GenomeStudio™ Software 2008.1 Release Notes

These release notes provide information about known issues in the commercial release of GenomeStudio Software 2008.1. In addition, new GenomeStudio features significantly different than those in BeadStudio are listed. Consult the following GenomeStudio Software User Guides for details about each module:

Module	User Guide	Part Number and Revision
GS	*GenomeStudio 2008.1 Framework User Guide*	#11318815 Rev. A
GT	*GenomeStudio Genotyping Module v1.0 User Guide*	#11319113 Rev. A
GX	*GenomeStudio Gene Expression Module v1.0 User Guide*	#11319121 Rev. A
M	*GenomeStudio Methylation Module v1.0 User Guide*	#11319130 Rev. A
PT	*GenomeStudio Protein Analysis Module v1.0 User Guide*	#11319148 Rev. A
CS	*GenomeStudio ChIP Sequencing Module v1.0 User Guide*	#11319084 Rev. A
DS	*GenomeStudio DNA Sequencing Module v1.0 User Guide*	#11319092 Rev. A
RS	*GenomeStudio RNA Sequencing Module v1.0 User Guide*	#11319105 Rev. A

1.   General/Framework

1.1.   Microsoft .NET 3.5 is required on the workstation where you plan to install GenomeStudio Software 2008.1. .NET 3.5 is available as a free download from Microsoft: http://www.microsoft.com/downloads/details.aspx?FamilyId=333325FD-AE52-4E35-B531-508D977D32A6&displaylang=en.

1.2.   After starting GenomeStudio Software 2008.1 for the first time, you will be asked to register your software. If you choose not to register, a registration reminder will pop up each time you open the application.

1.3.   Once you install GenomeStudio Software 2008.1, GenomeStudio software becomes the primary application associated both with BeadStudio projects and GenomeStudio projects (designated as *.bsc files).

  1.3.1.   To change *.bsc file association from GenomeStudio software back to BeadStudio, right-click a *.bsc file and select **Open With** | **BeadStudio**.

1.4.   A conversion tool that allows you to automatically convert BeadStudio projects to GenomeStudio projects has been built into GenomeStudio software. A similar tool that allows you to convert GenomeStudio projects to BeadStudio projects is built into the BeadStudio July 2008 release (GT v3.3, GX v3.4).

1.5.   If GenomeStudio detects an error, you are given the option to send an error report directly to Illumina Technical Support.

  1.5.1.   You can choose simply to submit the error, or to submit the error and receive feedback or technical support from Illumina.

1.6. You can also submit comments to Illumina by selecting **Help** | **Submit a comment**.

    1.6.1. Submit comments in the following categories:
Feature Request, Comment, Sales Question, Other.

    1.6.2. You have the option to attach files to your comments.

    1.6.3. You can request a response to your comment from an Illumina representative.

1.7. Data import speed and project creation time is variable depending upon network connection speed, disk speed of the local machine, and location of the data repository.

1.8. Installing GenomeStudio Software 2008.1 on a Windows Vista workstation does not require you to manually uninstall previous versions or modules of GenomeStudio software.

1.9. When using the line plot, the zoom level may be different depending on whether you zoom into the X-axis or the Y-axis.

1.10. Progress bars have been added to histograms and line plots while data is loading.

1.11. The **Jump to IGV** feature has been added to various tables in both sequencing and microarray modules. This feature allows you to right-click any row in a table with chromosome names and positions and jump to the corresponding location in the IGV.

Jump to IGV is available in the following GenomeStudio sequencing module tables:

	Sequences Table	Alleles Table	Genes Table	Exons Table	Junctions Table	Regions Table	Merged Regions Table	Peaks Table
ChIP Sequencing Module	X					X	X	X
DNA Sequencing Module	X	X						
RNA Sequencing Module	X	X	X	X	X			

Jump to IGV is available in the following GenomeStudio microarray module tables:

	Full Data Table	SNP Table	Paired Sample Table	Probe Tables
Genotyping Module	X	X	X	
Gene Expression Module				X
Methylation Module				
Protein Analysis Module				

2. Illumina Genome Viewer (IGV)

2.1. The features available in the IGV in GenomeStudio microarray modules are different than the features available in GenomeStudio sequencing modules. Please consult the *GenomeStudio 2008.1 Framework User Guide* to learn about these differences.

2.2. In GenomeStudio microarray modules, when you open the IGV, you are automatically directed to the **Add Favorite Data Plots** form.

2.3. Due to the extremely large amount of data included in the Sequences table (sequencing modules), IGV data plots may be drawn noticeably slower than data plots created using other tables.

2.4. When resizing the IGV horizontally or vertically, the zoom level changes correspondingly.

2.5. When scrolling vertically at the base-level view, there may be a delay if the stacked alignment contains an excessively large number of reads.

