BaseSpace Core Apps Software Release Notes

Amplicon DS v1.1

For BaseSpace

December 15, 2014

Introduction

The Amplicon DS app enables analysis of the TruSight Tumor samples. By using information from both strands, the app is able to employ a proprietary variant calling algorithm that is robust to degradation errors in FFPE or similarly compromised samples.

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:

- Amplicon DS v1.1

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New Features:

- Initial Release: see the User Guide for complete guidance on application features and functionality.
- This app includes expanded phased SNP calling capabilities that allows the identification of different, non-adjacent SNPs (up to 5 bases apart).

Defect Repairs:

- N/A

Known Issues:

- SNVs reported in the combined VCF output files may occasionally include filtered SNVs that should have been omitted from the gvcf due to being below the emit-cutoff, so they occurred at insufficient sequencing depth (<300x). These SNVs will be flagged as filtered in the combined VCF. These SNVs can be removed using Variant Studio by adding a depth filter of 300. Very few of the SNVs included in the combined VCF are expected to fall into this category.