Illumina DRAGEN TruSight Oncology 500 ctDNA Analysis Application on NovaSeq 6000Dx (RUO)

Release Notes

V2.6.1

For TruSight Oncology 500 ctDNA Assay

Introduction

These Release Notes detail the key features and known limitations of DRAGEN TruSight Oncology 500 ctDNA Analysis Application on NovaSeq 6000Dx (RUO). For full details, please consult user documentation for this software available on the support website.

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

The software is designed to be installed on NovaSeq 6000 Dx (RUO mode).

- DRAGEN TSO 500 ctDNA Pipeline Version: 2.6.1
- DRAGEN software version: 3.10.18
- DRAGEN TruSight Oncology 500 ctDNA Analysis Application on NovaSeq 6000Dx (RUO) version: 2.6.1-4v2

NEW FEATURES:

Initial release

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'.
- 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.
- The application will launch the analysis when the storage on the server is less than 4TB, which may lead to running out of space.
- The "Next" button in the application is not available while trying to create a run using Import Run with a sample sheet that contains the Sample_ID in the [TSO500L_Data] section but not in the [BCLConvert_Data] section.
- When using a Samplesheet where Sample_ID column is missing from BCLConvert_Data or tso500l_Data section for Import Run, error message is displayed, and there's a typo "sample_idin" (missing a space).

- Requeue analysis with no changes throws an error. It works when the second option "Edit run settings and requeue analysis" is selected.
- Run Details - Results section has Sample ID header (empty table).
- Analysis Run Results not displayed in run details for runs started by users with the role Sequencer Operator User.
- 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:

- The sample sheet must be configured as described in the provided templates, the User Guide or by using BaseSpace Run Planning tool.
- 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.
- Added validation for the storage size selection generates an error if "Small" or "Medium" values are selected ("Large" is required as a minimum) but the error message appears with a delay.
- 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

Version	Workflow#	Author	Description of Change
00	CN 1114085	Svetlana Bureeva	Initial Release