

BeadStudio Framework User Guide

A Modular Tool for Illumina Data Analysis

FOR RESEARCH ONLY

VERSION
3



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The BeadStudio Genotyping Module described herein is covered by U.S. Patent No. 7,035,740 and pending patent applications. The BeadStudio Methylation Module described herein is covered by pending patent applications.

Revision History

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Chapter 1

Overview

Topics

- 2 Introduction
- 2 Audience and Purpose
- 3 Installing BeadStudio
- 7 Starting BeadStudio
- 8 Verifying Your Version Numbers
- 9 Exiting BeadStudio

Introduction

BeadStudio is a modular software application that allows you to view and analyze genotyping and gene expression data.

BeadStudio includes the BeadStudio Framework and various add-on analysis software modules. The BeadStudio Framework includes a common user interface and tools that are available in all BeadStudio modules. Each BeadStudio module allows you to perform specific types of analyses.

To use the BeadStudio Framework, you must also install at least one licensed BeadStudio software module.

Audience and Purpose

This guide is written for researchers who want to use BeadStudio to view and analyze genotyping and gene expression data.

This guide provides procedures and user interface information for the BeadStudio Framework.

For information about the features specific to each BeadStudio analysis software module, refer to the user guide for that module:

- ▶ *BeadStudio Genotyping Module User Guide,*
Part # 11207066
- ▶ *BeadStudio Gene Expression Module User Guide,*
Part # 11207533
- ▶ *BeadStudio Methylation Module User Guide,*
Part # 11226064

Installing BeadStudio

To install the BeadStudio Framework:

1. Put the BeadStudio CD into your CD drive.
2. Do one of the following:
 - If the **Illumina BeadStudio Installation** screen appears (Figure 2), continue to Step 3.
 - If the CD does not load automatically, double-click the *BeadStudio<version>.exe* icon in the **BeadStudio** directory of the CD.

The BeadStudio application suite unzips (Figure 1).

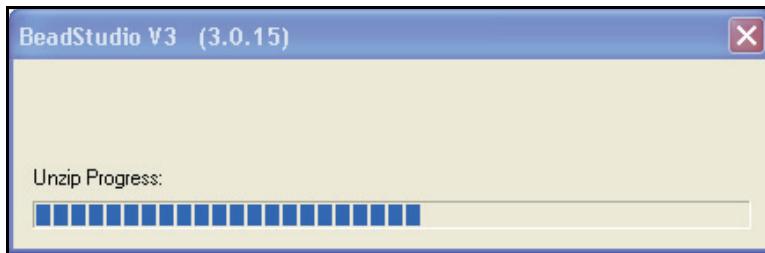


Figure 1 BeadStudio Application Unzipping

The **Illumina BeadStudio Installation** screen appears (Figure 2).

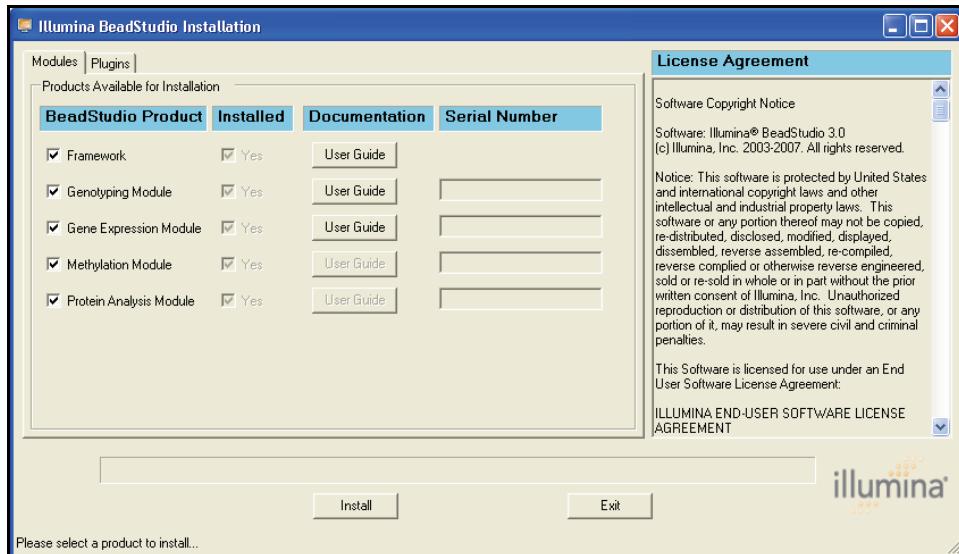


Figure 2 Illumina BeadStudio Installation, Modules Tab

3. Read the software license agreement in the right-hand side of the **Illumina BeadStudio Installation** window.
4. In the **BeadStudio Product** area, select **Framework**.



NOTE

The BeadStudio Framework works in conjunction with BeadStudio software modules. In addition to the Framework, select one or more BeadStudio modules to install. Have your serial number(s) available.

5. In the **Serial Number** area, enter your serial number(s) for the BeadStudio module(s) you want to install.



NOTE

Serial numbers are in the format #####-#####-#####-##### and can be found on an insert included with your BeadStudio CD.

6. **[Optional]** Enter the serial numbers for additional BeadStudio modules if you have licenses for additional BeadStudio modules and want to install them now.
7. **[Optional]** Install analysis algorithm plug-ins by performing the following steps:
 - a. Click the **Plug-ins** tab (Figure 3).

The **Plug-ins** tab lists the analysis algorithm plug-ins you can choose to install with BeadStudio.

b. Select the plug-ins you want to install.

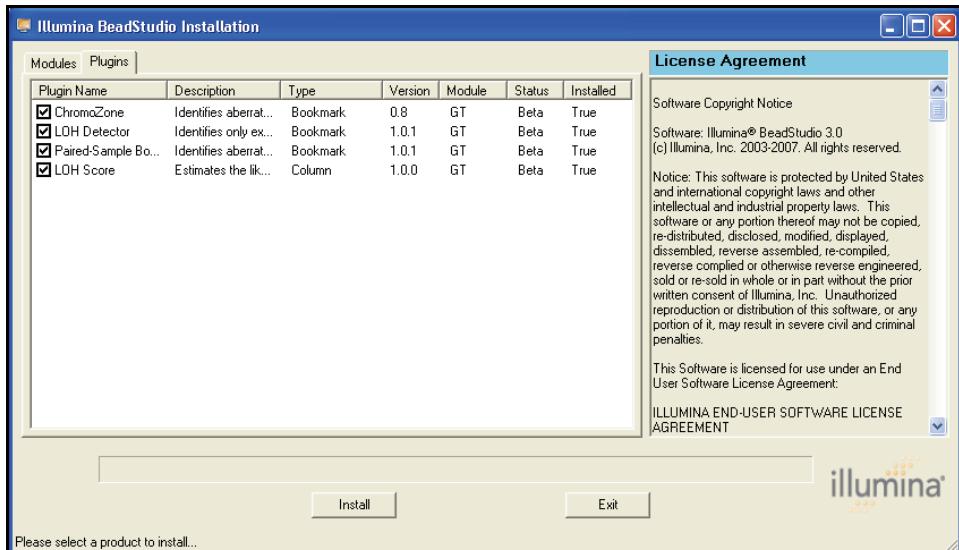


Figure 3 Illumina BeadStudio Installation, Plug-ins Tab

8. Click **Install**.

The **Software License Agreement** dialog box appears (Figure 4).



Figure 4 Accept License Agreement

9. Click **Yes** to accept the software license agreement.

The BeadStudio Framework is installed on your computer, along with any BeadStudio modules or plug-in algorithms you selected (Figure 5).

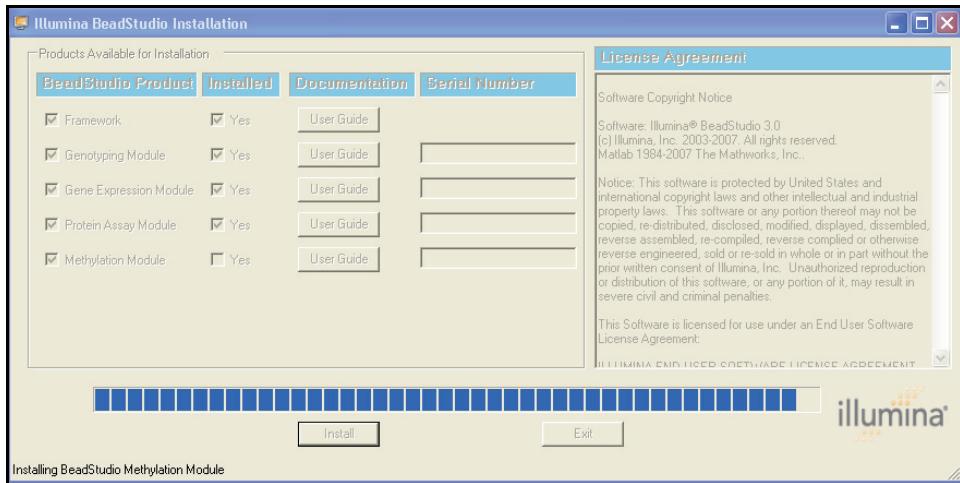


Figure 5 Illumina BeadStudio Installation

The **Installation Progress** dialog box notifies you that installation is complete (Figure 6).

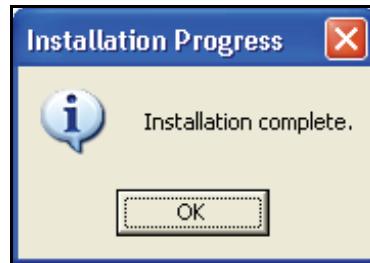


Figure 6 Installation Complete Dialog Box

10. Click **OK**.
 11. On the **Illumina BeadStudio Installation** screen (Figure 5), click **Exit**.
- You can now start a new BeadStudio project using any BeadStudio module you have installed with the Framework.

Starting BeadStudio

To start the BeadStudio Framework, do either of the following:

- ▶ Select **Start | Program Files | Illumina | BeadStudio.**

- ▶ Double-click the BeadStudio icon  on the desktop.

The BeadStudio application launches and opens to the main window (Figure 7).

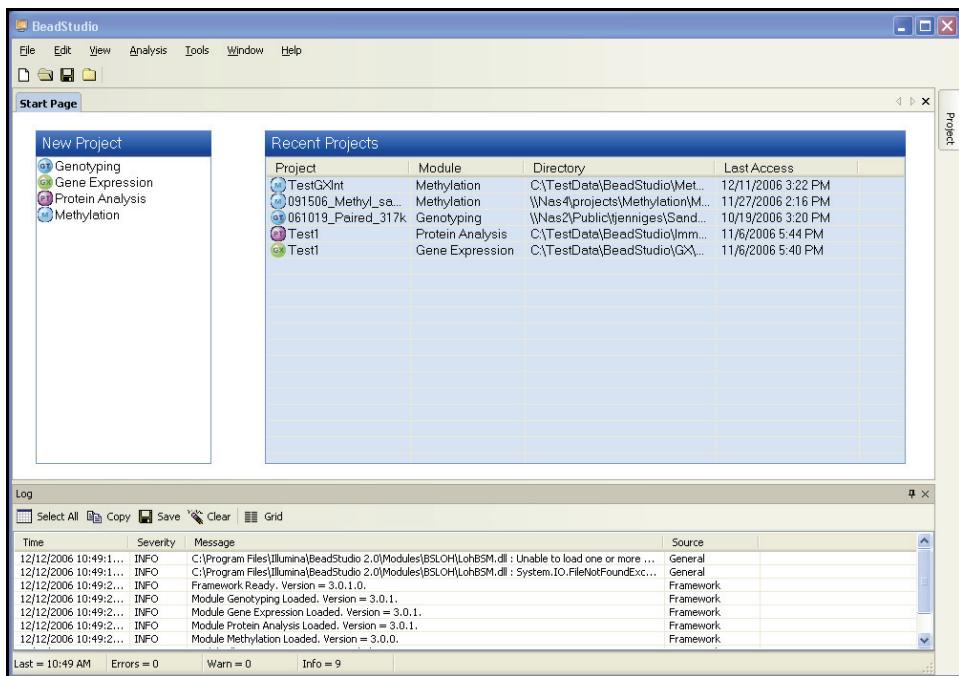


Figure 7 BeadStudio Main Window

Verifying Your Version Numbers

To verify the version number of the BeadStudio Framework you have installed, as well as the version numbers of any BeadStudio modules you have installed:

- Select **Help | About BeadStudio**.

The BeadStudio **About** box appears (Figure 8).

The version number of the BeadStudio **Framework** appears in the top portion of the BeadStudio **About** box.

The version numbers of any BeadStudio **modules** you have installed appear in the **Installed Components** area, in the middle portion of the BeadStudio **About** box.

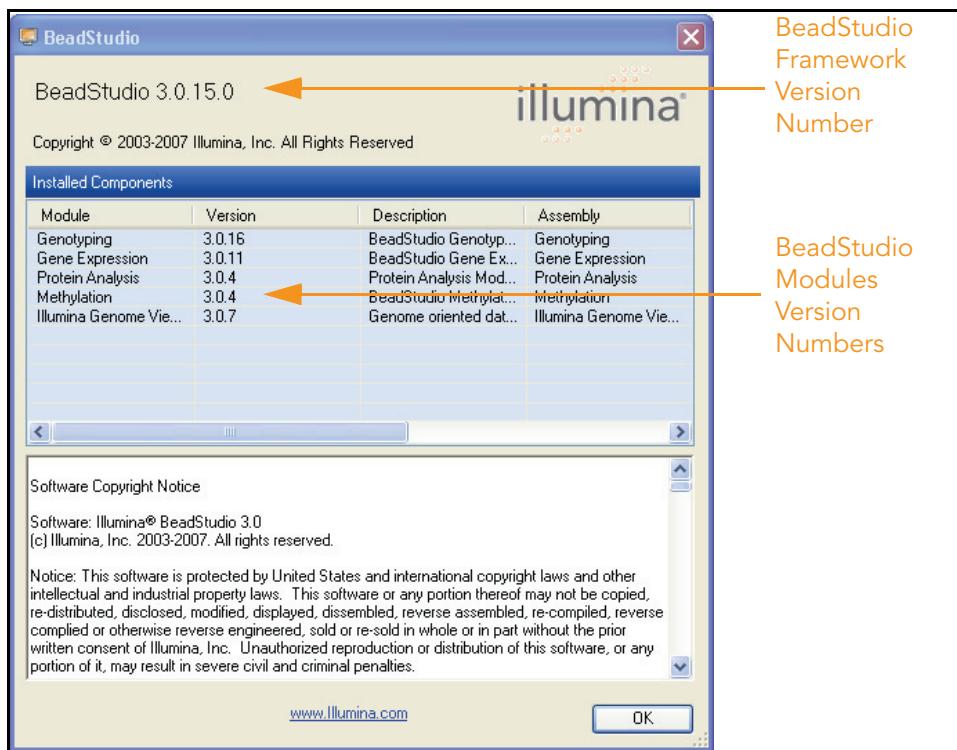


Figure 8 BeadStudio About Box

Exiting BeadStudio

To exit the BeadStudio application, do either of the following:

- ▶ Select **File | Exit**.
- ▶ Click  **Close** in the upper-right corner of the main window.
The BeadStudio application closes.

