allosomal Transcript Standard are not Conserved during test definition export.
- Expose RN (ALT field) for legacy cases with STRs in the Change column
- Unexpected change in DRAGEN 3.9.5/ 3.7.5 /3.8.4 results for a FASTQ case
- Archived clients can be edited.
- Case-specific custom annotation associated filter should not be selected in test definition.
- PLINK error terminates the pipeline when VCFs is empty
- CMP: The SFA case still can be saved and ingest success with the wrong relationship
- Gene Evidence is missed after editing it.
- Some long deletions called by the SV caller are inappropriately categorized as small variants instead of SV
- Columns are not consistent in saved filter view which cases ingested with different case datasets.
- SampleIDs are case sensitive in the case create form but are not the sample UI.
- Variant search function on Caselog tab does not work
- Column Sorting was not maintained after migration.

Open known issues from previous releases can be found in TruSight Software Suite Release Notes v.2.6.2 and earlier:

https://support.illumina.com/downloads/trusight-software-suite-release-notes.html