

Illumina TruSight™ Software Suite Release Notes

v1.5.1

August 2020

Introduction

TruSight™ Software Suite (TSS) is designed for translating genomic sequencing data in 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.1.5.1. 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
- Case Dashboard and Test Management
- IGV Visualization
- Complex custom filters
- Custom flagging of variants
- Custom annotation
- SpliceAI & PrimateAI
- Gene lists from phenotypes
- Storage of variant curation
- Visualization of aggregate data for genes or variants
- Customized report generation
- Audit logging
- Command-line interface for uploading FASTQs

- API documentation

NEW FEATURES

- Test Management
 - Ability to add up to 10 custom annotation files
- Case Management
 - Adding new Case Management APIs (query cases by status, get report details of a case)
 - Subject level opt-ins for Secondary Findings, Research Consent and DNA Storage
 - Provide additional search functionality (subject last name, record number, sample ID)
- Variant Grid / Filters
 - Persist sort order within a given filter view within Interpretation
 - Enable user to identify transcripts that are partially overlapped by the boundaries of a large variant call (new filter option)
- Variant Details
 - Support auto literature search
 - Import partial gene-level association information when creating variant-level association
 - Support custom disease names when creating association
- Admin
 - Align on naming convention (update Report Category to Report Section)
 - Available on Australia instance
- Release versions
 - TruSight Software Suite v.1.5.1 runs DRAGEN v.3.5.7 for secondary analysis
 - TSS v.1.5.1 uses Nirvana v.3.10 for annotation
 - TSS v.1.5.1 uses KNS-API v.0.8.0 beta.13149 and KNS-UI v.0.10.0-beta.484 for curation

RESOLVED ISSUES

- Defect repairs (bug fixes) from v1.5 release.

Issue Key	Description
<i>OLYM-11405</i>	Adding a case to CaseLog remains in pending state, without ingesting the case.
<i>OLYM-12231</i>	Only the most recent variant association shows in the variant details tab when "get new associations" is clicked. All associations show correctly in the variant grid.
<i>OLYM-12277</i>	When user has multiple tabs open and logs out of one tab, other tabs will be inactive but may show data last viewed. Workaround is to manually log out of each open TSS tab.
<i>OLYM-12270</i>	If an analysis fails with a WES error, downloading logs for the case does not work.
<i>OLYM-12290</i>	In certain rare cases, phenotype overlap ingestion fails and the case cannot be interpreted. User must contact tech support to manually clear the error and reupload the sample.
<i>OLYM-12034</i>	Custom columns include the text "CUSTOM COLUMN" as the column name.
<i>OLYM-12116</i>	If a case has been open for >1 hour, IGV URLs can expire and gene tracks will not load.
<i>OLYM-12098</i>	Links to previous cases shown under Caselog summary are working correctly.
<i>OLYM-12079</i>	When uploading an STR customer annotation file from Excel, a whitespace error is introduced and the genome build information cannot be read.
<i>OLYM-12002</i>	When 20 flags are used, the Interpretation header fails to load correctly.
<i>OLYM-10103</i>	When a locked filter tab with indications applied is duplicated, default phenotype genes are seen in the additional genes section.
<i>OLYM-10328</i>	For variants spanning many genes, the transcripts tab does not load correctly.
<i>OLYM-12288</i>	MT variants cannot be viewed in IGV when the reference genome is GRCh37.

KNOWN ISSUES

Issue Key	Description
<i>OLYM-9211</i>	Details about the OTHER variant are seen before those of the SIBLING variant in variant details.
<i>OLYM-10375</i>	Pop outs appear cut off at different points depending on the size of the user's screen.
<i>OLYM-10555</i>	When filtering with OMIM, gnomAD and ClinGen filters, the "exclude consequences" filter does not return correct results.
<i>OLYM-11337</i>	When a user with API Access role accesses interpretation page directly, user is redirected to a broken page. Workaround is to manually update the URL.
<i>OLYM-11406</i>	If a variant spans multiple genes and one of the genes has information associated with it, the user is unable to add information to other genes. A workaround exists by using the Edit button to manually enter gene information.
<i>OLYM-11967</i>	Any case ingested prior to v.1.5 will have all variants marked as "unreviewed" by showing a blue dot under "Status" column.
<i>OLYM-12059</i>	Gene list dropdown does not appear until user clicks out and back in after first selection. Workaround is to use autocomplete functionality.
<i>OLYM-12060</i>	In locked filter tabs, number of genes selected for filtering can exceed 7500.
<i>OLYM-12070</i>	Selecting a gene list in test management will close the entire Gene List modal, not just the gene list dropdown menu.
<i>OLYM-12077</i>	If a gene list name contains (_) underscore in it, the gene list autocomplete is unable to find it. Workaround is to create gene lists without (_).
<i>OLYM-12082</i>	WalkMe functionality is not set up in the UI.
<i>OLYM-12181</i>	In certain SVs, false positive de novo calls are seen. This issue has been fixed in a subsequent DRAGEN release.
<i>OLYM-12219</i>	CaseLog summary does not update with alternate allele frequency even after the addition of related cases.
<i>OLYM-12243</i>	The DRAGEN QC BED file requires the "chr" prefix for GRCh38 builds. Workaround is to ensure files have the "chr" prefix.
<i>OLYM-12244</i>	If a variant spans >2400 genes, the transcript tab fails to upload correctly.
<i>OLYM-12268</i>	It may take up to 24 hours to ingest cases into CaseLog.

<i>OLYM-12271</i>	When cases are added to CaseLog, there is a delay before they are seen in the CaseLog summary.
<i>OLYM-12280</i>	A VCF output from joint calling (JointGT-repeat-vcf) is not properly bgzipped and tabix indexed. Workaround is to manually fix the header.
<i>OLYM-12436</i>	IGV - tracks appear to be mis-aligned at the genome-wide view.
<i>OLYM-12471</i>	While working in the literature search section, idle timeout may occur. Workaround is to log back in and reopen the literature search.
<i>OLYM-12498</i>	When removing a variant from a report and then re-adding it back into the report, an error message is seen.
<i>OLYM-12519</i>	In a Test with multiple customer annotation files, user is unable to see the row names in the 'All Population' modal.
<i>OLYM-12522</i>	Whitespaces are not allowed when filtering tabs.
<i>OLYM-12532</i>	In the PDF report, association summaries are cut off at the right margin.
<i>OLYM-12565</i>	References are not removed (or auto deselected) when a variant has been removed from the report.
<i>OLYM-12567</i>	Some cases remain "In processing" and do not terminate or complete.
<i>OLYM-12573</i>	(AUS instance only) When creating a virtual variant in the IGV view and then clicking back to the Interpretation page, the user is stuck in the processing page. Workaround is to click to the Subject Info page and then click to the Interpretation page.
<i>OLYM-11801</i>	When the user creates a virtual variant, the variant is successfully created but there is no green success banner message.
<i>OLYM-12136</i>	For ROH variants, the partial overlap symbol does not work correctly, and is applied to all ROH variants regardless of overlap.
<i>OLYM-12147</i>	When adding InDels to a report, the variant ID may get cutoff in the report modal instead of wrapping around.