Chapter 2

Detachable Docking Windows

Topics

- 12 Introduction
- 12 Working with Detachable Docking Windows
 - 17 Saving Your Window Configuration
 - 17 Reverting to the Default Window Configuration

Introduction

The BeadStudio Framework is a flexible graphical user interface that allows you to organize and view your data in a number of ways. This chapter describes the configurable parts of the BeadStudio interface, known as detachable docking windows. All BeadStudio modules have detachable docking windows.

Working with Detachable Docking Windows

The BeadStudio Framework is flexible and customizable. When you launch BeadStudio, the interface appears in default view. However, you can reposition the detachable docking windows to suit your analysis needs and preferences.

Use the docking icons shown in Figure 9 through Figure 12 to reposition BeadStudio's detachable docking windows.

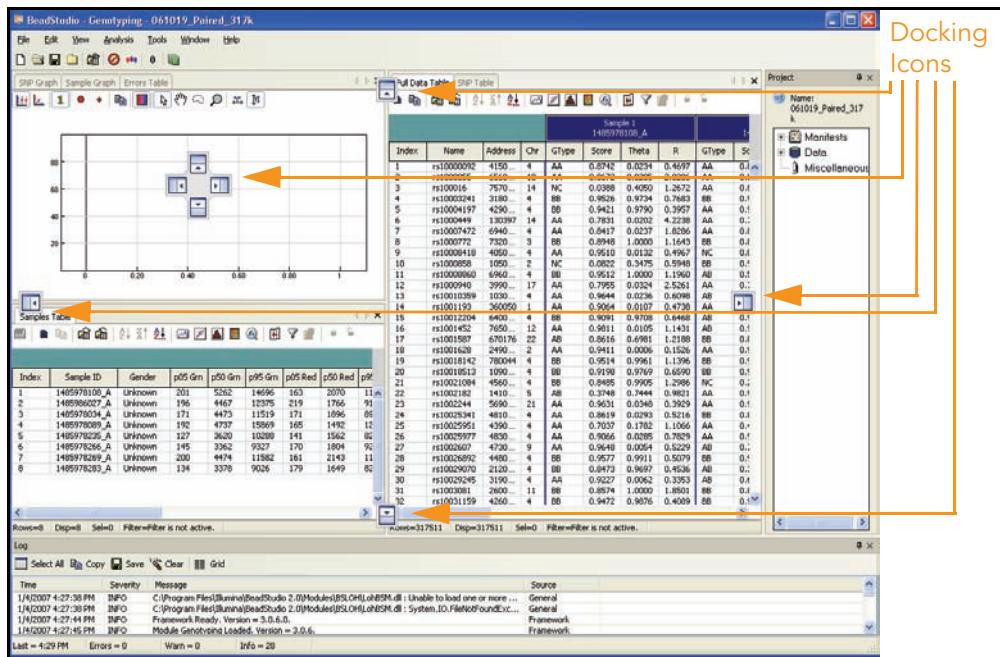


Figure 9 Docking Icons

Use the following techniques to detach and dock a window:

1. To select a detachable docking window, click its title tab.
2. Drag the detachable docking window over the main window.

BeadStudio displays icons (shown in Figure 9), which indicate potential docking positions and locations for the window.

When you drag a window over a docking icon, BeadStudio draws an outline or shades an area indicating where the window would be docked if you were to release the mouse button at that location.

BeadStudio's detachable docking windows allow you to customize the graphical user interface for your individual needs. The best way to learn how to configure the BeadStudio interface is to experiment, using the general guidelines that follow.

To dock a window on top of another window (so that it becomes a member of the tab group):

- ▶ Drag the mouse over the center docking icon in the destination window (Figure 10) and release the mouse button.

The window you moved is now docked on top of another window, and is a member of that window's tab group.

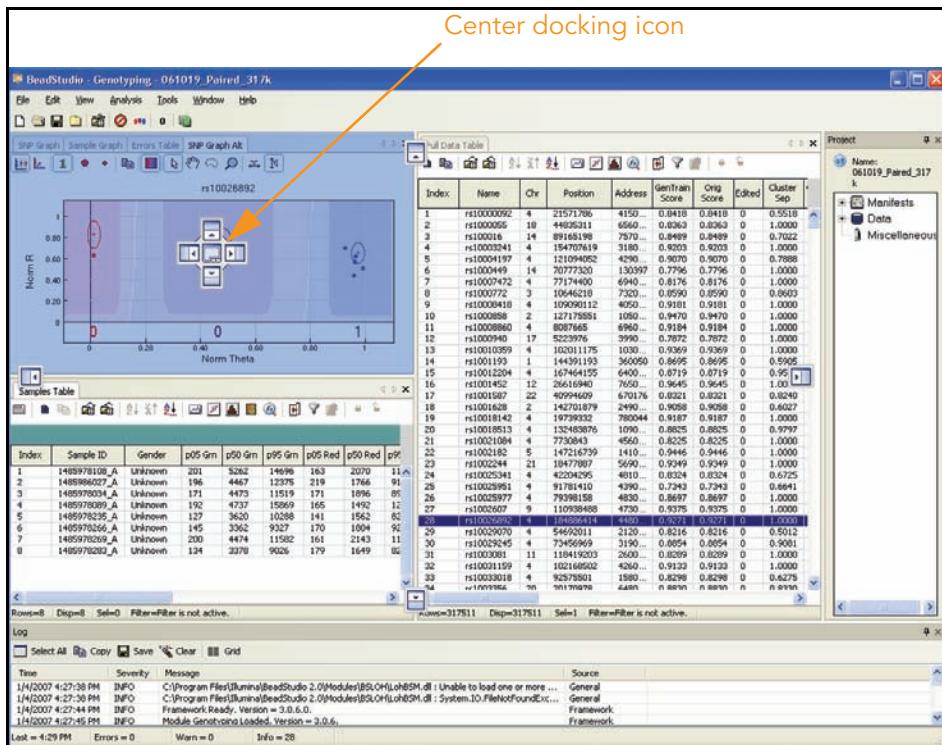


Figure 10 Docking a Window Over Another Window

To dock a window to the right of another window:

- ▶ Drag the mouse over the right docking icon , as shown in Figure 11, and release the mouse button.

The window you moved is now docked to the right of the window.

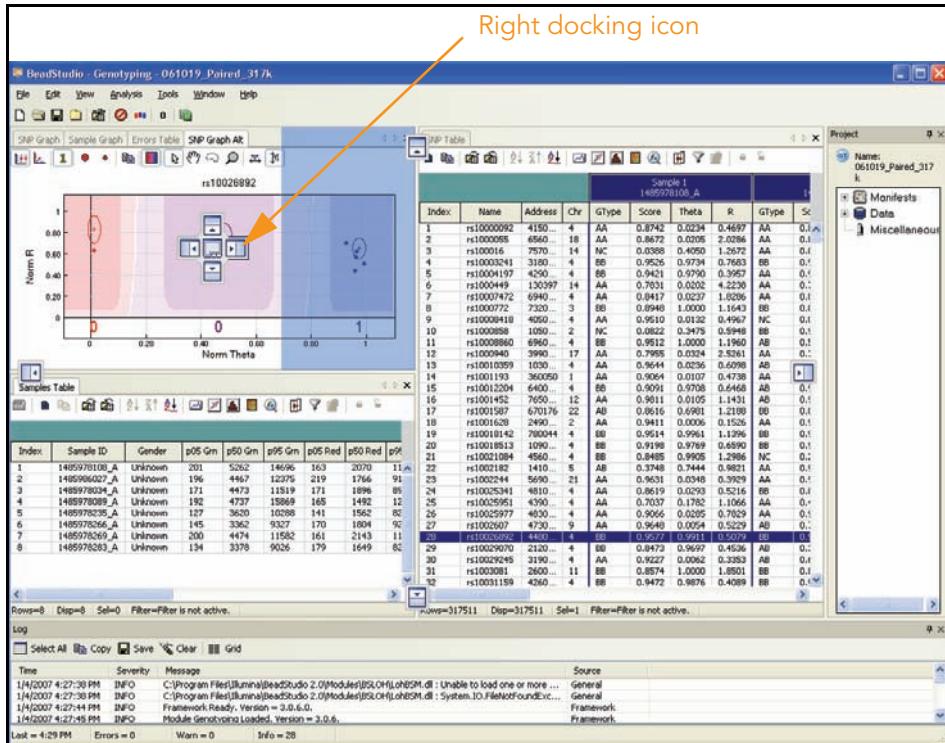


Figure 11 Docking a Window to the Right of Another Window

To dock a window to the left of another window:

- ▶ Drag the mouse over the left docking icon , as shown in Figure 11, and release the mouse button.

The window you moved is now docked to the left of another window.

To dock the window so it occupies an entire side of the main window:

- ▶ Drag the mouse over the docking icon at the edge of the main window, as shown in Figure 12, and release the mouse button.

The window you moved is now docked in an entire side of the main window.

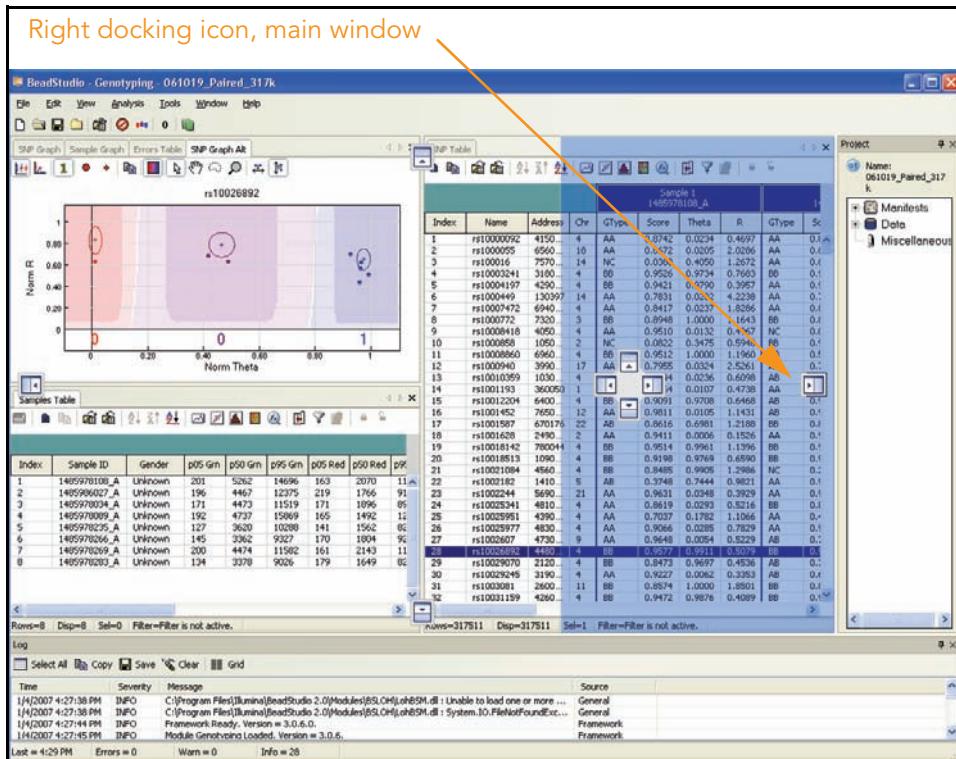


Figure 12 Docking a Window to Occupy Side of Main Window

Saving Your Window Configuration

When you quit BeadStudio, your window configuration is saved. The next time you start BeadStudio, your saved window configuration appears.

Reverting to the Default Window Configuration

To revert to the default window configuration at any time, select **View | Restore Default View** (Figure 13).

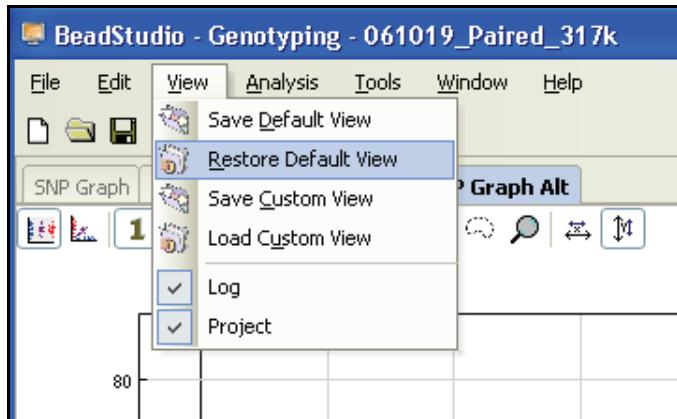


Figure 13 Restoring the Default View

Chapter 3

Tables

Topics

- 20 Introduction
- 20 Working with Tables
 - 21 Selecting Rows
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 - 39 Filtering Rows
- 46 Locking Columns
- 49 Unlocking Columns

Introduction

Tables behave the same way across all BeadStudio modules. This chapter describes how to work with tables in BeadStudio to manipulate, view, and analyze your data.

Working with Tables

There are two types of tables: those without subcolumns, and those with subcolumns. For example, in the BeadStudio Genotyping Module, the **SNP Table** (Figure 14) does not have subcolumns. The **Full Data Table** (Figure 15) does have subcolumns. It is important to note that tables with subcolumns have the same subcolumns for each column.

