

BaseSpace Sequence Hub 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 BaseSpace Sequence Hub (BSSH) UMI Error Correction App.

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

The software package includes:

- BaseSpace Sequence Hub UMI Error Correction App version v1.0.0
- Burrows-Wheeler Aligner (BWA version 0.7.15)

I. BaseSpace Sequence Hub 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 FASTQ files generated from bcl2fastq v2.18 or higher. Output is collapsed BAM, aligned BAM and stitched BAM files.

KNOWN ISSUES:

1. BSSH UMI Error Correction App analyses should be submitted at least 2 minutes apart, since submission can fail when analysis jobs are launched simultaneously.
2. Analysis log files can have both Local and UTC time zones. The correct (local) time is provided in the CSV report.
3. Sample IDs may not contain special characters and underscores. BaseSpace will automatically change these characters to dashes (-) during FASTQ generation resulting in different Sample IDs.
4. When using custom manifests, Median Target Coverage Values and Noise Allele Frequency metrics double count manifest regions with regions overlapping by 1 base. Custom manifests should either not overlap or should overlap by 2 or more bases.

Release History

Version	ER#	Description of Change
00	DIR Workflow	Initial Release