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BaseSpace Core Apps Software Release Notes

TruSeq Phasing Analysis v1.1

For BaseSpace

February 23, 2015

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Introduction

These Release Notes detail the key changes to software components for the TruSeq Phasing Analysis v1.1 App. This v1.1 release consists mainly of changes to summary statistics reporting, input VCF file filtering and general code stability.

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:

• TruSeq Phasing Analysis v1.1

I. TruSeq Phasing Analysis v1.1

New Features:

- General stability and robustness improvements.
- Faster start-up time of the app without having to pre-download input files
- Gene stats now calculated only on autosomes for consistency.
- Variant counts and %ICF calculations reported in the phasing metrics section and in library characteristics file now only calculated against chromosomes for which phasing was attempted.
- Increased code stability when number of variants in input VCF is low.
- Change in the calculation of number of aligned reads. Affects the %mapped reads metric displayed in the PDF file.
- Improved app logging. Please see user guide for description of log file contents

DEFECT REPAIRS:

- Fixes were made to the verification of input VCF file format and content so that they now adhere to the rules outlined in the user guide.
- A fix was made in the calculation of the percent of genes phased. Previously, the number of genes considered for phasing estimation (the denominator), was not consistently calculated. This is issue is now resolved.

KNOWN ISSUES:

• TruSeq Synthetic Long-Read Samples that are merged in BaseSpace are not compatible with the app.