The screenshot shows a table titled "SNP Table" within the "Full Data Table" module. The table contains 45 rows of SNP data. Each row includes columns for Index (rsID), Name (e.g., rs10000992), Chr (chromosome number), Position (base position), Address (physical address), GenTrain Score, Orig Score, Edited status, Cluster Sep, ChiTest1, Ht Excess, AA Freq, AB Freq, and BB Freq. The "Edited" column shows a mix of checked and unchecked boxes. The "Cluster Sep" column values range from 0.0000 to 1.0000. The "ChiTest1" column values range from 0 to 3500. The "Ht Excess" column values range from -0.0000 to 1.0000. The "AA Freq", "AB Freq", and "BB Freq" columns show frequencies ranging from 0.0000 to 1.0000. Row 45 is labeled "v11m4XvNn" with an "A" suffix.

| Index | Name | Chr | Position | Address | GenTrain Score | Orig Score | Edited | Cluster Sep | ChiTest1 | Ht Excess | AA Freq | AB Freq | BB Freq |
|-------|------------|-----|-----------|----------|----------------|------------|--------|-------------|----------|-----------|---------|---------|---------|
| 1 | rs10000992 | 4 | 21571786 | 4150... | 0.8418 | 0.8418 | 0 | 0.5518 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.00 |
| 2 | rs10000959 | 18 | 44835311 | 6560... | 0.8363 | 0.8363 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 3 | rs100016 | 14 | 69165198 | 7570... | 0.8489 | 0.8489 | 0 | 0.7022 | 0.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0001 |
| 4 | rs10003241 | 4 | 154707619 | 3180... | 0.9203 | 0.9203 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 5 | rs10004197 | 4 | 121094052 | 4290... | 0.9070 | 0 | 0.7888 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 6 | rs1000449 | 14 | 70777320 | 130397 | 0.7796 | 0.7796 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 7 | rs1000772 | 3 | 10646210 | 7320... | 0.8589 | 0.8589 | 0 | 0.8603 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 9 | rs10008418 | 4 | 109090112 | 4050... | 0.9181 | 0.9181 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 10 | rs1000858 | 2 | 127175581 | 1050... | 0.9470 | 0.9470 | 0 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0001 |
| 11 | rs1000860 | 4 | 8087665 | 6960... | 0.9184 | 0.9184 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 12 | rs1000940 | 17 | 5223976 | 3990... | 0.7872 | 0.7872 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 13 | rs10010359 | 4 | 102011175 | 1030... | 0.9369 | 0.9369 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 14 | rs1001193 | 1 | 14439119 | 36005... | 0.8695 | 0.8695 | 0 | 0.5956 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 15 | rs10012204 | 4 | 167464158 | 6400... | 0.8719 | 0.8719 | 0 | 0.9548 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 16 | rs1001452 | 12 | 26616940 | 7650... | 0.9645 | 0.9645 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 17 | rs1001587 | 22 | 40994609 | 670176 | 0.8321 | 0.8321 | 0 | 0.8240 | 0.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0001 |
| 18 | rs1001628 | 2 | 142701879 | 2490... | 0.9058 | 0.9058 | 0 | 0.6027 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 19 | rs10016342 | 4 | 19739332 | 780194 | 0.9187 | 0.9187 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 20 | rs10016519 | 2 | 132480706 | 1090... | 0.8625 | 0.8625 | 0 | 0.9977 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 21 | rs1001694 | 4 | 7735493 | 4560... | 0.8235 | 0.8235 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 22 | rs100182 | 5 | 147216739 | 1410... | 0.9446 | 0.9446 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 23 | rs1002244 | 21 | 18477887 | 5690... | 0.9349 | 0.9349 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 24 | rs10025341 | 4 | 42204295 | 4810... | 0.8324 | 0.8324 | 0 | 0.6725 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 25 | rs10025951 | 4 | 91781410 | 4390... | 0.7343 | 0.7343 | 0 | 0.6641 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 26 | rs10025977 | 4 | 73996159 | 4830... | 0.8697 | 0.8697 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 27 | rs1002607 | 9 | 110938488 | 4730... | 0.9375 | 0.9375 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 28 | rs10026892 | 4 | 184886414 | 4480... | 0.9271 | 0.9271 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 29 | rs10029070 | 4 | 54692011 | 2120... | 0.8216 | 0.8216 | 0 | 0.5012 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 30 | rs10029245 | 4 | 73456969 | 3190... | 0.8854 | 0.8854 | 0 | 0.9081 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 31 | rs1003081 | 11 | 11841920 | 2600... | 0.8282 | 0.8289 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 32 | rs10031159 | 4 | 102168952 | 4280... | 0.8133 | 0.9133 | 0 | 1.0000 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 33 | rs10032636 | 4 | 92575905 | 1950... | 0.8205 | 0.8205 | 0 | 0.4959 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 34 | rs100356 | 20 | 20313973 | 4480... | 0.8830 | 0.8830 | 0 | 0.8330 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 35 | rs1003497 | 4 | 8675056 | 4670... | 0.8250 | 0.8250 | 0 | 0.9438 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 36 | rs1003563 | 12 | 6294838 | 4540... | 0.8153 | 0.8153 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 37 | rs10037067 | 5 | 90021638 | 3460... | 0.9608 | 0.9608 | 0 | 1.0000 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 38 | rs1003743 | 2 | 15517246 | 3450... | 0.8441 | 0.8441 | 0 | 0.7713 | 1.0000 | 0.0000 | 1.0000 | 0.0000 | 0.0001 |
| 39 | rs10039021 | 11 | 133032378 | 3850... | 0.7721 | 0.7721 | 0 | 0.5559 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 40 | rs1003921 | 11 | 17756912 | 6960... | 0.7776 | 0.7776 | 0 | 0.8677 | 0.0000 | 1.0000 | 0.0000 | 0.0000 | 1.00 |
| 41 | rs10041674 | 5 | 92659577 | 9404... | 0.8435 | 0.8435 | 0 | 0.6702 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 1.00 |
| 42 | rs10043779 | 5 | 93969574 | 4290... | 0.8347 | 0.8347 | 0 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0001 |
| 43 | rs10044860 | 5 | 149711264 | 6480... | 0.8607 | 0.8607 | 0 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0001 |
| 44 | rs1004616 | 9 | 23940994 | 4830... | 0.8261 | 0.8261 | 0 | 1.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0000 | 0.0001 |
| 45 | v11m4XvNn | A | 1AR11KHN | 4281... | n | n | n | n | n | n | n | n | n |

Figure 14 Table without Subcolumns

In Figure 15, the subcolumns for each sample column include **X Raw**, **YRaw**, **B Allele Freq**, and **Log R Ratio**.

The screenshot shows a software interface titled "Full Data Table" with a "SNP Table" tab selected. The table has two main sections: "Sample 1" and "Sample 2". Each section contains a header row with columns for Index, Name, Address, Chr, X Raw, Y Raw, B Allele Freq, and Log R Ratio. Below these headers are data rows. The first few rows of the table are as follows:

| Index | Name | Address | Chr | Sample 1 | | | | Sample 2 | | | | Sam | |
|-------|------------|-----------|-----|----------|-------|---------------|-------------|----------|-------|---------------|-------------|-------|-------|
| | | | | X Raw | Y Raw | B Allele Freq | Log R Ratio | X Raw | Y Raw | B Allele Freq | Log R Ratio | X Raw | Y Raw |
| 1 | rs10000092 | 4150... | 4 | 1915 | 259 | 0.0022 | -0.3685 | 2355 | 247 | 0.0000 | 0.2798 | 2056 | 204 |
| 2 | rs1000055 | 6560... | 18 | 8071 | 664 | 0.0061 | -0.1017 | 4748 | 464 | 0.0058 | -0.5129 | 357 | 9663 |
| 3 | rs100016 | 7570... | 14 | 3086 | 4205 | 0.3274 | 0.1787 | 2136 | 453 | 0.0126 | -0.5579 | 335 | 461 |
| 4 | rs10003241 | 3180... | 4 | 265 | 5673 | 0.984 | -0.2820 | 253 | 5845 | 0.999 | 0.0025 | 1950 | 3597 |
| 5 | rs1004197 | 4230... | 4 | 150 | 3600 | 0.9940 | -0.2380 | 1757 | 270 | 0.0000 | 0.1947 | 1200 | 314 |
| 6 | rs1004197 | 130397... | 14 | 16786 | 1200 | 0.0001 | -0.2441 | 1313 | 797 | 0.0000 | 0.3495 | 13532 | 1005 |

Below the table, status information is displayed: Rows=317511 Disp=317511 Sel=1 Filter=Filter is not active.

Figure 15 Table with Subcolumns

Selecting Rows

Select one or more table rows by doing one of the following:

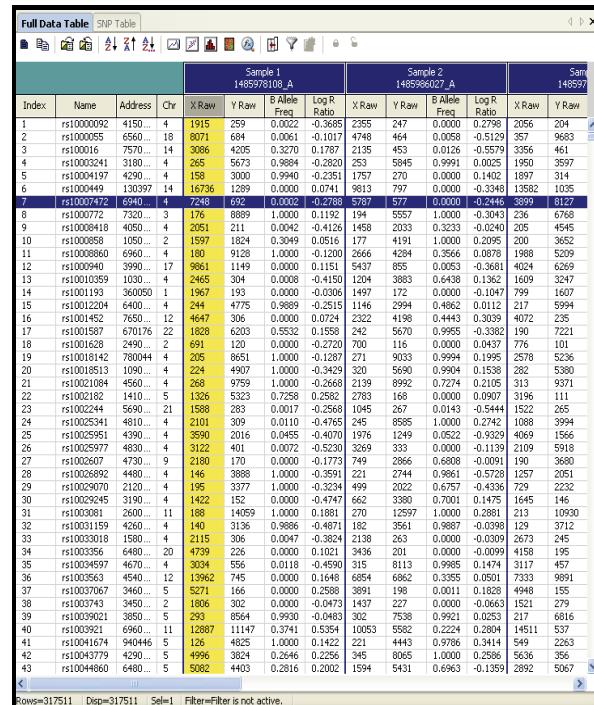
- ▶ Select a single table row by clicking in the row, using the up and down arrow keys on your keyboard, or scrolling with the mouse wheel.
- ▶ Select multiple contiguous table rows by pressing and holding the **Shift** key and clicking the mouse button on the first and last rows of the range you want to select.
- ▶ Select multiple noncontiguous table rows by pressing and holding the **Ctrl** key and clicking the mouse button once on each row you want to select.
- ▶ Select all rows by clicking **Select All Rows** in the toolbar. The selected rows are highlighted in dark blue.

Selecting Columns

To select a column:

- ▶ Place the cursor in a column header and click the mouse button.

The column is highlighted in yellow (Figure 16).



The screenshot shows a software window with a toolbar at the top. Below the toolbar is a menu bar with 'File', 'Edit', 'View', 'Tools', 'Help', and 'SNP Table'. The main area contains a table with three columns: 'Sample 1', 'Sample 2', and 'Sample 3'. Each column has several sub-headings: Index, Name, Address, Chr, X Raw, Y Raw, Allele Freq, Log R Ratio, and X Raw, Y Raw. The 'Sample 1' column header is highlighted in yellow. The table contains approximately 43 rows of SNP data. At the bottom left, there is a status bar with 'Rows=31751', 'Disp=31751', 'Sel=1', and 'Filter=Filter is not active.'

Figure 16 Table with a Column Selected

Showing & Hiding Columns

To show or hide columns:

1. In the **Table** toolbar, click  **Column Chooser**.

The **Column Chooser** appears (Figure 17).



Figure 17 Column Chooser

2. Select columns or subcolumns to show or hide by clicking a column name.
3. Click **Show** or **Hide**.

Alternatively, you can drag and drop the columns between the **Displayed Columns** area and the **Hidden Columns** area.

Searching Within Tables

To search within tables:

1. Place the cursor in a column header and click the mouse button.
The column is highlighted in yellow.
2. Right-click on the column header.
The context menu appears (Figure 18).

| | | | | Sample 1 1485978108_A | | | | | |
|-------|------------|---------|-----|--------------------------|-------|----------|-------------|-------|-------|
| Index | Name | Address | Chr | X Raw | Y Raw | B Allele | Log R ratio | X Raw | Y Raw |
| 1 | rs10000092 | 4150... | 4 | 1915 | | | | 3685 | 2355 |
| 2 | rs1000055 | 6560... | 18 | 8071 | | | | 1017 | 4748 |
| 3 | rs100016 | 7570... | 14 | 3086 | 4205 | 0.3270 | 0.1787 | 2135 | 453 |
| 4 | rs10003241 | 3180... | 4 | 265 | 5673 | 0.9884 | -0.2820 | 253 | 5845 |
| 5 | rs10004197 | 4290... | 4 | 158 | 3000 | 0.9940 | -0.2351 | 1757 | 270 |
| 6 | rs1000449 | 130397 | 14 | 16736 | 1289 | 0.0000 | 0.0741 | 9813 | 797 |
| 7 | rs10007472 | 6940... | 4 | 7248 | 692 | 0.0002 | -0.2788 | 5787 | 577 |
| 8 | rs1000772 | 7320... | 3 | 176 | 8889 | 1.0000 | 0.1192 | 194 | 5557 |
| 9 | rs10008418 | 4050... | 4 | 2051 | 211 | 0.0042 | -0.4126 | 1458 | 2033 |
| 10 | rs1000858 | 1050... | 2 | 1597 | 1824 | 0.3049 | 0.0516 | 177 | 4191 |
| 11 | rs10008860 | 6960... | 4 | 180 | 9128 | 1.0000 | -0.1200 | 2666 | 4284 |
| 12 | rs1000940 | 3990... | 17 | 9861 | 1149 | 0.0000 | 0.1151 | 5437 | 855 |
| 13 | rs10010359 | 1030... | 4 | 2465 | 304 | 0.0008 | -0.4150 | 1204 | 3883 |

Context menu

Figure 18 Tables Context Menu

3. Select **Find** from the context menu.

The **Find Row** dialog box appears (Figure 19).



Figure 19 Find Row Dialog Box

4. Enter a search term.

5. Click **Find Next**.

The table row that contains the next instance of your search term is highlighted in the table (Figure 20).

The screenshot shows a BeadStudio interface with a 'Full Data Table' window. A search dialog box titled 'Find Row...' is open, containing the text 'that starts with 732'. Below the dialog, two buttons are visible: 'Find Next' and 'Close'. The main table area displays two samples: Sample 1 (rs100016 to rs100049) and Sample 2 (rs100049 to rs1000940). The row for rs100049 (Index 8) is highlighted in blue, indicating it is the first instance of the search term '732'. An orange arrow points from the search term '732' in the dialog to the highlighted row in the table. Another orange arrow points from the 'Find Next' button in the dialog to the highlighted row in the table. The status bar at the bottom of the window shows 'Rows=317511 Disp=317511 Sel=1 Filter=Filter is not active.'

Figure 20 Highlighted Table Row Containing Search Term

6. **[Optional]** Click **Find Next** one or more additional times to find the next instance of the search term in the table.

Marking Table Rows

Mark table rows if you want to set them apart visually, for the purpose of viewing or analyzing a subset of your data.

