

# BaseSpace Core Apps Software Release Notes

## mtDNA Variant Analyzer

### for BaseSpace

**April 23, 2016**

FOR RESEARCH USE ONLY

© 2015 Illumina, Inc. All rights reserved.

Illumina, 24sure, BaseSpace, BeadArray, BlueFish, BlueFuse, BlueGnome, cBot, CPro, CytoChip, DesignStudio, Epicentre, GAllx, Genetic Energy, Genome Analyzer, GenomeStudio, GoldenGate, HiScan, HiSeq, HiSeq X, Infinium, iScan, iSelect, ForenSeq, MiSeq, MiSeqDx, MiSeq FGx, NeoPrep, Nextera, NextBio, NextSeq, Powered by Illumina, SeqMonitor, SureMDA, TruGenome, TruSeq, TruSight, Understand Your Genome, UYG, VeraCode, verifi, VeriSeq, the pumpkin orange color, and the streaming bases design are trademarks of Illumina, Inc. and/or its affiliate(s) in the U.S. and/or other countries. All other names, logos, and other trademarks are the property of their respective owners.

## Introduction

- I. These Release Notes detail notable items for the mtDNA Variant Analyzer BaseSpace Core App v1.0.0 release.

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).

[http://support.illumina.com/sequencing/sequencing\\_software/basespace/documentation.ilmn](http://support.illumina.com/sequencing/sequencing_software/basespace/documentation.ilmn)

The mtDNA Variant Analyzer app, in conjunction with the mtDNA Processor, allows for variant analysis and easy visualization of mitochondrial sequence data. This workflow can analyze any part of the full circular genome, without any origin dead zone, using quality and coverage thresholds customized by the user. Generate Excel reports to view data outside of BaseSpace. FASTQ files from the sequencer are processed into VCF files using the mtDNA Processor app. The VCF output is then displayed in the web-based mtDNA Analyzer app for visualization and optional report generation in an Excel file format.

The software package includes:

- mtDNA Variant Analyzer (initial release)

## II. mt Variant Analyzer

### NEW FEATURES:

- Use of BaseSpace allows data from any Illumina sequencer to be used
- Comparison tool allows thousands of samples to be viewed at once or seen against the rCRS reference
- User modifications can be saved
- Custom thresholds and manifests are accepted for flexibility
- Variant reports can be exported as an Excel file for easy data review

### KNOWN ISSUES:

- The Zero Differences filter of Sample Compare Mode will remain applied after switching to the Reference Genome Compare Mode. To rectify, enter Sample Compare Mode and clear the Zero Differences filter.
- The Sample Name shown has spaces replaced with underscores and an "\_S1" appended to the end of the name.
- When the authentication to BaseSpace has expired, the prompt to log back in to BaseSpace will present in a new browser window.

- The User Modified indicator will remain present after returning modified calls to the original call from the mtDNA Variant Processor.