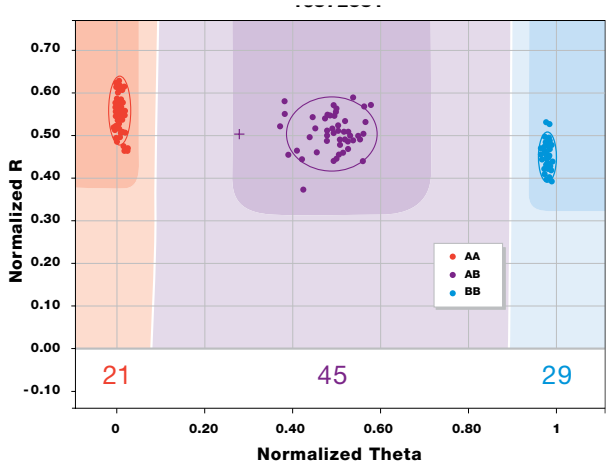
Illumina® BeadStudio Data Analysis Software Modules

Illumina's BeadStudio delivers high-quality software for cutting-edge data analysis and advanced visualization tools for the following applications: Genotyping, Gene Expression, and Loss of Heterozygosity (LOH).

Highlights of Illumina Beadstudio Software

- **Cutting Edge:**
  powerful data manipulation, heat map and cluster analysis
- **See Your Data in a New Way:**
  graphical analysis using the Illumina Genome Viewer (IGV) and Illumina Chromosome Browser (ICB)
- **High Performance:**
  sophisticated algorithms to support of genomic investigation
- **Integrative:**
  common framework that supports multiple application modules
- **Freedom for Downstream Analysis:**
  export data for use in downstream applications (e.g., SpotFire® or GeneSpring®)

Figure 1: Genoplot Generated Using Beadstudio Genotyping Module



The BeadStudio Genotyping Module clusters genotype calls into AA, AB and BB clusters for easy viewing and data analysis.

Introduction

As high-throughput systems enable increasingly complex bioinformatics data to be generated, researchers turn towards managing, processing, and analyzing their data. Thus, there is a need for a state-of-the-art software package that combines ease-of-use with powerful visualization tools to streamline the analysis process. Illumina is proud to introduce its leading-edge analytical software platform, BeadStudio, and its associated analytical modules (GT, GX, LOH Plus).

BeadStudio's modular design allows Illumina to respond to evolving customer needs by releasing add-on software modules that integrate seamlessly into the BeadStudio Framework.

As new assays or applications are developed, the BeadStudio product family will continue to support scientists' most current needs. This modular approach also enables users to rely on a familiar common graphical user interface, minimizing the need for new software training as additional modules are released. In addition, the BeadStudio architecture is designed to support the flow of information for downstream data compatibility with other applications (e.g., SpotFire or GeneSpring).

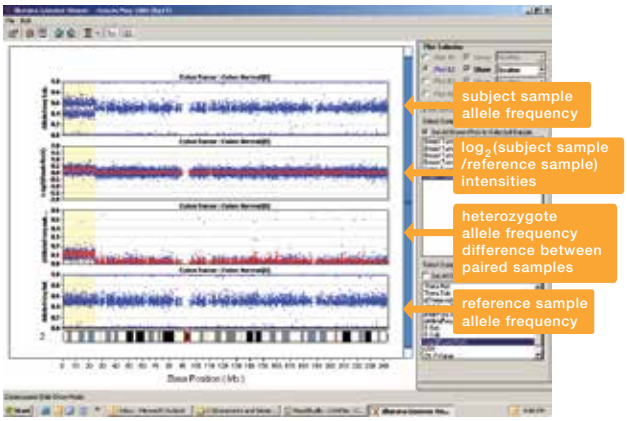
Genotyping (GT) Module

The BeadStudio GT Module provides automated genotype calling and sample clustering. Powerful visualization tools and quality-control features enable graphical output of data and reports on reproducibility and Mendelian consistency. Bead Studio Framework features such as heat maps, scatter plots, the Illumina Genome Viewer (IGV), the Illumina Chromosome Browser (ICB), the Illumina Sequence Viewer (ISV) and Infinium Laboratory Information Management System (LIMS) integration add additional functionality to this module.