To mark table rows:

1. Select the rows you want to mark by one of the following methods:
 - To select a single row:
 - Click on the row you want to mark.
 - To select multiple contiguous rows:
 - Click on the first row in the range you want to mark.
 - Press and hold the **Shift** key.
 - Click on the last row in the range you want to mark.
 - To select multiple non-contiguous rows:
 - Press and hold the **Ctrl** key.
 - Click on each row you want to mark.
2. Right-click once in the window to display the context menu.
3. Select **Configure Marks** (Figure 21).

| Sample 1 1485978108_A | | | | | | | | |
|--------------------------|------------|-------------------------|-----|-------|-------|---------------|-------------|---------|
| Index | Name | Address | Chr | X Raw | Y Raw | B Allele Freq | Log R Ratio | X Raw |
| 3 | rs100016 | 7570... | 14 | 3086 | 4205 | 0.3270 | 0.1787 | 2135 |
| 4 | rs10003241 | 3180... | 4 | 265 | 5673 | 0.9884 | -0.2820 | 253 |
| 5 | rs10004197 | 4290... | 4 | 158 | 3000 | 0.9940 | -0.2351 | 1757 |
| 6 | rs1000449 | 130397 | 14 | 16736 | 1289 | 0.0000 | 0.0741 | 9813 |
| 7 | rs10007472 | 6940... | 4 | 7248 | 692 | 0.0002 | -0.2788 | 5787 |
| 8 | rs1000772 | 7320... | 3 | 176 | 8889 | 1.0000 | 0.1192 | 194 |
| 9 | rs10008418 | 4050... | 4 | 2051 | 211 | 0.0042 | -0.4126 | 1458 |
| 10 | rs101 | Show Only Selected Rows | | | | 1824 | 0.3049 | 0.0516 |
| 11 | rs101 | Configure Marks... | | | | 9128 | 1.0000 | -0.1200 |
| 12 | rs101 | Configure Marks... | | | | 1149 | 0.0000 | 0.1151 |
| 13 | rs101 | Mark Selected Rows | | | | 304 | 0.0008 | -0.4150 |
| 14 | rs101 | Select Marked Rows | | | | 193 | 0.0000 | -0.0306 |
| 15 | rs101 | Select Marked Rows | | | | 4775 | 0.9889 | -0.2515 |
| 16 | rs101 | Clear Marks | | | | 306 | 0.0000 | 0.0724 |
| 17 | rs1001587 | 670176 | 22 | 1828 | 6203 | 0.5532 | 0.1558 | 242 |
| 18 | rs1001628 | 2490... | 2 | 691 | 120 | 0.0000 | -0.2720 | 700 |
| 19 | rs10018142 | 780044 | 4 | 205 | 8651 | 1.0000 | -0.1287 | 271 |

Figure 21 Configure Marks Selected

The **Configure Marks** dialog box appears (Figure 22).

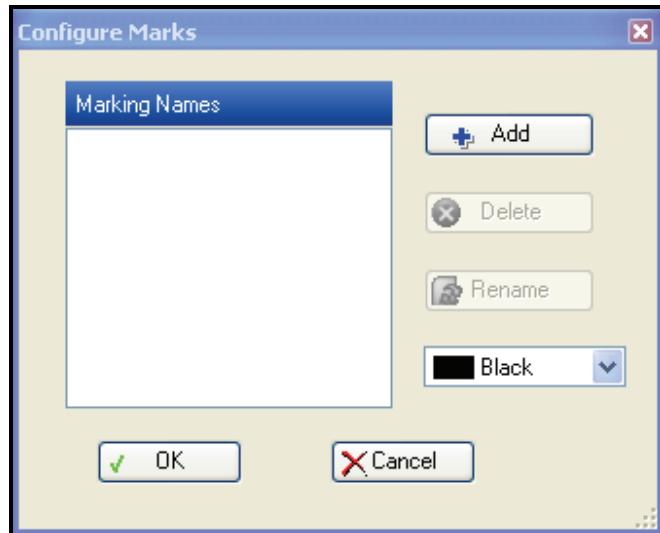


Figure 22 Configure Marks Dialog Box

4. Click **Add**.

The **Select Mark Name** dialog box appears (Figure 23).

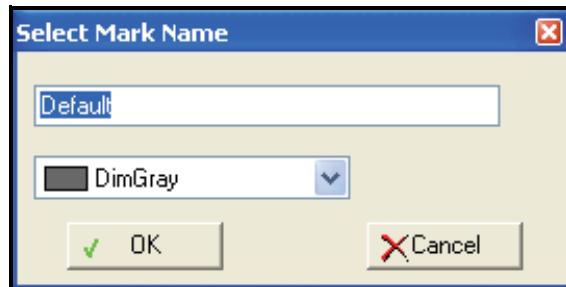


Figure 23 Select Mark Name Dialog Box

5. Type a label for your mark in the text field.
6. Choose a color for your mark from the color dropdown menu.

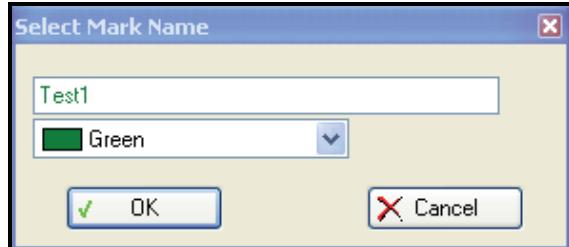


Figure 24 Selecting Mark Color

7. Click OK.

The mark is displayed in the color you chose (Figure 25).

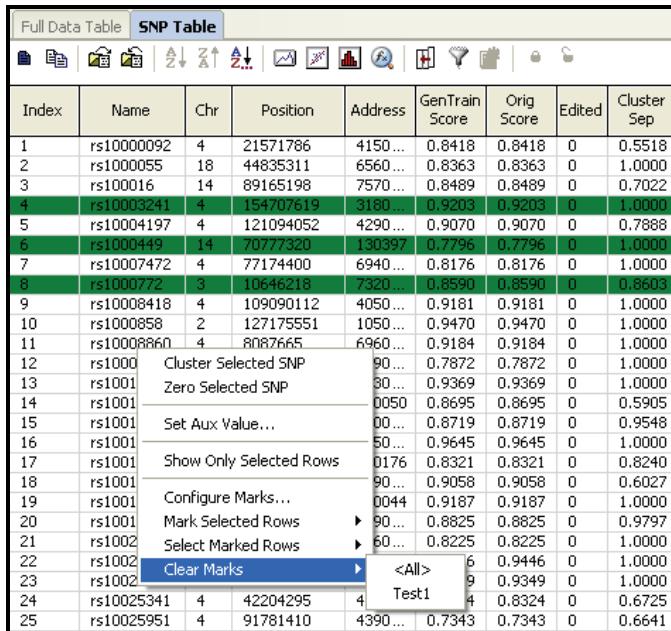
| Index | Name | Address | Chr | Sample 1 1465978108_A | | | | Sample 2 146596027_A | | | | Sample | 146597 |
|-------|------------|-----------|-----|-----------------------|-------|---------------|-------------|----------------------|-------|---------------|-------------|--------|--------|
| | | | | X Raw | Y Raw | B Allele Freq | Log R Ratio | X Raw | Y Raw | B Allele Freq | Log R Ratio | | |
| 1 | rs10000092 | 4150... | 4 | 1915 | 259 | 0.0022 | -0.3695 | 2355 | 247 | 0.0000 | -0.2798 | 2056 | 204 |
| 2 | rs1000055 | 6560... | 18 | 8071 | 684 | 0.0061 | -0.1017 | 4748 | 464 | 0.0058 | -0.5129 | 357 | 9683 |
| 3 | rs100016 | 7570... | 14 | 3086 | 4205 | 0.3270 | 0.1787 | 2135 | 453 | 0.0126 | -0.5579 | 3356 | 461 |
| 4 | rs10003241 | 3180... | 4 | 265 | 5973 | 0.9884 | -0.2860 | 253... | 5945 | 0.9991 | 0.0025 | 1950 | 3597 |
| 5 | rs10004197 | 4290... | 4 | 158 | 3000 | 0.9940 | -0.2351 | 1757 | 270 | 0.0000 | 0.1402 | 1897 | 314 |
| 6 | rs10004447 | 130397... | 14 | 16736 | 1289 | 0.0000 | 0.0741 | 9613 | 797 | 0.0000 | -0.3334 | 13852 | 1035 |
| 7 | rs10004742 | 6940... | 4 | 7248 | 692 | 0.0002 | -0.2788 | 5787 | 577 | 0.0000 | -0.4454 | 3899 | 8127 |
| 8 | rs10005110 | 4320... | 4 | 1776 | 1389 | 0.0000 | 0.0192 | 194... | 559 | 0.0000 | 0.0000 | 609 | 609 |
| 9 | rs1000618 | 4560... | 4 | 2051 | 241 | 0.0000 | -0.2454 | 1458 | 2033 | 0.0233 | -0.0240 | 205 | 4565 |
| 10 | rs1000659 | 1050... | 2 | 1597 | 234 | 0.3949 | 0.0516 | 1373 | 4191 | 1.0000 | 0.0926 | 200 | 3553 |
| 11 | rs10008660 | 6960... | 4 | 180 | 9128 | 1.0000 | -0.1200 | 2666 | 4284 | 0.3566 | 0.0878 | 998 | 5209 |
| 12 | rs1000940 | 2990... | 17 | 9861 | 1149 | 0.0000 | 0.1151 | 5437 | 855 | 0.0053 | -0.3651 | 4924 | 6269 |
| 13 | rs10010359 | 1030... | 4 | 2465 | 304 | 0.0008 | -0.4150 | 1204 | 3883 | 0.6438 | 0.1362 | 1609 | 3247 |
| 14 | rs1001193 | 360050... | 1 | 1967 | 193 | 0.0000 | -0.0306 | 1497 | 172 | 0.0000 | -0.1047 | 799 | 1607 |
| 15 | rs10012204 | 6400... | 4 | 244 | 4775 | 0.9889 | -0.2515 | 1146 | 2994 | 0.4862 | 0.0112 | 217 | 5994 |
| 16 | rs1001452 | 7650... | 12 | 4647 | 306 | 0.0000 | 0.0724 | 2322 | 4198 | 0.4443 | 0.3039 | 4072 | 235 |
| 17 | rs1001587 | 670176... | 22 | 1828 | 6203 | 0.5532 | 0.1558 | 242 | 5670 | 0.9955 | -0.3382 | 190 | 7221 |
| 18 | rs1001628 | 2490... | 2 | 691 | 120 | 0.0000 | -0.2720 | 700 | 116 | 0.0000 | 0.0437 | 776 | 101 |
| 19 | rs10018142 | 780044... | 4 | 205 | 8651 | 1.0000 | -0.1287 | 271 | 9033 | 0.9994 | 0.1995 | 2578 | 5236 |
| 20 | rs10018513 | 1090... | 4 | 224 | 4907 | 0.0000 | -0.3429 | 321 | 5690 | 0.9904 | 0.1533 | 282 | 5380 |
| 21 | rs1002084 | 4560... | 4 | 268 | 9759 | 1.0000 | -0.2658 | 2139 | 8992 | 0.7274 | 0.2105 | 311 | 9371 |
| 22 | rs1002156 | 151... | 1 | 1326 | 5232 | 0.0000 | 0.0071 | 2787 | 1484 | 0.0000 | -0.0027 | 3196 | 111 |
| 23 | rs1002244 | 5690... | 21 | 1588 | 233 | 0.0017 | -0.2568 | 1475 | 267 | 0.0143 | -0.5444 | 226 | |
| 24 | rs1002541 | 4810... | 4 | 2101 | 309 | 0.0110 | -0.4765 | 245 | 8585 | 1.0000 | 0.2742 | 1088 | 3994 |
| 25 | rs10025951 | 4390... | 4 | 3590 | 2016 | 0.0456 | -0.4070 | 1976 | 1249 | 0.0532 | -0.9329 | 4069 | 1566 |
| 26 | rs10025977 | 4830... | 4 | 3122 | 401 | 0.0072 | -0.5230 | 3269 | 333 | 0.0000 | -0.1139 | 2109 | 5918 |
| 27 | rs1002607 | 4730... | 9 | 2180 | 170 | 0.0000 | -0.1773 | 749 | 2866 | 0.6808 | 0.0991 | 190 | 3680 |
| 28 | rs10026892 | 4480... | 4 | 146 | 3888 | 1.0000 | -0.3591 | 221 | 2744 | 0.9961 | -0.5728 | 1257 | 2051 |
| 29 | rs1002907 | 2120... | 4 | 195 | 3377 | 1.0000 | -0.3234 | 199 | 2023 | 0.6757 | -0.4333 | 729 | 2232 |
| 30 | rs10029245 | 3190... | 4 | 1422 | 152 | 0.0000 | -0.4747 | 662 | 3380 | 0.7001 | 0.1475 | 1645 | 146 |
| 31 | rs1003081 | 2600... | 11 | 180 | 14059 | 1.0000 | 0.1881 | 270 | 12597 | 1.0000 | 0.2861 | 213 | 10930 |
| 32 | rs10031159 | 4260... | 4 | 141 | 3136 | 0.9868 | -0.4871 | 185 | 3561 | 0.9887 | -0.0398 | 129 | 3712 |
| 33 | rs10033011 | 1580... | 4 | 2115 | 306 | 0.0047 | -0.3824 | 2138 | 263 | 0.0000 | -0.0309 | 2673 | 245 |
| 34 | rs1003356 | 6480... | 20 | 4739 | 226 | 0.0000 | 0.1021 | 3436 | 201 | 0.0000 | -0.0199 | 4158 | 195 |
| 35 | rs10033771 | 4290... | 4 | 1420 | 4795 | 0.0000 | -0.4090 | 2409 | 8119 | 0.6288 | 0.1144 | 1403 | 1167 |
| 36 | rs1003563 | 4510... | 12 | 15962 | 45 | 0.0000 | 0.1646 | 6554 | 6852 | 0.3555 | -0.0501 | 7333 | 9891 |
| 37 | rs10037067 | 2460... | 5 | 5271 | 166 | 0.0000 | 0.2588 | 3891 | 198 | 0.0111 | 0.1828 | 4949 | 155 |
| 38 | rs1003743 | 3450... | 2 | 1806 | 302 | 0.0000 | -0.0473 | 1437 | 227 | 0.0000 | -0.0663 | 1521 | 279 |
| 39 | rs10039021 | 3850... | 5 | 293 | 8564 | 0.9930 | -0.0493 | 302 | 7538 | 0.9921 | 0.0253 | 217 | 6816 |
| 40 | rs1003921 | 6960... | 11 | 12887 | 11147 | 0.3741 | 0.0534 | 10053 | 5582 | 0.2224 | 0.2804 | 14511 | 537 |
| 41 | rs10041674 | 940446 | 5 | 126 | 4825 | 1.0000 | 0.1422 | 221 | 4443 | 0.9786 | 0.3414 | 549 | 2263 |
| 42 | rs10043779 | 4290... | 5 | 4996 | 3624 | 0.2646 | 0.2256 | 345 | 8065 | 1.0000 | 0.2586 | 5636 | 356 |
| 43 | rs10044960 | 6480... | 5 | 5082 | 4403 | 0.2816 | 0.2002 | 1594 | 5431 | 0.6963 | -0.1359 | 2892 | 5067 |

Figure 25 Selected Rows Marked in the Table

Clearing Marks

To clear any marks in a table:

1. Select **Clear Marks** from the context menu (Figure 26).



The screenshot shows a BeadStudio interface with a table titled "SNP Table". The table has columns: Index, Name, Chr, Position, Address, GenTrain Score, Orig Score, Edited, and Cluster Sep. Rows are numbered 1 to 25. Rows 4, 6, 8, 10, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21, 22, 23, 24, and 25 have green backgrounds, indicating they are selected. A context menu is open over row 22, with the "Clear Marks" option highlighted in blue. Other options in the menu include "<All>" and "Test1".

Figure 26 Clear Selected Marks from the Table

2. Choose whether you want to clear all marks, or clear only a specific mark.

The selected marks are removed and the table rows return to BeadStudio default colors.

Exporting to a File

The **Export Displayed Data to File** function allows you to export currently-displayed table data to a tab-delimited file.

