TSO500 ctDNA Local App Software

Release Notes

V1.0.1

For TruSight Oncology 500 ctDNA Assay

**17-FEB-2020**

Introduction

These Release Notes detail the key features and known limitations to software components for the Local TSO500 ctDNA App v1.0.1.

This software is intended for use with the TruSight Oncology 500 ctDNA Assay.

* Software Version: 1.0.1
* Docker Image ID: 455a34f55041

The software installer includes:

* illumina-tso500-ctdna\_1.0.1.tar – a tar file of the TSO500 ctDNA docker image.
* test\_TSO500\_ctDNA-1.0.1.sh – a test script for validating the installation of TSO500 ctDNA.
* uninstall\_TSO500\_ctDNA-1.0.1.sh – a script for uninstalling TSO500 ctDNA.
* resources/ - a directory containing all resources files necessary for TSO500 ctDNA.
* dragen-3.4.5.el7.x86\_64.run – the DRAGEN installer.
* \*.rpm – Mulitple RPM files used to install docker and its dependencies.
* install.sh – The script used to install TSO500 ctDNA based on the contents listed here.

New Features:

* Initial release

Defect Repairs:

* None

Known Issues:

* Moving 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.
* The Indel Realignment and Read Stitching algorithm can produce output stitched BAM files that vary by as many as 5 reads due to a reproducibility issue with sorting order, which can lead to variation in DNA QC Metrics less than .01%
* Across multiple analyses on the same compute environment, the phased variant algorithm can produce different variant calls for variants with equal levels of supporting evidence within EGFR exon 19 region (seen in less than 1/100 samples)
* Performance not verified using reads other than 2 x 151, paired end, dual index.

Product Limitations:

* The sample sheet must be configured as described in the User Guide and the template in the resource bundle.
* 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.
* If more than one user would like to run the Self-Test, they will need to either delete the /staging/tmp directory or change the permissions so that all users can write to the location using the following command: chmod 777 /staging/tmp
* Unmapped long insertions are not likely to occur on shorter indels because there is sufficient reference-matching sequence in the reads. Product claims only indels up to 25 base pairs.
* Complex variants are specifically output only for a specific region of the EGFR gene, component and phased variants would both be contained in the output
* Incorrect calculation of variant allele frequency can occur in variants near the start and end of genomic reads, but variation in read start and end positions in an enrichment assay is sufficient to make incorrect variant allele frequency in output variants a low-probability situation.
* Germline estimation uses latest publicly available population data and estimated to be representative of targeted population, the impact of rare germline mutations is expected to be limited

# Release History

| **Version** | **ER#** | **Author** | **Description of Change** |
| --- | --- | --- | --- |
| 00 | DIR Workflow | Andrea Hatlen | Initial Release |