2.6. When scrolling horizontally at the base-level view, the vertical order of reads may change as additional reads are brought in or out of view.

2.7. Tooltip information is only available on data plots in microarray modules.

2.8. Alignment cursors have been added to the **View** menu in the Illumina Chromosome Browser (ICB). This allows you to create vertical lines to align data or stacked alignment plots with the chromosome and with annotation tracks.

2.9. The Data Plots window now contains the following buttons: **Add**, **Delete**, **Check Plots** and **Uncheck Plots**.

2.10. In stacked alignment view in the sequencing modules, a red number showing the highest peak (number of reads) for that given location in the genome is displayed.

2.11. For large Genotyping projects (>500,000 SNPs), only the first chromosome is displayed in the Genome Viewer, instead of the Whole Genome View.

3. Sequencing Modules

3.1. General

3.1.1. Due to the large amount of data included in the Sequences table (sequencing modules), table functions (including graphing functions) in the Sequences table may be noticeably slower than the same functions in other tables.

3.2. DNA and RNA Sequencing Modules

3.2.1. There are two options for creating ADT (Assay Design Tool) Reports:

3.2.1.1. The **ADT dbSNP Report** provides start and stop positions for a region flanking the SNP.

3.2.1.2. The **ADT Sequence Report** provides the actual sequence for the region flanking the SNP.

3.2.2. The header in the ADT report has been made optional by adding a checkbox in the report wizard.

3.3. ChIP Sequencing Module

3.3.1. When creating a new ChIP Sequencing project, the calculation for regions and peaks can be disabled. To load your reads directly into the module, clear the Automatically Calculate Regions and Peaks checkbox.

3.3.2. To calculate the regions and peaks after the project creation, go to **Analysis** | **Run Regions and Peaks Finder**.

3.3.3. The counting algorithm for ChIP Sequencing has been modified to report actual counts rather than summed counts.

    3.3.3.1. The counts displayed in the Regions table reflect the total number of bases seen when displaying the region/peak region in the IGV.

    3.3.3.2. The values displayed in the Peaks table reflect summed counts divided by the region window size.

3.3.4. Only regions which are greater than or equal to the region finder window size are shown in the Regions table.

3.3.5. A new Merged Regions table has been added, which combines regions that have less than a user-specified base pair distance between them. Go to **Windows** | **Merged Regions** to display this table.

3.3.6. The Peaks table includes a column specifying where the peak was found.

    3.3.6.1. **Jump to Region** allows you to jump from a peak in the Peaks table to the region (in the Regions table) where it was found.

3.3.7. Regions which have counts slightly larger than the region count threshold (default 200) may not show up in the Regions Table. A possible workaround is to enter a smaller count threshold (30-40 base pairs smaller) than the desired value.

4. Microarray Modules

4.1. Genotyping Module

4.1.1. A slight difference seen in normalization between 32-bit and 64-bit systems in BeadStudio software has been resolved in GenomeStudio software. You should now see virtually no difference between normalized values (and subsequent calculations such as call rates) on 32-bit and 64-bit systems.

4.1.2. Data from Illumina Human-1M and Human-1M-Duo BeadChips can now be combined in the same GenomeStudio project.

    4.1.2.1. Projects with multiple versions of a manifest can now only be created on 64-bit workstations with 4+GB of RAM. (This includes CNV-370/CNV-370 Quad, 550-Duo/610 Quad, and 1M/1M-Duo.)

4.1.3. An issue regarding SAM-OPA association being inverted when loading a GoldenGate® multiple-OPA project on 64-bit workstations has been resolved.

4.1.4. A heat map is now automatically created during project creation.

4.1.5. When reopening a Genotyping project, the heat map window is disabled. You can reactivate the heat map by clicking in the window.

    4.1.5.1. In large Genotyping projects, the heat map may become blank due to the large amount of data.

4.2. Gene Expression Module

4.2.1. An issue seen when excluding probes with missing data with certain Human-HT12 BeadChips has been resolved. You can now choose to impute or exclude probes with missing data.

4.2.2. A **Select All** button has been added to the Gene Expression Final Report to enable you to select all columns, instead of manually choosing each column.

4.3. Methylation Module

4.3.1. An issue regarding creating dendrogram plots in GoldenGate and VeraCode® Assay types has been resolved.

4.3.2. The parameter Bead_StdErr_B has now been fixed to use NBEADS_B instead of NBEADS_A.

4.3.3. An issue regarding importing columns into the Samples Table for GoldenGate and Infinium® Methylation products has been resolved.

4.4. Protein Analysis Module

4.4.1. An issue involving creating a Protein Analysis project in BeadStudio has been resolved.