To export a column:

1. Click  **Export Displayed Data to File**.
The **Save As** dialog box appears.
2. In the **File Name** text field, enter a name for the file.
3. Click **Save**.
If there are rows selected in the table, the following prompt appears:



Figure 27 Exporting Data Dialog Box

4. Do one of the following:
 - To export the entire visible table, click **Yes**.
 - To export only the selected rows and columns, click **No**.
The file is saved in the location you specified.

Importing a Column

The **Import Columns** function allows you to import data into BeadStudio from a preexisting file.



NOTE

In BeadStudio v3, you can import subcolumns in addition to columns.

To import a column or a subcolumn:

1. Click  **Import Columns**.
The **Import** dialog box appears (Figure 28).

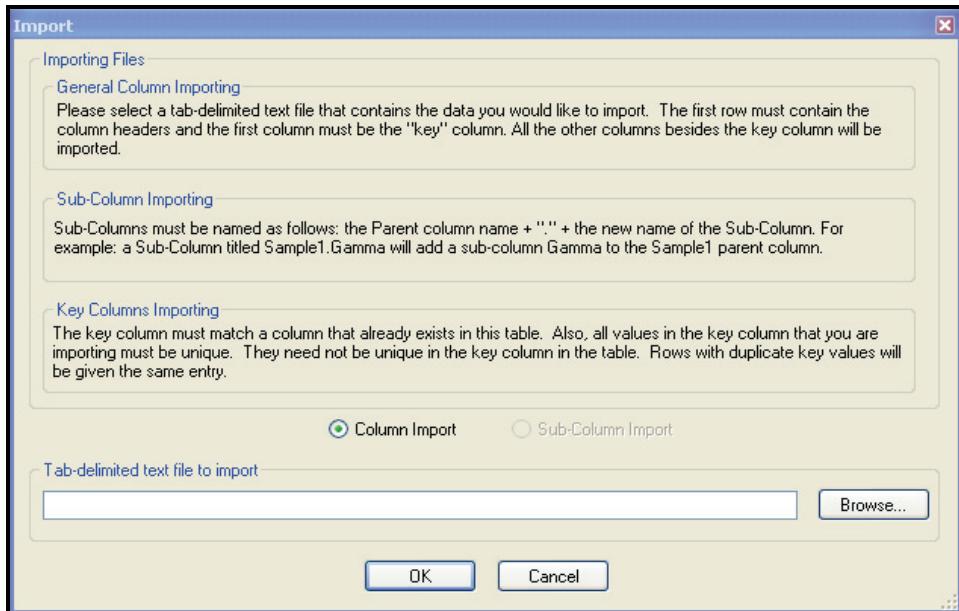


Figure 28 Import Dialog Box

2. Use the following guidelines, shown in the **Import** dialog box, to import columns or subcolumns:

Table 1 Guidelines for Importing Columns or Subcolumns

| To import a... | Do the following... |
|----------------|--|
| General column | Select a tab-delimited file that you would like to import. The first row must contain the column headers and the first column must be a "key" column. All other columns besides the key column will be imported. |
| Subcolumn | Name the subcolumns you want to import as follows: the parent column name, followed by a period, followed by the name of the subcolumn. For example, a subcolumn titled "Sample1.Gamma" adds a subcolumn "Gamma" to the "Sample1" parent column. |
| Key column | A "key" column must match a column that already exists in this table. Also, all values in the key column that you are importing must be unique. They need not be unique in the key column in the table. Rows with duplicate key values will be given the same entry. |

3. Select **Column Import** or **Subcolumn Import**.

4. Click **Browse**.

5. Browse to the tab-delimited file you want to import.

6. Click **Open**.

7. Click **OK**.

The imported column or subcolumn appear in the table.



NOTE

You can perform the same actions on an imported column that you can perform on a standard column.

Sorting by Column

To sort a column in ascending order:

- ▶ Select the column and click **Sort Column (Ascending)**.

To sort a column in descending order:

- ▶ Select the column and click **Sort Column (Descending)**.

To sort by more than one column:

1. Click **Sort by Column(s)** to bring up the **Sort** dialog box (Figure 29).

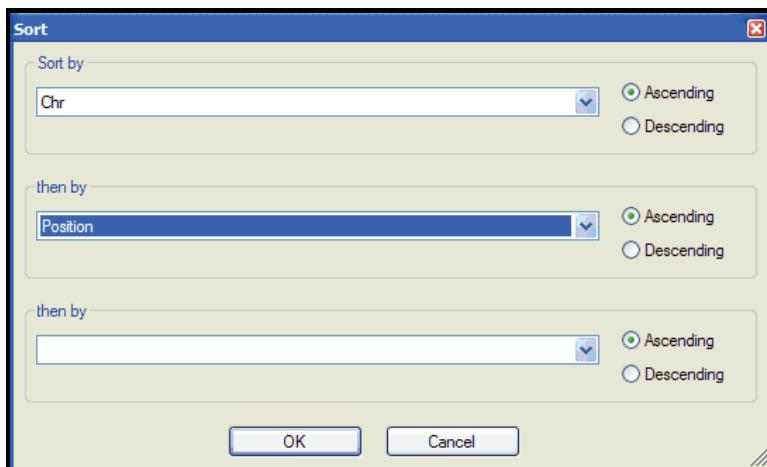


Figure 29 Multiple-Column Sort Dialog Box

2. Select the columns you want to sort by, and choose ascending or descending sort order for each one.

3. Click **OK**.

The columns are sorted as you specified.

Creating a New Column

The BeadStudio Framework allows you to create new columns and add them to a table.

In BeadStudio v3, you can create columns that are functions of other columns, or you can create annotation columns.

To create a new column:

1. Click  **New Column**.

The **New Column** dialog box appears (Figure 30).

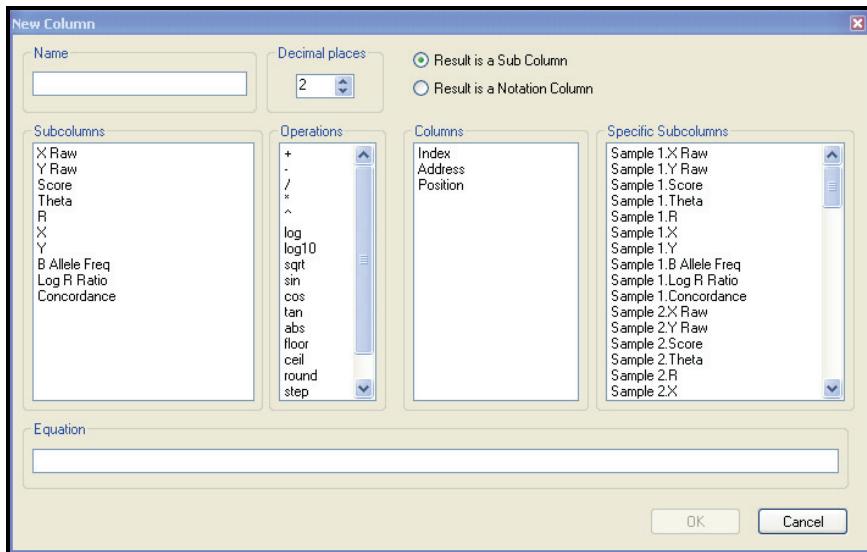


Figure 30 New Column Dialog Box

2. In the **Name** text field, type a name for the new column you want to create.
3. In the **Decimal Places** listbox, select or type the number of decimal places you want to display in the new column.
4. Choose one of the following options:
 - Select **Result is a Subcolumn** if you want to create a new subcolumn for every parent column.
 - Select **Result is a Notation Column** if you want to create a new, independent column.
5. Build the column's equation by clicking the columns and operations you want to include (Table 2).

Table 2 New Column Operations & Descriptions

| Operation | Description |
|-----------|--|
| + | Addition |
| - | Subtraction |
| / | Division |
| * | Multiplication |
| ^ | Power |
| log | Log |
| log10 | Log base 10 |
| abs | Absolute value |
| floor | Greatest integer that is less than or equal to x |
| ceil | Smallest integer that is greater than or equal to z |
| round | Closest integer to x |
| step | 0 if x is less than 0 1 if x is greater than or equal to zero |
| mod | Remainder |
| sgn | Sign |

The equation appears in the **Equation** text field (Figure 31).

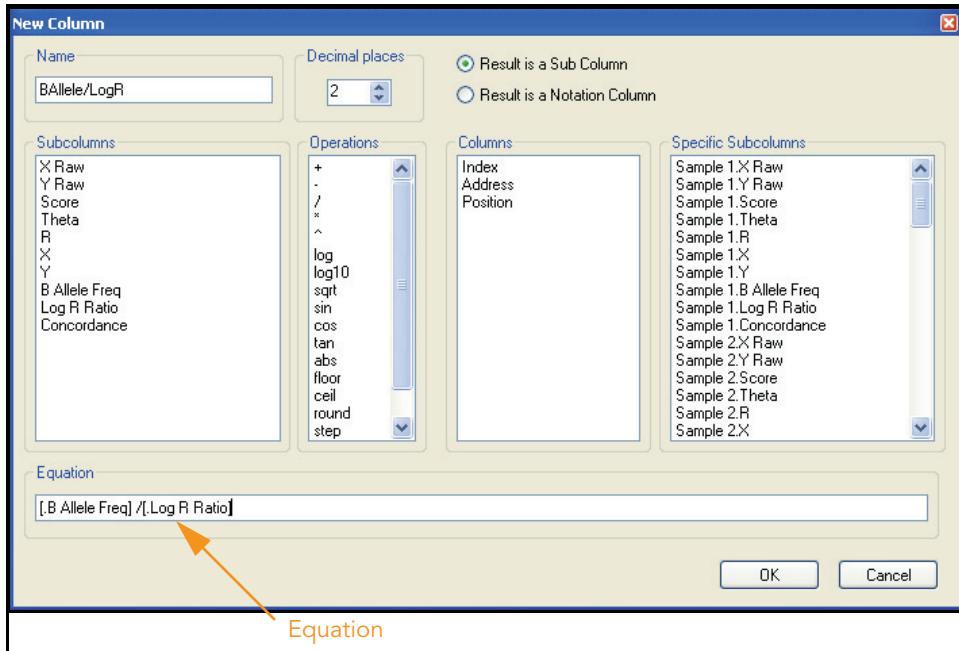


Figure 31 Building an Equation

6. Click OK.

The new column is added to the table for every sample (Figure 32).

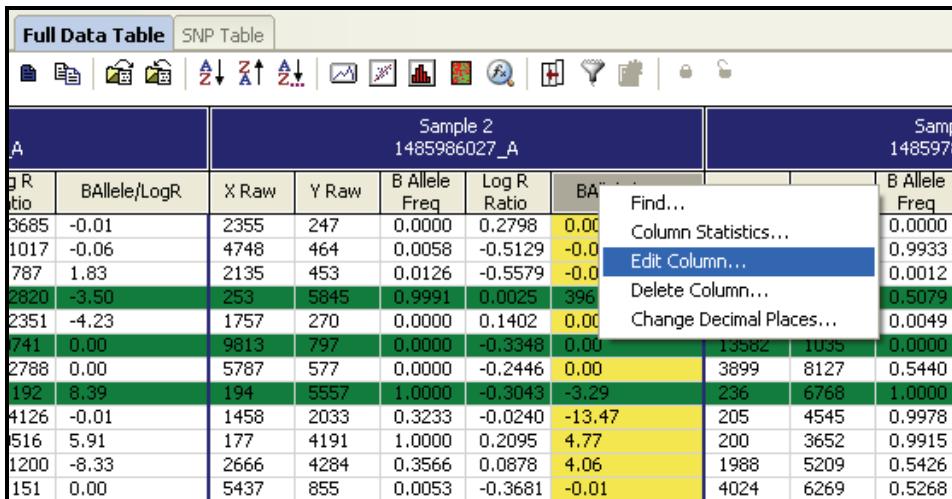
| A | | Sample 2 1485986027_A | | | | | Sample 3 1485978034_A | | | | |
|-------------|---------------|--------------------------|-------|------------------|----------------|---------------|--------------------------|-------|------------------|----------------|---------------|
| g R atio | BAAllele/LogR | X Raw | Y Raw | B Allele Freq | Log R Ratio | BAAllele/LogR | X Raw | Y Raw | B Allele Freq | Log R Ratio | BAAllele/LogR |
| 3685 | -0.01 | 2355 | 247 | 0.0000 | 0.2794 | 0.00 | 2056 | 204 | 0.0000 | -0.0871 | 0.00 |
| 1017 | -0.06 | 4748 | 464 | 0.0058 | -0.5129 | -0.01 | 357 | 9683 | 0.9933 | -0.0219 | -45.38 |
| 787 | 1.83 | 2135 | 453 | 0.0126 | -0.5579 | -0.02 | 3356 | 461 | 0.0012 | -0.0724 | -0.02 |
| 2620 | -3.50 | 253 | 5845 | 0.9991 | 0.0025 | 396.63 | 1950 | 3597 | 0.5079 | 0.0432 | 11.76 |
| 2351 | -4.23 | 1757 | 270 | 0.0000 | 0.1402 | 0.00 | 1897 | 314 | 0.0049 | 0.1254 | 0.04 |
| 741 | 0.00 | 9813 | 797 | 0.0000 | 0.2318 | 0.00 | 13692 | 1035 | 0.0000 | 0.0393 | 0.00 |
| 2788 | 0.00 | 5787 | 577 | 0.0000 | -0.2446 | 0.00 | 3899 | 8127 | 0.5440 | 0.0590 | 9.21 |
| 192 | 6.59 | 194 | 5557 | 1.0000 | 0.5043 | 3.29 | 236 | 5768 | 1.0000 | 0.0162 | 54.90 |
| 4126 | -0.01 | 1458 | 2033 | 0.3233 | -0.0240 | -13.47 | 205 | 4545 | 0.9978 | -0.0588 | -16.96 |
| 516 | 5.91 | 177 | 4191 | 1.0000 | 0.2095 | 4.77 | 200 | 3652 | 0.9915 | 0.0686 | 14.40 |
| 1200 | -8.33 | 2666 | 4284 | 0.3566 | 0.0878 | 4.06 | 1988 | 5209 | 0.5426 | -0.0452 | -12.00 |
| 151 | 0.00 | 5437 | 855 | 0.0053 | -0.3681 | -0.01 | 4024 | 6269 | 0.5268 | -0.0367 | -14.37 |
| 4150 | 0.00 | 1204 | 3883 | 0.6438 | 0.1362 | 4.73 | 1609 | 3247 | 0.5124 | 0.0885 | 5.79 |
| 0306 | 0.00 | 1497 | 172 | 0.0000 | -0.1047 | 0.00 | 799 | 1607 | 0.5258 | -0.2242 | -2.34 |
| 2515 | -3.93 | 1146 | 2994 | 0.4862 | 0.0112 | 43.24 | 217 | 5994 | 1.0000 | 0.3265 | 3.06 |
| 724 | 0.00 | 2322 | 4198 | 0.4443 | 0.3039 | 1.46 | 4072 | 235 | 0.0000 | 0.0612 | 0.00 |
| 558 | 3.55 | 242 | 5670 | 0.9955 | -0.3382 | -2.94 | 190 | 7221 | 1.0000 | 0.0084 | 119.39 |
| 2720 | 0.00 | 700 | 116 | 0.0000 | 0.0437 | 0.00 | 776 | 101 | 0.0000 | 0.0976 | 0.00 |
| 1287 | -7.77 | 271 | 9033 | 0.9994 | 0.1995 | 5.01 | 2578 | 5236 | 0.4976 | 0.0764 | 6.51 |

