DRAGEN TSO500 ctDNA Analysis Software

Customer Release Notes

V2.6.2

For TruSight Oncology 500 ctDNA Assay

December 12, 2024

Introduction

These Release Notes detail the key changes to software components for the DRAGEN TSO500 ctDNA Analysis Software v2.6.2 on DRAGEN server. For full details, please consult the DRAGEN TSO 500 ctDNA Analysis Software v2.6.2 User Guide available on the support website.

This software is intended for use with the TruSight Oncology 500 ctDNA and TruSight Oncology 500 ctDNA v2 assays.

- Software Version: 2.6.2
- DRAGEN software version inside of the TSO500 Docker Image: 3.10.18
- DRAGEN software version on the host DRAGEN server: 3.10.19

The software package includes:

- dragen_tso500_ctdna_2.6.2.tar – a tar file of the DRAGEN TSO 500 ctDNA v2.6.2 Analysis Software docker image containing DRAGEN 3.10.18
- dragen_tso500_ctdna_2.6.2.sif – singularity image format file of the DRAGEN TSO 500 ctDNA v2.6.2 Analysis Software
- uninstall_DRAGEN_TSO500_CTDNA-2.6.2.sh - bash script used to uninstall DRAGEN TSO500 ctDNA
- resources/ - workflow resource bundle for DRAGEN TSO500 ctDNA Analysis Software v2.6.2
- DRAGEN installer:
- dragen-3.10.19 -11.multi.el7.x86_64.run – the DRAGEN installer for servers running CentOS 7.
- dragen-3.10.19-11.multi.el8.x86_64.run – the DRAGEN installer for servers running Oracle Linux 8.
- check_DRAGEN_TSO500_CTDNA-2.6.2.sh - bash script used to validate DRAGEN TSO500 ctDNA installation is successful
- install_DRAGEN_TSO500_CTDNA-2.6.2.run - script used to install TSO500 ctDNA
- DRAGEN_TSO500_CTDNA-2.6.2.sh - bash script used to launch DRAGEN TSO 500 ctDNA

NEW FEATURES:

- Updated installer with multi-version DRAGEN support: DRAGEN TSO500 ctDNA Analysis Software v2.6.2 can run on the same DRAGEN server with DRAGEN pipelines v4.3 or higher (e.g., DRAGEN Enrichment 4.3)

KNOWN ISSUES:

- There are minor differences in outputs of exon_cov_report and gene_cov_report from DRAGEN TSO 500 Analysis Software on ICA and DRAGEN server/NovaSeq 6000Dx Analysis Application

- NTC sample (No-Template Control samples with 0 reads) is marked as 'False' (under the header 'Completed_All_steps') in the metrics output file instead of being marked 'TRUE'.
- Moving or modifying files during the analysis may cause the analysis to fail or provide incorrect results.
- Using control-c during a running analysis may cause an FPGA error. To recover from an FPGA error, shut down and restart the server.
- The sample sheet should not have blank rows between samples in the [Data] section, this may cause a run failure.
- Performance not verified using reads other than 2 x 151, paired end, dual index.
- The software does not notify the user when InterOp files for RunQC are missing or corrupted.
- Analysis fails when starting from V1 sample sheets due to missing adapter sequences in V1 sample sheet template. Users are recommended to start with V2 sample sheet template or add adapter sequences manually.
- Pipeline does not exit early and continues to the next DragenCaller step due to TSO500 ctDNA FASTQ validation failure if Fastq_list.csv is missing.
- High chimeric read count results in incorrect TMB calculation.
- In the V2 CNV cutoff bed file, gene "MYCL" should be listed instead of "MCYL1".
- Variant consequences are not assigned consistently for co-occurring variants when stop_gained is introduced.

PRODUCT LIMITATIONS:

- Sample sheets generated for auto-launch on ICA are not compatible and cannot be reused without changes for DRAGEN TSO500 ctDNA Analysis Software on a Local DRAGEN server, and vice versa.
- The values in the Run Metrics section will be listed as 'NA' if the analysis was started from FASTQs or if the analysis was started from BCLs but the InterOp files are missing or corrupted.
- Germline estimation uses the latest publicly available population data and is estimated to be representative of targeted population, the impact of rare germline mutations is expected to be limited.
- The Illumina Annotation Engine (aka Nirvana) may report incorrect HGVS c. and HGVS p. notation for small variants occurring in RefSeq transcripts that exhibit transcript sequences differing from the genomic reference (i.e., RNA-edits). Currently the HGVS c. error rate is 0.00527% and the HGVS p. error rate is 0.00737%.
- The CNV caller has slightly higher noise for sample types that are not included in the baseline used for normalization (eg., cell lines). The baseline samples consist of mostly healthy donor clinical samples and SeraCare-contrived samples.
- MSAF output has had limited testing and needs to be used with caution. Updates to the small variant calling have led to an increased MSAF in samples with higher DNA input.

Release History

Revision	Release Reference	Originator	Description of Change
00	CN 1116532	Svetlana Bureeva	Initial Release