Local UMI Error Correction App Software

Release Notes

V1.0.0

For TruSight Oncology UMI Reagents

April 10, 2018

Introduction

These Release Notes detail the key features and known limitations to software components for the Local UMI Error Correction App.

This software is intended for use with the TruSight Oncology UMI Reagents.

The software package includes:

- Local UMI Error Correction App version v1.0.0
- Bcl2fastq2 v 2.19.1
- Burrows-Wheeler Aligner (BWA) v 0.7.15

I. Local UMI Error Correction App version v1.0.0

NEW FEATURES:

- First release

DEFECT REPAIRS:

- None

NOTES:

- The UMI Error Correction App reduces the error rate in cfDNA samples meeting QC metrics to <0.007%.
- App input is a run folder or FASTQ files generated from bcl2fastq v2.18 or higher. Output is collapsed BAM, aligned BAM and stitched BAM files.

KNOWN ISSUES:

1. Analysis log files can have both Local and UTC time zones. The correct (local) time is provided in the CSV report.
2. Sample IDs may not contain special characters or spaces. Attempting to run analysis using a sample sheet with forbidden Sample IDs will provide an error and require renaming the samples to continue.
3. When using custom manifests, Median Target Coverage Values and Noise Allele Frequency metrics double count manifest regions with regions overlapping by 1 base. Custom manifest bases should either not overlap, or should overlap by 2 or more bases.
4. Different Mapping Quality scores are produced by BWA depending on the number of threads used. To cap thread count, and ensure consistent results between systems/runs, the Local override parameter BwaThreadCount can be used.
5. Memory issues during Noise Allele Frequency calculation may occur when running 48 or more samples in a single run, and may require more than the minimum RAM specification.

Release History

Version	CO#	Description of Change
00	DIR Workflow	Initial Release