Figure 32 New Column Displayed for Every Sample

Editing a User-Defined Subcolumn

To edit a user-defined subcolumn:

1. Right-click in the heading of the subcolumn you want to edit.
2. Select **Edit Column** (Figure 33) from the context menu.



| A | | Sample 2 1485986027_A | | | | | Samp | |
|-------------|--------------|--------------------------|-------|---------------|-------------|---------|--------------------------|---------------|
| Log R Ratio | BAllele/LogR | X Raw | Y Raw | B Allele Freq | Log R Ratio | BAllele | Find... | B Allele Freq |
| 3685 | -0.01 | 2355 | 247 | 0.0000 | 0.2798 | 0.00 | Column Statistics... | 0.0000 |
| 1017 | -0.06 | 4748 | 464 | 0.0058 | -0.5129 | -0.0 | Edit Column... | 0.9933 |
| 787 | 1.83 | 2135 | 453 | 0.0126 | -0.5579 | -0.0 | Delete Column... | 0.0012 |
| 2820 | -3.50 | 253 | 5945 | 0.9991 | 0.0025 | 396 | Change Decimal Places... | 0.5079 |
| 2351 | -4.23 | 1757 | 270 | 0.0000 | 0.1402 | 0.00 | | 0.0049 |
| 741 | 0.00 | 9813 | 797 | 0.0000 | -0.3348 | 0.00 | | 0.0000 |
| 2788 | 0.00 | 5787 | 577 | 0.0000 | -0.2446 | 0.00 | | 0.5440 |
| 192 | 8.39 | 194 | 5557 | 1.0000 | -0.3043 | -3.29 | | 1.0000 |
| 4126 | -0.01 | 1458 | 2033 | 0.3233 | -0.0240 | -13.47 | | 0.9978 |
| 516 | 5.91 | 177 | 4191 | 1.0000 | 0.2095 | 4.77 | | 0.9915 |
| 1200 | -8.33 | 2666 | 4284 | 0.3566 | 0.0878 | 4.06 | | 0.5426 |
| 151 | 0.00 | 5437 | 855 | 0.0053 | -0.3681 | -0.01 | | 0.5268 |

Figure 33 Editing a User-Defined Subcolumn

The **Edit Column** screen appears (Figure 34).

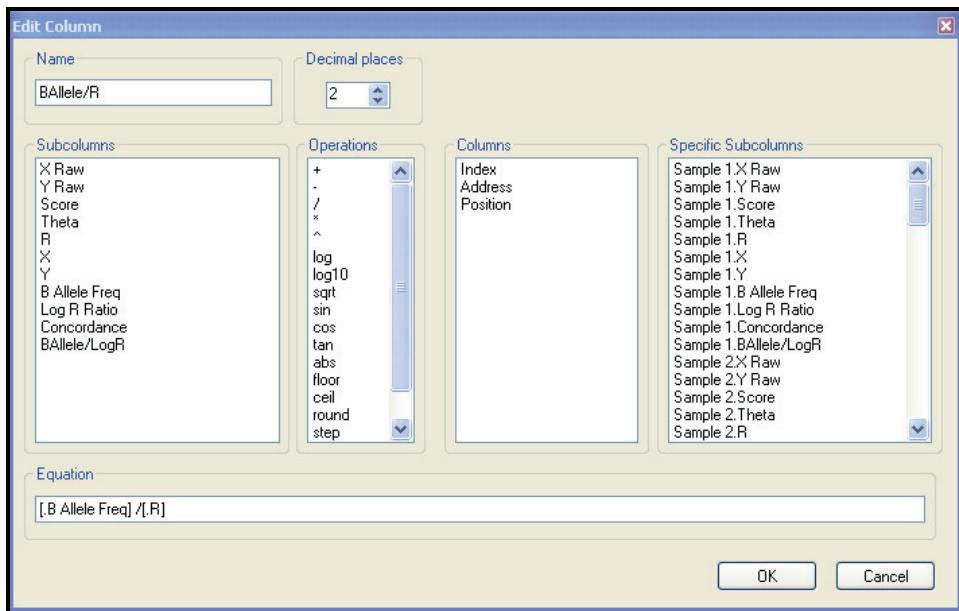
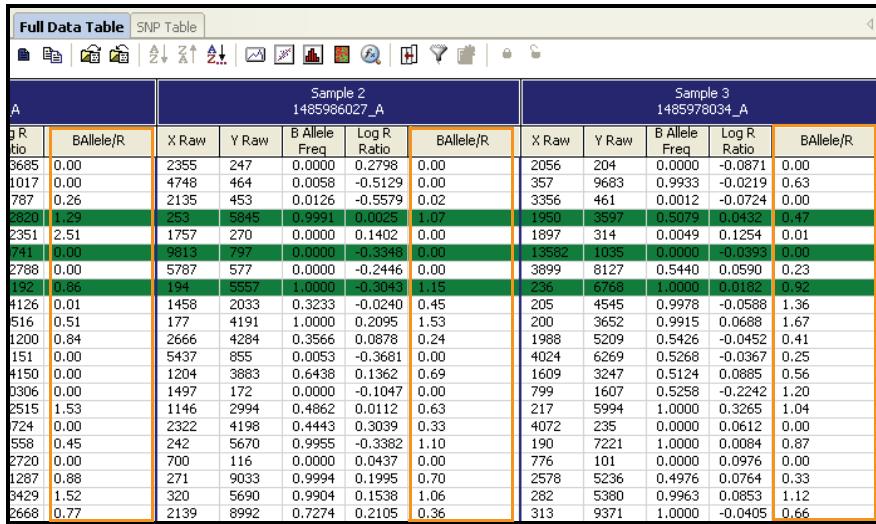


Figure 34 Changing Subcolumn Attributes

3. In the **Edit Column** screen, edit the subcolumn by changing the subcolumn attributes.
4. Click **OK**.

The changes you made to the subcolumn are reflected in the table.



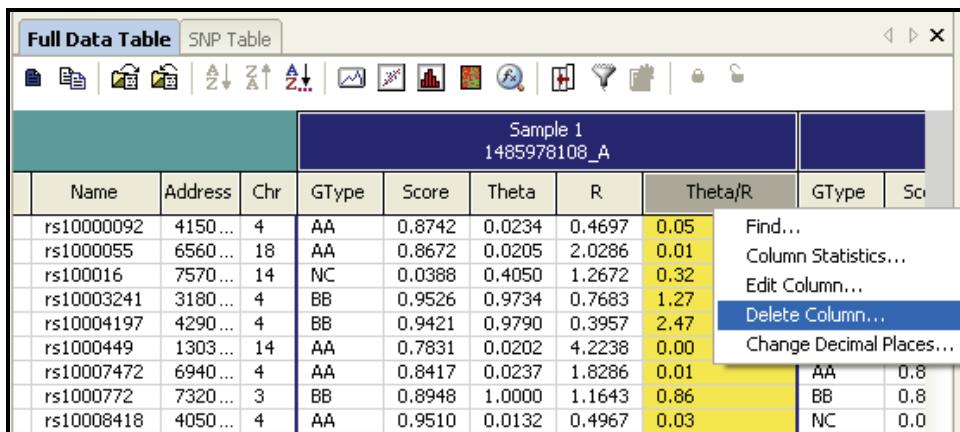
| A | | Sample 2 1485986027_A | | | | Sample 3 1485978034_A | | | | | |
|-------------|-----------|--------------------------|-------|------------------|----------------|--------------------------|-------|-------|------------------|----------------|-----------|
| g R atio | BAllele/R | X Raw | Y Raw | B Allele Freq | Log R Ratio | BAllele/R | X Raw | Y Raw | B Allele Freq | Log R Ratio | BAllele/R |
| 3685 | 0.00 | 2355 | 247 | 0.0000 | 0.2798 | 0.00 | 2056 | 204 | 0.0000 | -0.0871 | 0.00 |
| 1017 | 0.00 | 4748 | 464 | 0.0058 | -0.5129 | 0.00 | 357 | 9683 | 0.9933 | -0.0219 | 0.63 |
| 787 | 0.26 | 2135 | 453 | 0.0126 | -0.5579 | 0.02 | 3356 | 461 | 0.0012 | -0.0724 | 0.00 |
| 2820 | 1.29 | 253 | 5845 | 0.9991 | 0.0025 | 1.07 | 1950 | 3597 | 0.5079 | 0.0432 | 0.47 |
| 2351 | 2.51 | 1757 | 270 | 0.0000 | 0.1402 | 0.00 | 1897 | 314 | 0.0049 | 0.1254 | 0.01 |
| 741 | 0.00 | 9813 | 797 | 0.0000 | -0.3348 | 0.00 | 13582 | 1035 | 0.0000 | -0.0393 | 0.00 |
| 2788 | 0.00 | 5787 | 577 | 0.0000 | -0.2446 | 0.00 | 3899 | 8127 | 0.5440 | 0.0590 | 0.23 |
| 192 | 0.86 | 194 | 5557 | 1.0000 | -0.3043 | 1.15 | 236 | 6764 | 1.0000 | 0.0182 | 0.92 |
| 4126 | 0.01 | 1458 | 2033 | 0.3233 | -0.0240 | 0.45 | 205 | 4545 | 0.9978 | -0.0588 | 1.36 |
| 516 | 0.51 | 177 | 4191 | 1.0000 | 0.2095 | 1.53 | 200 | 3652 | 0.9915 | 0.0688 | 1.67 |
| 1200 | 0.84 | 2666 | 4284 | 0.3566 | 0.0878 | 0.24 | 1988 | 5209 | 0.5426 | -0.0452 | 0.41 |
| 151 | 0.00 | 5437 | 855 | 0.0053 | -0.3681 | 0.00 | 4024 | 6269 | 0.5268 | -0.0367 | 0.25 |
| 4150 | 0.00 | 1204 | 3883 | 0.6438 | 0.1362 | 0.69 | 1609 | 3247 | 0.5124 | 0.0885 | 0.56 |
| 0306 | 0.00 | 1497 | 172 | 0.0000 | -0.1047 | 0.00 | 799 | 1607 | 0.5258 | -0.2242 | 1.20 |
| 2515 | 1.53 | 1146 | 2994 | 0.4862 | 0.0112 | 0.63 | 217 | 5994 | 1.0000 | 0.3265 | 1.04 |
| 724 | 0.00 | 2322 | 4198 | 0.4443 | 0.3039 | 0.33 | 4072 | 235 | 0.0000 | 0.0612 | 0.00 |
| 558 | 0.45 | 242 | 5670 | 0.9955 | -0.3382 | 1.10 | 190 | 7221 | 1.0000 | 0.0084 | 0.87 |
| 2720 | 0.00 | 700 | 116 | 0.0000 | 0.0437 | 0.00 | 776 | 101 | 0.0000 | 0.0976 | 0.00 |
| 1287 | 0.88 | 271 | 9033 | 0.9994 | 0.1995 | 0.70 | 2578 | 5236 | 0.4976 | 0.0764 | 0.33 |
| 3429 | 1.52 | 320 | 5690 | 0.9904 | 0.1538 | 1.06 | 282 | 5380 | 0.9963 | 0.0853 | 1.12 |
| 2668 | 0.77 | 2139 | 8992 | 0.7274 | 0.2105 | 0.36 | 313 | 9371 | 1.0000 | -0.0405 | 0.66 |

Figure 35 Subcolumn Changes Reflected in the Table

Deleting a User-Defined Subcolumn

To delete a user-defined subcolumn:

1. Right-click in the heading of the subcolumn you want to edit.
2. Select **Delete Column** from the context menu (Figure 36).



| Sample 1 1485978108_A | | | GType | Score | Theta | R | Theta/R | | GType | Score |
|--------------------------|---------|-----|-------|--------|--------|--------|---------|----|--------------------------|-------|
| Name | Address | Chr | GType | Score | Theta | R | Theta/R | | GType | Score |
| rs10000092 | 4150... | 4 | AA | 0.8742 | 0.0234 | 0.4697 | 0.05 | | Find... | |
| rs1000055 | 6560... | 18 | AA | 0.8672 | 0.0205 | 2.0286 | 0.01 | | Column Statistics... | |
| rs100016 | 7570... | 14 | NC | 0.0388 | 0.4050 | 1.2672 | 0.32 | | Edit Column... | |
| rs10003241 | 3180... | 4 | BB | 0.9526 | 0.9734 | 0.7683 | 1.27 | | Delete Column... | |
| rs10004197 | 4290... | 4 | BB | 0.9421 | 0.9790 | 0.3957 | 2.47 | | Change Decimal Places... | |
| rs1000449 | 1303... | 14 | AA | 0.7831 | 0.0202 | 4.2238 | 0.00 | | | |
| rs10007472 | 6940... | 4 | AA | 0.8417 | 0.0237 | 1.8286 | 0.01 | AA | 0.8 | |
| rs1000772 | 7320... | 3 | BB | 0.8948 | 1.0000 | 1.1643 | 0.86 | BB | 0.8 | |
| rs10008418 | 4050... | 4 | AA | 0.9510 | 0.0132 | 0.4967 | 0.03 | NC | 0.0 | |

Figure 36 Deleting a Column

A dialog box appears (Figure 37).

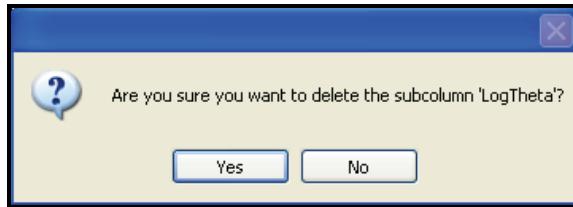


Figure 37 Delete Subcolumn Dialog Box

3. Click Yes.

The subcolumn is deleted from the table (Figure 38).