Gene Expression (GX) Module

The BeadStudio GX Module is designed for analysis of Whole-Genome Gene Expression BeadChips and Focused Arrays for the Direct Hyb and DASL® Assays. Advanced analytical features include gene-level analysis, statistical tools for differential expression analysis, sample controls, heat maps (Figure 3), bar plots and clustering tools. Enhanced data management enables hierarchical organization of samples, groups, groupsets and associated project analysis.

Figure 2: LOH Plus Module



The Illumina Genome Viewer enables classic LOH analysis by comparing reference and subject sample data from the same donor.

Loss of Heterozygosity Plus (LOH Plus) Module

The BeadStudio LOH Plus Module is designed to address chromosomal aberrations and allelic imbalance in paired samples (e.g., matched normal/tumor pair). Key features of this module include: automated detection of chromosomal aberrations, custom bookmarking of genomic regions of interest and a chromosomal heat map to reflect LOH/Copy Number-related intensity differences along chromosomes. The Bookmarked Samples Report also allows the user to create a summary of various aberrations.

Beadstudio Framework Features

The BeadStudio Framework is the basis of BeadStudio and includes many customizable features and formats for powerful data analysis.

Project Wizard

The BeadStudio Project Wizard helps the user initiate and organize projects with ease. Using the Project Wizard simplifies decision-making so less time is required for setup, allowing more time for analysis.

Illumina Genome Viewer (IGV)

The IGV allows visualization of sample-related data on a genome-wide scale or at the chromosomal level. The IGV can display up to four plots at a time (a single data series each) over a chosen chromosome (Figure 2).

Illumina Chromosome Browser (ICB)

Using the ICB, the user can browse imported SNPs within the context of a chromosome (e.g., SNPs relative to exons). Both the IGV and ICB offer a wide range of visualization options for data analysis.

Data Tables

Advanced data table features allow the user to customize data viewing for efficient and effective data analysis. Options include:

- **importing columns**
- **adding derived columns**
- **sorting**
- **selecting and marking rows**
- **advanced data filtering**
- **exporting data**

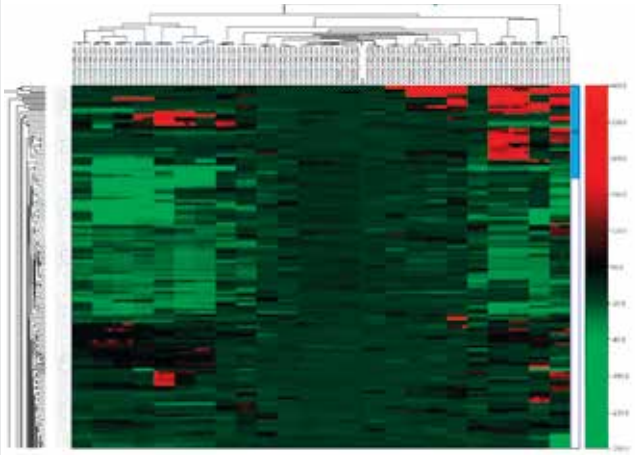
Reports

Custom or standard reports can be created for various types of analysis. Implementing consistent report formats can streamline analysis and discovery.

Table 1: Beadstudio Tools

Feature	Highlight
BeadStudio Project Wizard	Easy project set-up and organization
Illumina Genome Viewer	View data patterns across the genome
Illumina Chromosome Browser	Browse data at the chromosome and gene level
Illumina Sequence Viewer	Browse data at the sequence level
Table Filters	Configure with easy-to-use, rule-based options
Import Data	Import cluster files, import columns into tables
Export Data	Export cluster files, export table data
Generate Reports	Create standard or custom reports
Scatter Plots	For graphical viewing of data
Heat Map	For graphical viewing of data
LOH/Copy Number	Analyze Loss of Heterozygosity/Copy Number
Auto Detection	Search for chromosomal aberrations (in addition to manual curation)
Data Integration	Customize reports for integration into other downstream applications

Figure 3: Gene Expression Module



The heat map dendrogram clusters rows (Target ID) and columns (Differential Scores). Using the Heat Map tools within the Gene Expression Module allows the investigator to easily visualize and analyze large amounts of data.

