

GenomeStudio Software 2.0

Release Notes

GenomeStudio 2.0

Genotyping Module 2.0

Polyploid Genotyping Module 2.0

Introduction

These Release Notes detail the key changes to the GenomeStudio Software since the release of GenomeStudio Software v2011.1. This is a required software update for genotyping customers currently using GenomeStudio Software v2011.1, as this release includes important features and bug fixes outlined below.

Note that GenomeStudio Software v2.0 only includes the Genotyping Module and the Polyploid Genotyping Module. Users interested in running the Methylation Module and the Gene Expression Module should continue to run GenomeStudio Software v2011.1. GenomeStudio Software v2.0 and GenomeStudio Software v2011.1 can be installed and run simultaneously on the same system.

Genotyping Module 2.0

IMPROVEMENTS:

- Added new clustering and genotyping option, GenTrain 3.0
- Added support for clustering on Infinium XT products
- Improved accuracy for sample normalization of Infinium I bead types
- Added support for NextSeq 550 data
- Added ability to set genotyping and clustering intensity thresholds:
 - Genotyping intensity threshold (one per project) filters samples prior to clustering process
 - Clustering intensity threshold (one per locus) filter samples prior to genotyping
- Improved user interface for folder selection
- Added support for multi-species products when creating new projects from Illumina LIMS

DEFECT REPAIRS:

- Addressed issue with incorrect handling of intensity data for bead types with a small number of non-outlier beads
- Addressed issues with viewing of text reports after report generation

KNOWN ISSUES:

- Software installer does not create a desktop shortcut. Note that GenomeStudio Software 2.0 is installed by default to "C:\Program Files\Illumina\GenomeStudio 2.0\GenomeStudio.exe", while GenomeStudio Software 2011.1 is installed by default to "C:\Program Files (x86)\Illumina\GenomeStudio\GenomeStudio.exe".
- The Genotyping Module uses heterozygosity of SNPs on the X chromosome to estimate the gender of the sample. Female samples for which the heterozygosity of SNPs on the X chromosome is low may be incorrectly called as males. This can occur when X chromosome SNPs have a low average minor allele frequency (MAF) or when

the sample has a substantial loss of heterozygosity (LOH) whether copy neutral or not.

- Intensity Only status of loci is ignored when using cluster files that specify genotypes for Intensity Only loci
- Box plots may be slow to load and cause the software to crash

OTHER:

- Removed support for Illumina Controls Database
- Removed support for custom cluster definitions for polyploid loci. For polyploid loci, please use the Polyploid Genotyping module
- Removed GoldenGate Contamination Dashboard
- Removed cluster definition export to Illumina LIMS
- Removed license key requirements for installation. Registration using MyIllumina credentials will be required upon first launch of the software. Users without internet connectivity on the computer where GenomeStudio 2.0 is installed should contact Illumina tech support

NOTES:

- New GenomeStudio 2.0 projects and cluster files cannot be opened in GenomeStudio 2011.1. However, projects and cluster files created from GenomeStudio 2011.1 can be opened in GenomeStudio 2.0

Polyploid Genotyping Module 2.0 (formerly Polyploid Clustering Module)

IMPROVEMENTS:

- Renamed from "Polyploid Clustering Module" to "Polyploid Genotyping Module"
- Added automatic per-sample genotype calling for data originating from polyploid organisms
- Added PolyGentrain algorithm for generating polyploid genotyping clusters
- Added new clustering options to specify level of ploidy (e.g. tetraploidy, hexaploidy, octaploidy), type of ploidy (i.e. autopolyploid vs. allopolyploid), and whether or not samples are obtained from an inbred population
- Improved accuracy for sample normalization of Infinium I bead types
- Added support for NextSeq 550 data
- Added ability to set genotyping and clustering intensity thresholds:
 - Genotyping intensity threshold (one per project) filters samples prior to clustering process

- Clustering intensity threshold (one per locus) filter samples prior to genotyping
- Improved user interface for folder selection
- Added support for multi-species products when creating new projects from Illumina LIMS
- Added text report generation

DEFECT REPAIRS:

- Address issue with incorrect handling of intensity data for bead types with a small number of non-outlier beads

KNOWN ISSUES:

- Genotype scores associated with no-calls may be represented as a negative number

OTHER:

- Removed OPTICS clustering algorithm

NOTES:

- Cluster files generated from Polyploid Clustering Module 1.0 cannot be used with the Polyploid Genotyping Module 2.0 software. Likewise, cluster files generated from Polyploid Genotyping Module 2.0 cannot be used with the Polyploid Clustering Module 1.0 software