 A screenshot of a software interface titled 'Full Data Table' and 'SNP Table'. The main area displays a table with two sample sections: 'Sample 1' (1485978108_A) and 'Sample 2' (1485986027_A). The table has columns for Name, Address, Chr, GType, Score, Theta, R, and GType. The 'Theta' column is present in Sample 1 but absent in Sample 2. The table contains approximately 15 rows of data.

| | Name | Address | Chr | GType | Score | Theta | R | GType | Score | Theta | R | GType |
|------------|---------|---------|-----|--------|--------|--------|----|--------|--------|--------|----|-------|
| rs10000092 | 4150... | 4 | AA | 0.8742 | 0.0234 | 0.4697 | AA | 0.8742 | 0.0149 | 0.7335 | AA | |
| rs1000055 | 6560... | 18 | AA | 0.8672 | 0.0205 | 2.0286 | AA | 0.8659 | 0.0203 | 1.5255 | BB | |
| rs100016 | 7570... | 14 | NC | 0.0388 | 0.4050 | 1.2672 | AA | 0.8830 | 0.0491 | 0.6957 | AA | |
| rs10003241 | 3180... | 4 | BB | 0.9526 | 0.9734 | 0.7683 | BB | 0.9526 | 0.9834 | 0.9334 | AB | |
| rs10004197 | 4290... | 4 | BB | 0.9421 | 0.9790 | 0.3957 | AA | 0.9421 | 0.0260 | 0.5475 | AA | |
| rs1000449 | 1303... | 14 | AA | 0.7831 | 0.0202 | 4.2238 | AA | 0.7831 | 0.0188 | 3.1822 | AA | |
| rs10007472 | 6940... | 4 | AA | 0.8417 | 0.0237 | 1.8266 | AA | 0.8417 | 0.0223 | 1.8723 | AB | |
| rs1000772 | 7320... | 3 | BB | 0.8948 | 1.0000 | 1.1643 | BB | 0.8948 | 0.9955 | 0.8707 | BB | |
| rs10008418 | 4050... | 4 | AA | 0.9516 | 0.0132 | 0.4967 | NC | 0.0274 | 0.3944 | 0.7256 | BB | |
| rs1000858 | 1050... | 2 | NC | 0.0822 | 0.3475 | 0.5948 | BB | 0.9709 | 0.9937 | 0.6534 | BB | |
| rs10008660 | 6960... | 4 | BB | 0.9512 | 1.0000 | 1.1960 | AB | 0.5004 | 0.4383 | 1.4567 | AB | |

Figure 38 Subcolumn Deleted from the Table

Filtering Rows

If you want to work with a portion of your data that falls within certain parameters, you can filter the data table to display only rows that meet certain criteria.

To filter a table:

1. Click Filter Rows.

The **Filter Table Rows** dialog box appears (Figure 39).

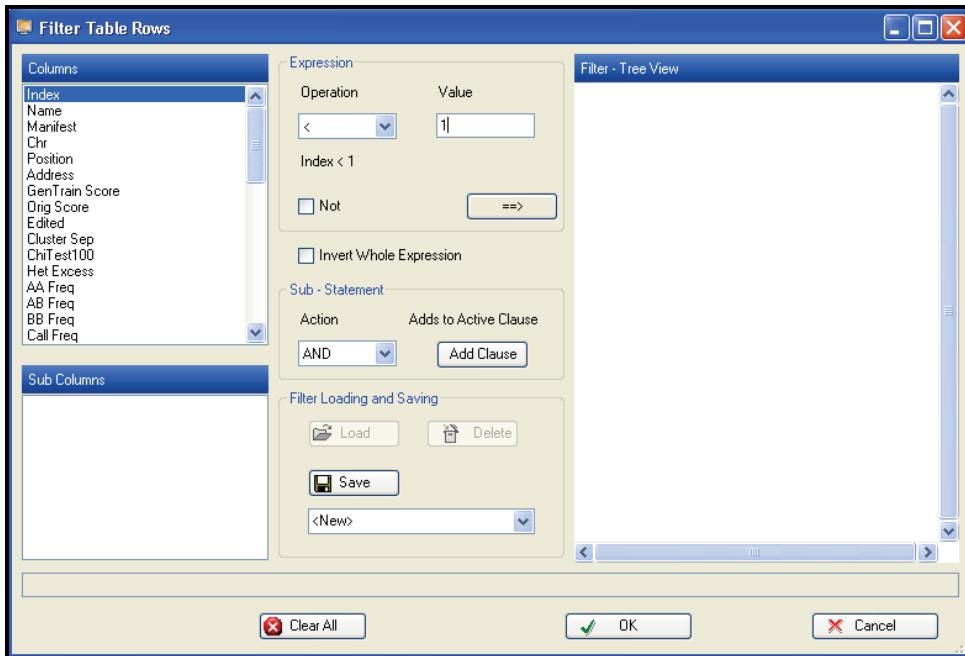


Figure 39 Filter Table Rows Dialog Box

2. Select a column.
If the column you have selected has subcolumns, select a subcolumn as well.
3. Select an operation from the **Operation** dropdown menu.
4. Enter a value in the **Value** text field.
5. Click **==>** to add the column to the filter.
6. To add a substatement, select an action in the **Sub-Statement** area.
7. Click **Add Clause**.



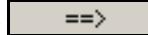
You may enter multiple expressions linked by AND, OR, etc.

8. Click **OK**.

Alternatively, you can filter table rows in some tables by selecting one or more rows and right-clicking. Choose **Show Only Selected Rows** from the context menu. The table is filtered according to the rows you selected.

Creating a Simple Filter

For example, if you want to filter your table so that only SNPs on chromosome 1 are displayed:

1. Select the **Chr** column.
2. Select the **=** operation.
3. Enter **1** in the **Value** text field.
4. Click .

The dialog box should look similar to the one shown in Figure 40.

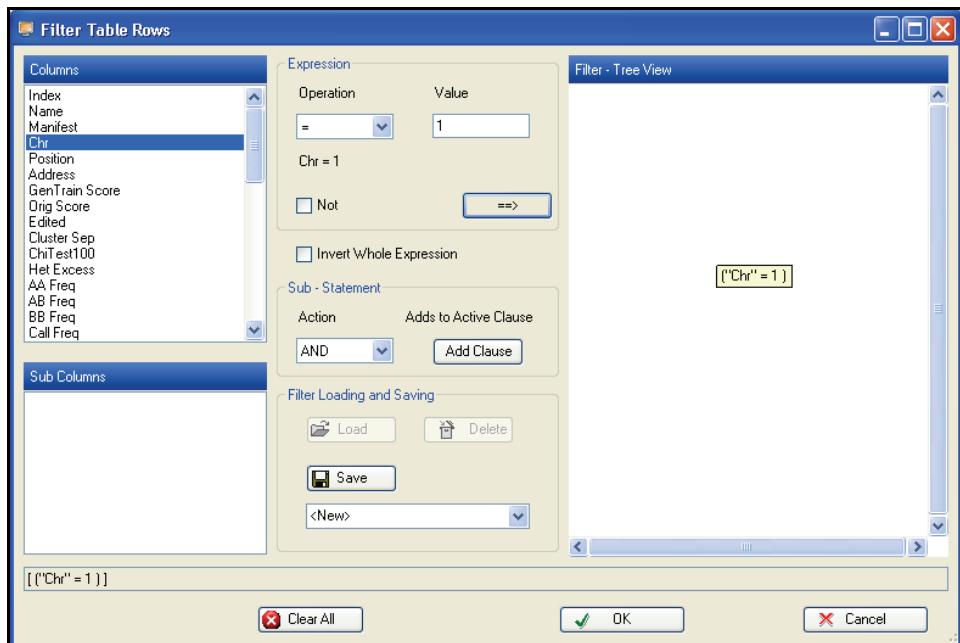
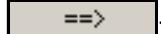
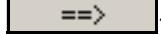
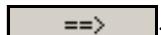


Figure 40 Creating a Simple Filter

Creating a More Complex Filter

You can also create more complex boolean expressions.

For example, you can select all SNPs on chromosome 1 where Sample 1's genotype is AA or Sample 2's score is greater than 0.5:

1. Select the **Chr** column.
2. Select the **=** operation.
3. Enter 1 in the **Value** text field.
4. Click .
5. Select **OR** from the **Action** pulldown menu.
6. Click **Add Clause**.
- This adds a circle with an OR in the tree view. The circle is highlighted in pink, indicating that this is the active node to which leaves will be added (Figure 41).
7. Select **Sample 1** in the **Columns** listbox.
8. Select **GType** in the **Subcolumns** listbox.
9. Type AA in the **Value** text field.
10. Click .
11. Select **Sample 2** in the **Columns** listbox.
12. Select **Score** in the **Subcolumns** listbox.
13. Select **>** in the **Operation** pulldown menu.
14. Type 0.5 in the **Value** text field.
15. Click .

The dialog box should look similar to the one shown in Figure 41.

Note that your filter formula is displayed as a formula near the bottom of the window. In this case, it is [(Chr = 1) AND [(Sample 1.GType = AA) OR (Sample 2.Score > 0.5)]].



The sub-statement root node is always AND unless you right-click it and select an alternate root node. AND is implied for the first action. AND will only appear once you add a clause.

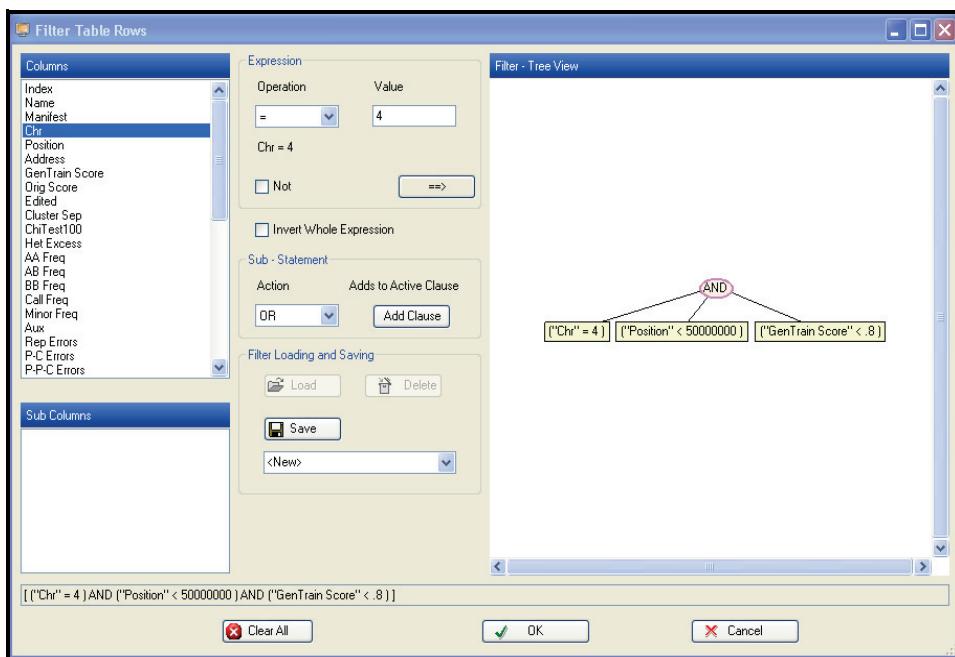


Figure 41 Creating a More Complex Filter

Saving a Filter

Filters can be saved and used later in different projects.

To save a filter for later use:

1. Build your filter.
2. In the **Filter Loading and Saving** area of the **Filter Table Rows**, click **Save**.

The **Enter Name for Filter** dialog box appears.



Figure 42 Entering a Name for a Filter

3. Enter a name for your filter in the text field.



NOTE

To ensure that BeadStudio can identify the filter, use only alphanumeric characters in the filter name.

4. Click **OK**.

Your filter is saved to *C:\Documents and Settings\<username>\Application Data\Illumina\BeadStudio\<filter name>.flt*

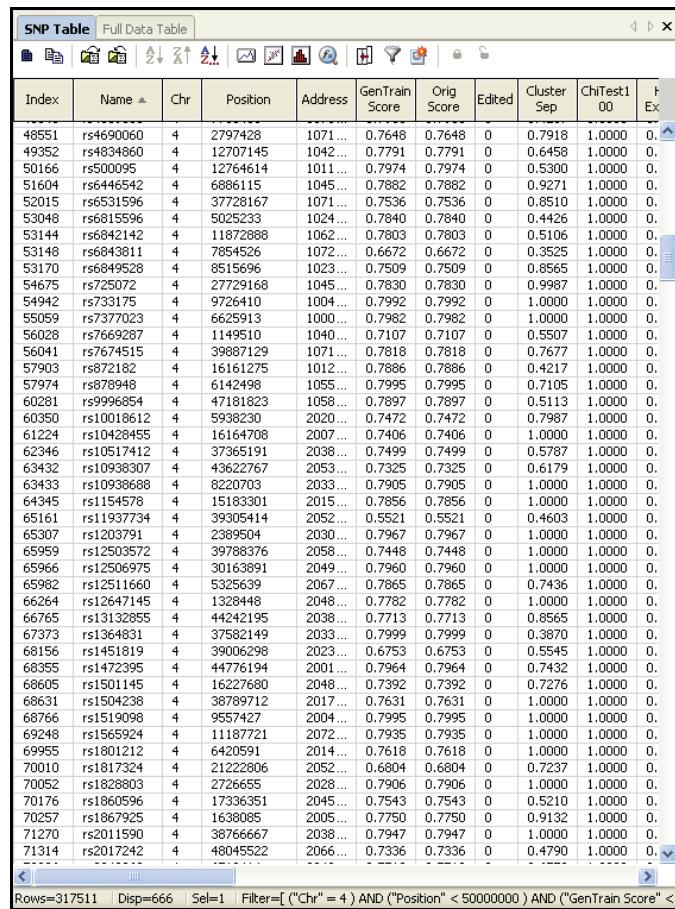
Loading a Filter

To load and use a filter you previously saved:

1. In the **Filter Loading and Saving** area of the **Filter Table Rows**, select the filter you want to use from the dropdown menu.
The **Load** button becomes active.
2. Click **Load**.
Your filter is loaded and ready to use.

Results of Filtering

Figure 43 shows the results of the example filter shown in Figure 41. The filter formula is displayed in the status bar of the table, along with the number of rows that passed the filter.



The screenshot shows the BeadStudio SNP Table interface. The window title is "SNP Table | Full Data Table". The table has the following columns: Index, Name (sorted), Chr, Position, Address, GenTrain Score, Orig Score, Edited, Cluster Sep, ChiTest1 00, and F Ex. The data grid contains approximately 31,751 rows of SNP information. The status bar at the bottom displays the filter formula: "Filter=[("Chr" = 4) AND ("Position" < 50000000) AND ("GenTrain Score" < 0.7536)]".

Figure 43 Results of Filtering

Clearing a Filter

To clear the filter:

- ▶ Click  **Clear Filter**.
The filter is cleared.