Graphs

BeadStudio graphic display options allow the user to plot table data in a number of ways, including the following formats:

- **Histograms**
- **Line Graphs**
- **Scatter Plots**
- **Heat Maps**
- **Cluster Plots**
- **Many More Module-Specific**
- **Capabilities**

Data Integration

Customizable reports and data export options simplify integration with other downstream analytical applications.

Summary

BeadStudio is designed to be leading-edge, modular and integrative. BeadStudio consists of individual software modules specific to various product applications. Users can rely on the familiar BeadStudio Framework while minimizing additional product training as they expand their analytical capabilities from a single module to multiple modules. BeadStudio is designed for downstream data compatibility with other applications. This modular architecture enhances the flow of information from data analysis to downstream scientific discovery.

Additional Information

Visit www.illumina.com or contact us at the address below to learn more about BeadStudio or other solutions from Illumina.

Table2: Hardware and Software Requirements

Feature	Highlights
CPU Speed	Intel® Pentium® 2.0 GHz or above
Memory Size	≥ 2 GB
Hard Drive	≥ 100 GB
Video Display	1280 x 1024 recommended, 1024 x 768 required
Monitor Display	LCD or CRT 17” recommended
Operating System	Windows XP - SP2 (32bit) or Windows XP - SP2 (64 bit)
Specific OS Requirements	Requires Microsoft .NET framework 1.1 or above

Ordering Information

Catalog No.	Product	Description
SW-10-101	BeadStudio Genotyping (GT) Module (Single Seat) - New License	New single-seat license for BeadStudio Genotyping (GT) Module provides advanced tools for SNP data analysis.
SW-10-102	BeadStudio Genotyping (GT) Module (Five Seats) - New License	New single-seat license for BeadStudio Genotyping (GT) Module provides advanced tools for SNP data analysis.
SW-10-200	BeadStudio Genotyping (GT) Module (Single Site w/ Warranty) - Upgrade	Single-site upgrade license for systems with warranty coverage. BeadStudio Genotyping (GT) Module provides advanced tools for SNP data analysis.
SW-10-300	BeadStudio Genotyping (GT) Module (Enterprise w/ Warranty) - Upgrade	Enterprise upgrade license for system with warranty coverage. BeadStudio Genotyping (GT) Module provides advanced tools for SNP data analysis.
SW-10-400*	BeadStudio Genotyping (GT) Module (Five Seats w/Warranty) - Upgrade	Complimentary five-seat upgrade license for system with warranty coverage. BeadStudio Genotyping (GT) Module provides advanced tools for SNP data analysis.
SW-20-101	BeadStudio Gene Expression (GX) Module (Single Seat) - New License	New single-seat license for BeadStudio Gene Expression (GX) Module provides advanced tools for gene expression data analysis.
SW-20-400*	BeadStudio Gene Expression (GX) Module (Five Seats w/Warranty) - Upgrade	Complimentary five-seat upgrade license for system with warranty coverage. BeadStudio Gene Expression (GX) Module provides advanced tools for Gene Expression data analysis.
SW-30-101	BeadStudio LOH Plus Module (Single Seat) - New License	New single-seat license for BeadStudio Loss of Heterozygosity Plus (LOH Plus) Module provides advanced tools for LOH/Copy Number data analysis.
SW-30-400*	BeadStudio LOH Plus Module (Five Seats w/Warranty) - Upgrade	Complimentary five-seat upgrade license for system with warranty coverage. BeadStudio Loss of Heterozygosity Plus (LOH Plus) Module provides advanced tools for LOH/Copy Number data analysis.

*free upgrade for current customers

For a complete listing of BeadStudio Products, contact Customer Solutions at address below.

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