

BaseSpace Core Apps Software Release Notes

BWA Enrichment v2.1

For BaseSpace

March 4, 2015

Introduction

These Release Notes detail the key changes to software components for the BWA Enrichment BaseSpace Core App since the release of the BWA Enrichment 2.0 app. This v2.1 release enables upload of custom manifest files (e.g., probes and targeted regions files), generated by Illumina DesignStudio upon ordering a custom enrichment panel.

This app is deployed on BaseSpace Cloud.

For more information about this app and how to use it, refer to the app User Guides, available from the details page of each app, and the BaseSpace Support Page, Documentation and Literature, on illumina.com.

http://support.illumina.com/sequencing/sequencing_software/basespace/documentation.ilmn

The software package includes:

- BWA Enrichment v2.1

I. BWA Enrichment v2.1

NEW FEATURES:

- Updated to support multi-node launch for up to 96 samples. Users can expect to see a marked improvement in time-to-results for analysis of multiple samples.
- Renamed workflow setting name in PDF report from "Trim Nextera Rapid Capture Adapters" to "Trim Adapters".

REMOVED FEATURES:

- Aggregate summary output files including csv, pdf and html are no longer provided.

DEFECT REPAIRS:

- The manifest for Nextera Rapid Capture Exome manifest v1.2 was replaced to match the released version.
- Fixed an issue with annotation and report generation: previously, the app would attempt to report and annotate variants in regions outside of the targeted region. This issue has been fixed and this will no longer occur. Set depth threshold to 1 for generation of gaps files. This ensures that the gaps file only contains zero-coverage regions.

KNOWN ISSUES:

- The vertical bins in the Depth of Sequencing Coverage Table are off by a value of 1 in the pdf report.

II. BWA Enrichment v2.0

NEW FEATURES:

- Includes BWA-MEM as the default aligner. This is generally recommended for high-quality alignment as it is faster and more accurate. BWA-MEM also has better performance than BWA-backtrack for 70-100bp Illumina reads.
- Updated the following module and algorithm components:
 - GATK (Variant Caller) 1.6-23-gf0210b3
 - SAMtools 0.1.19-isis-1.0.1
- Ability to use custom manifest files which have been imported to the BaseSpace project of interest.
- Updated to Illumina Annotation Service (IAS) version 3, which includes improved IAS annotation stability and robustness.
- Added tags to VCF header (e.g. annotator, variant caller version number).
- Only on-target variants are now reported.
- Improved logging and renamed workflow log files as WorkflowLog.txt and WorkflowError.txt.
- Improved reporting of software version numbers in PDF report.
- Improved naming of workflow steps to provide greater consistency. (e.g. "Statistics generation" is now used instead of the previous "Generate statistics").
- Updated PDF report content (plots, notes, stats). See the User Guide for more details.
- Remove minimum depth threshold for variant calling to increase accuracy.

DEFECT REPAIRS:

- A fix was made to the integration of IAS and the ISAAC Enrichment v2 app so that Deflate Stream errors will now longer occur.
- A fix was made to the reporting of adapter counts and resulting adapter trimming statistics. Trimming is now reported only for reads passing filter (PF) instead of all reads.
- A fix was made so that in the rare instance when a bam file containing no aligned reads is generated, this can be properly handled.

A fix was made to the bam statistics generation to improve robustness of this operation by adding retry logic.

KNOWN ISSUES:

- None