# GenomeStudio™ Software v2011.1 Release Notes

## Release Highlights

1. Methylation module now supports the Illumina LIMS workflow.
2. The performance for genotyping project creation has been improved.
3. New polyploidy features have been added into the Genotyping module.
4. DNA and RNA Sequencing modules are compatible with CASAVA1.8 output data set.
5. Online help is available for the GenomeStudio Framework and GenomeStudio Methylation Module.

NOTE: This release notes contains new features and changes since the previous GenomeStudio release (GenomeStudio v2010.3). Please refer to the GenomeStudio User Guide and previous release notes (GenomeStudio v2010.3, GenomeStudio v2010.2 Release Notes, GenomeStudio v2010.1 Release Notes, GenomeStudio v2009.1 Release Notes and GenomeStudio v2009.2 Release Notes) for instruction and changes since the GenomeStudio v2008.1 release.

# 1. Genotyping Module v1.9

1.1. The performance for genotyping project creation has been improved.

1.1.1. Please refer to the performance benchmark for the detail information.

1.2. Please refer to the recommended workflow tech note to create infinium based project in GenomeStudio.

1.3. The following new polyploidy features have been added:

1.3.1. Users are able to define and edit the customer cluster in the SNP graph.

1.3.1.1. After changing the Custom Cluster number in the Full Data table, go to SNP graph and select the samples, and define the genotype from the context menu. The ellipse will be drawn for each defined polyploidy cluster.

1.3.1.2. Edit the customer cluster size and position as the normal cluster in the SNP graph.

1.3.2. Users are able to save custom cluster definitions to the project that will load when the project is opened later.

1.3.3. Users are able to export the custom cluster file.

1.3.3.1. Choose File | Export Cluster Position to export

1.3.4. Added "**Custom Cluster #**" column to the SNP Table

1.3.4.1. The value in the column is synchronized with the "**Customer Cluster#**" in the Full Data table.

1.3.5. Users are able to import that custom cluster file to another Genotyping project

1.4. Added option to use plus/minus strand when importing or exporting allele calls .

1.4.1. Added "Plus/Minus Alleles" sub-column in Full Data table

1.5. XY CNV regions are now shown in the Genome Viewer. This is a known issue in the previous release.

1.6. Resolved issue in previous release having to do with to calculating the **Chi100**, **HetExcess** and **HW Equil**.

1.7. Resolved the known issue in last release (GenomeStudio v2010.3): p05 Grn, p50 Grn, p95 Grn, p05 Red, p50 Red, and p95 Red in Sample Table did not get updated in the project that was saved with the "Save reduced snp set" option from original project..

1.8. Resolved known issue that caused 0.1% error in the calculation for the "**Rep Error Rate**"

in the Samples Table.

1.8.1. The value will match the data in the Reproducibility report.

## 2. Methylation Module v1.9

2.1. Illumina LIMS workflow is now available for Methylation Infinium HD assay:

2.1.1. For detail usage, please refer to the Methylation Module online help.

2.1.2. Check the option "Use LIMS to create project" option to enable the LIMS connection.

2.1.3. Create project from LIMS option: will load the project from Illumina LIMS.

2.1.4. Create only sample sheet option: from LIMS will create just a sample sheet from LIMS without creating a project and running analysis.

2.1.5. Create project from LIMS sample sheet option: will allow user to browse to select a LIMS sample sheet to create the Methylation project.

2.1.6. Apply filter file option is used to apply sample filter file.

2.1.6.1. This is a text file that contains list of sample ID to exclude from the project.

2.1.6.2. One sample ID per line in the filter file. Check online help for a sample filter file.

2.1.7. Duplicated sample names from LIMS are modified to make them unique. If there are three identical sample names, the first one will remain the same, the second and the third ones will get "_2" and "_3" suffixes respectively.

2.2. The issue of "Insufficient system resources exist to complete the requested service" when creating a project over network has been resolved.

## 3. Gene Expression Module v1.9

3.1. Resolved the issue of "Insufficient system resources exist to complete the requested service" when creating a project over network.

## 4. DNA and RNA Sequencing Modules v1.9

4.1. New option "*Show Density Plot for Regions Depth Above*" is added in the project creation wizard.

4.1.1. This option will show a histogram plot in IGV when the stack alignment is higher than the specified value. The default value is 1000.

4.2. If the folder which contains the BAM data in a project is moved , you will be prompted to browse and select the new folder location in order to open the sequencing project.

4.3. You cannot use the CASAVA build and the generic bam dataset in the same project.

4.4. If you change the BAM folder name, you are not able to open the GenomeStudio projects created before using the BAM file.

4.5. If you use CASAVA1.8 build in GenomeStudio, it is expected that the Lanes table is gone, and the Samples table display 0 value in the columns.

4.6. Known issue: Jump to splice mate does not function as expected in IGV.

4.7. Known issue: you are not able to add new columns in Allele and Indel tables (through data track, new function calculation, or import function).

## 5. ChIP Sequencing Modules v1.9

5.1. Known issue: You cannot create a ChIP seq project using CASAVA 1.8. This is because CASAVA1.8 no longer generates the sorttd.txt file from Gerald, which is required to create the ChIP seq project in GenomeStudio. To create a ChIP seq project, you must create a sort.txt file. One method of doing that is to use CASAVA 1.7.

5.1.1. You can view the stack alignment read using the CASAVA1.8 build, without the peak and region finder function, by loading the bam data into DNA sequencing module.

5.1.2. The Chip Seq module will support the bam input in the future release.