

GenomeStudio Software 2.0.3

Release Notes

GenomeStudio 2.0.3

Genotyping Module 2.0.3

Polyploid Genotyping Module 2.0.3

Introduction

These Release Notes detail the key changes to the GenomeStudio Software since the release of GenomeStudio Software v2.0.2. This is a required software update for genotyping customers currently using GenomeStudio Software v2.0.2, as this release includes important features and bug fixes outlined below.

Note that GenomeStudio Software v2.0.3 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 v2.0.3 and GenomeStudio Software v2011.1 can be installed and run simultaneously on the same system.

Genotyping Module 2.0.3

IMPROVEMENTS:

- Improved gender estimation algorithm to improve accuracy in situations where the heterozygosity of SNPs on the X chromosome is low due to low average minor allele frequencies (MAF) or when the sample has a substantial loss of heterozygosity (LOH).
- Added support for the hg38 reference genome in the Illumina Genome Viewer

KNOWN ISSUES:

- 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

Polyploid Genotyping Module 2.0.3

No changes relative to Polyploid Genotyping Module 2.0.2

KNOWN ISSUES:

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