

GenomeStudio Software 2.0.2

Release Notes

GenomeStudio 2.0.2

Genotyping Module 2.0.2

Polyploid Genotyping Module 2.0.2

Introduction

These Release Notes detail the key changes to the GenomeStudio Software since the release of GenomeStudio Software v2.0.0. This is a required software update for genotyping customers currently using GenomeStudio Software v2.0.0, as this release includes important features and bug fixes outlined below.

Note that GenomeStudio Software v2.0.2 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 v2.0.2 and GenomeStudio Software v2011.1 can be installed and run simultaneously on the same system.

Genotyping Module 2.0.2

IMPROVEMENTS:

- Improved performance of final report generation when writing directly to network drives

DEFECT REPAIRS:

- Fixed issue where the software installer would not create a desktop shortcut.
- Fixed issue where projects created with the "Pre-Calculate" option selected would not update genotypes when using the "Set Genotyping Intensity Threshold for Selected SNP" feature.

KNOWN ISSUES:

- 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

Polyploid Genotyping Module 2.0.2

No changes relative to Polyploid Genotyping Module 2.0.0

KNOWN ISSUES:

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