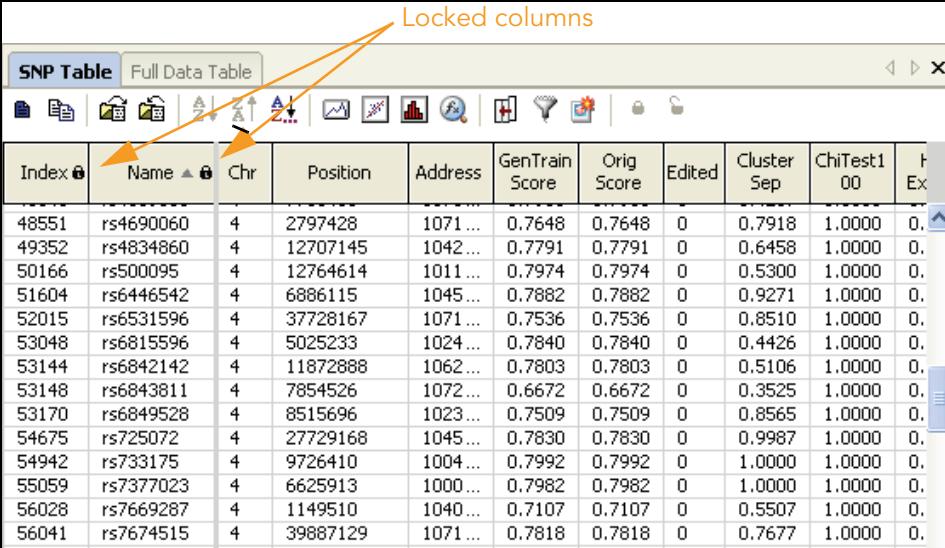
Filtering Notes

- ▶ Leaves are added to the currently selected node (highlighted in pink).
- ▶ To change the active node, click another node.
- ▶ To change the action for a node, right-click the node and select the action from the popup menu.
- ▶ To modify a leaf's value, first click it to select it, then change options within the expression area of the **Filter Table Rows**.
- ▶ To delete a node or a leaf, right-click it and select **Delete** from the context menu.
- ▶ To clear all actions and expressions, click **Clear All**.

Locking Columns

Locking columns can be useful when there are many columns in a table and you want to keep a particular column or a few columns visible. Locking a column “freezes” a column on the left side of the display, so that you can see it while scrolling through other columns.

A Locked column is indicated by a  lock symbol in the column's header (Figure 44).



Locked columns

| Index | Name | Chr | Position | Address | GenTrain Score | Orig Score | Edited | Cluster Sep | ChiTest1 00 | H Ex |
|-------|-----------|-----|----------|----------|----------------|------------|--------|-------------|-------------|------|
| 48551 | rs4690060 | 4 | 2797428 | 1071 ... | 0.7648 | 0.7648 | 0 | 0.7918 | 1.0000 | 0. |
| 49352 | rs4834860 | 4 | 12707145 | 1042 ... | 0.7791 | 0.7791 | 0 | 0.6458 | 1.0000 | 0. |
| 50166 | rs500095 | 4 | 12764614 | 1011 ... | 0.7974 | 0.7974 | 0 | 0.5300 | 1.0000 | 0. |
| 51604 | rs6446542 | 4 | 6886115 | 1045 ... | 0.7882 | 0.7882 | 0 | 0.9271 | 1.0000 | 0. |
| 52015 | rs6531596 | 4 | 37728167 | 1071 ... | 0.7536 | 0.7536 | 0 | 0.8510 | 1.0000 | 0. |
| 53048 | rs6815596 | 4 | 5025233 | 1024 ... | 0.7840 | 0.7840 | 0 | 0.4426 | 1.0000 | 0. |
| 53144 | rs6842142 | 4 | 11872888 | 1062 ... | 0.7803 | 0.7803 | 0 | 0.5106 | 1.0000 | 0. |
| 53148 | rs6843811 | 4 | 7854526 | 1072 ... | 0.6672 | 0.6672 | 0 | 0.3525 | 1.0000 | 0. |
| 53170 | rs6849528 | 4 | 8515696 | 1023 ... | 0.7509 | 0.7509 | 0 | 0.8565 | 1.0000 | 0. |
| 54675 | rs725072 | 4 | 27729168 | 1045 ... | 0.7830 | 0.7830 | 0 | 0.9987 | 1.0000 | 0. |
| 54942 | rs733175 | 4 | 9726410 | 1004 ... | 0.7992 | 0.7992 | 0 | 1.0000 | 1.0000 | 0. |
| 55059 | rs7377023 | 4 | 6625913 | 1000 ... | 0.7982 | 0.7982 | 0 | 1.0000 | 1.0000 | 0. |
| 56028 | rs7669287 | 4 | 1149510 | 1040 ... | 0.7107 | 0.7107 | 0 | 0.5507 | 1.0000 | 0. |
| 56041 | rs7674515 | 4 | 39887129 | 1071 ... | 0.7818 | 0.7818 | 0 | 0.7677 | 1.0000 | 0. |

Figure 44 Table with Two Locked Columns

To lock columns:

To see a list of locked columns in the **Column Chooser**:

- Click  **Column Chooser**.

Locked columns are displayed in the **Display-Locked Columns** area (Figure 45).

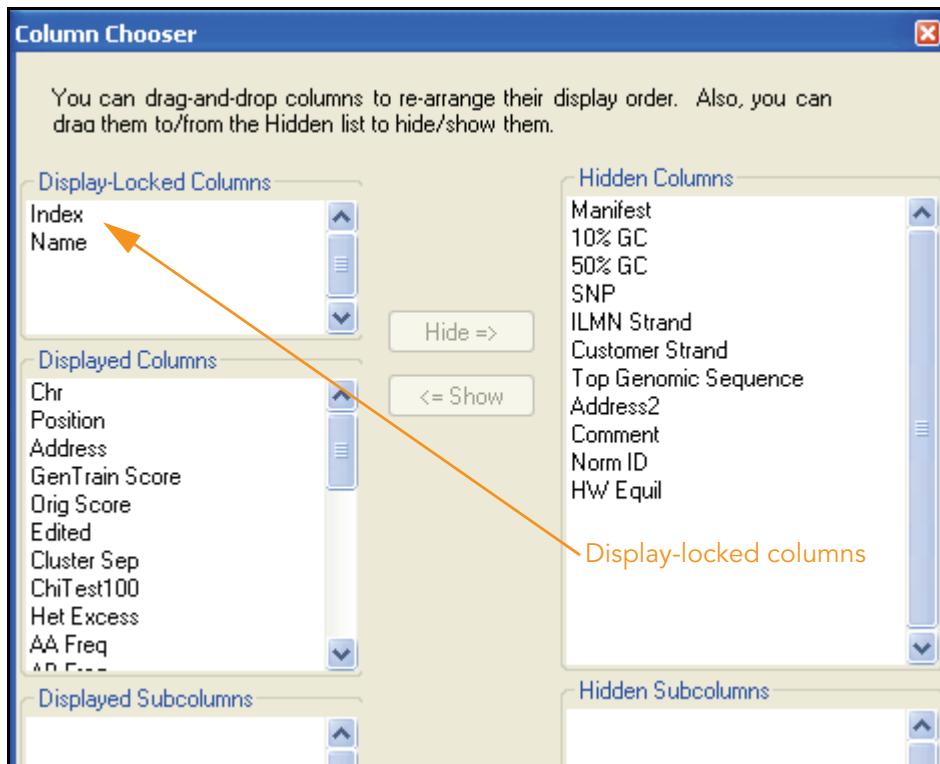


Figure 45 Column Chooser, Display-Locked Columns

Unlocking Columns

To unlock columns:

1. Select the column header of each locked column you want to unlock (Figure 46).

Select the columns you want to unlock, then click **Unlock**.

| Index | Name | Chr | Position | Address | GenTrain Score | Orig Score | Edited | Cluster Sep | ChiTest1 00 | t Ex |
|-------|-----------|-----|----------|---------|----------------|------------|--------|-------------|-------------|------|
| 2137 | rs1392592 | 4 | 21978621 | 6960... | 0.7950 | 0.7950 | 0 | 1.0000 | 1.0000 | 0. |
| 2168 | rs1402037 | 4 | 12467551 | 3120... | 0.7969 | 0.7969 | 0 | 0.6969 | 1.0000 | 0. |
| 2326 | rs1465522 | 4 | 20314215 | 6980... | 0.7664 | 0.7664 | 0 | 0.4769 | 1.0000 | 0. |
| 2985 | rs1913332 | 4 | 21613078 | 3450... | 0.7543 | 0.7543 | 0 | 1.0000 | 1.0000 | 0. |
| 3737 | rs2285084 | 4 | 2943293 | 4860... | 0.7792 | 0.7792 | 0 | 1.0000 | 1.0000 | 0. |
| 4898 | rs3733591 | 4 | 9598399 | 7150... | 0.7936 | 0.7936 | 0 | 0.5073 | 1.0000 | 0. |
| 5132 | rs3843438 | 4 | 7394613 | 1110... | 0.7704 | 0.7704 | 0 | 0.7055 | 1.0000 | 0. |
| 5373 | rs4298137 | 4 | 14480063 | 4880... | 0.7978 | 0.7978 | 0 | 0.4064 | 1.0000 | 0. |
| 5403 | rs4331786 | 4 | 38591974 | 4570... | 0.7891 | 0.7891 | 0 | 1.0000 | 1.0000 | 0. |
| 5470 | rs4407490 | 4 | 26515729 | 6860... | 0.7808 | 0.7808 | 0 | 0.8918 | 1.0000 | 0. |
| 5699 | rs4689562 | 4 | 7052727 | 1110... | 0.7846 | 0.7846 | 0 | 0.5472 | 1.0000 | 0. |

Figure 46 Selecting Columns to Unlock

2. Click **Unlock**.

The selected columns are unlocked (Figure 47).

The selected columns are unlocked.

| Index | Name | Chr | Position | Address | GenTrain Score | Orig Score | Edited | Cluster Sep | ChiTest1 00 | t Ex |
|-------|-----------|-----|----------|---------|----------------|------------|--------|-------------|-------------|------|
| 2137 | rs1392592 | 4 | 21978621 | 6960... | 0.7950 | 0.7950 | 0 | 1.0000 | 1.0000 | 0. |
| 2168 | rs1402037 | 4 | 12467551 | 3120... | 0.7969 | 0.7969 | 0 | 0.6969 | 1.0000 | 0. |
| 2326 | rs1465522 | 4 | 20314215 | 6980... | 0.7664 | 0.7664 | 0 | 0.4769 | 1.0000 | 0. |
| 2985 | rs1913332 | 4 | 21613078 | 3450... | 0.7543 | 0.7543 | 0 | 1.0000 | 1.0000 | 0. |
| 3737 | rs2285084 | 4 | 2943293 | 4860... | 0.7792 | 0.7792 | 0 | 1.0000 | 1.0000 | 0. |
| 4898 | rs3733591 | 4 | 9598399 | 7150... | 0.7936 | 0.7936 | 0 | 0.5073 | 1.0000 | 0. |
| 5132 | rs3843438 | 4 | 7394613 | 1110... | 0.7704 | 0.7704 | 0 | 0.7055 | 1.0000 | 0. |
| 5373 | rs4298137 | 4 | 14480063 | 4880... | 0.7978 | 0.7978 | 0 | 0.4064 | 1.0000 | 0. |
| 5403 | rs4331786 | 4 | 38591974 | 4570... | 0.7891 | 0.7891 | 0 | 1.0000 | 1.0000 | 0. |
| 5470 | rs4407490 | 4 | 26515729 | 6860... | 0.7808 | 0.7808 | 0 | 0.8918 | 1.0000 | 0. |
| 5699 | rs4689562 | 4 | 7052727 | 1110... | 0.7846 | 0.7846 | 0 | 0.5472 | 1.0000 | 0. |

Figure 47 Selected Columns Unlocked

Chapter 4

Graphs

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Introduction

The graphing functions of BeadStudio allow you to plot data displayed in tables in a number of ways.

Histograms

Use the **Histogram** option to plot the distribution of values for a particular column.

1. In the table toolbar, click  **Histogram**.

The **BeadStudio Histogram** window appears (Figure 48).

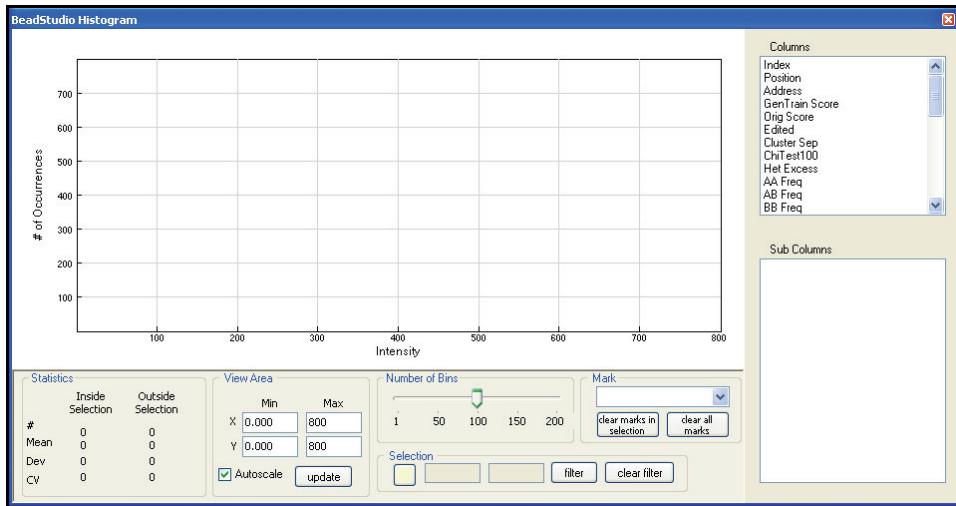


Figure 48 BeadStudio Histogram Window

2. Select graphing options from the **Columns** and **Subcolumns** listboxes.

A histogram displaying the options you selected appears (Figure 49).

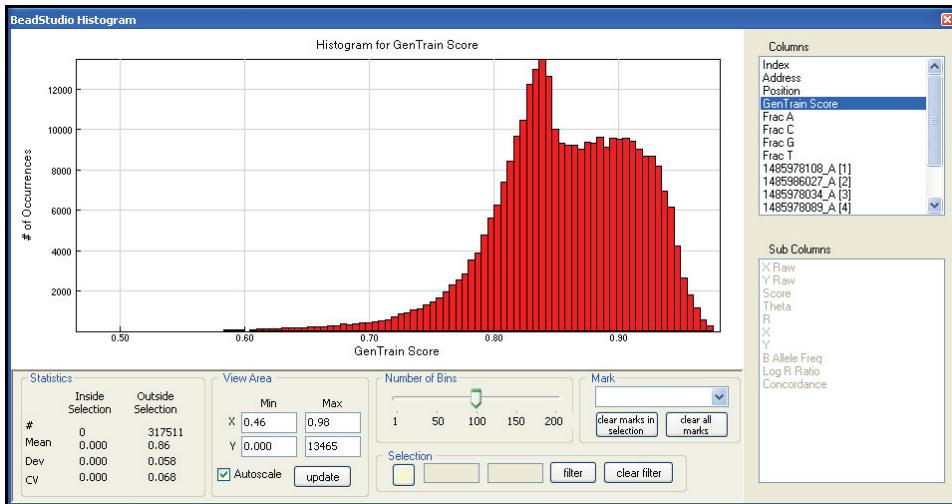


Figure 49 Histogram

Adjusting the View Area

If you want to view a subset of the table data, you can customize a histogram to display only data that falls within a specific range.

To adjust the view area in a histogram, do the following:

1. In the **View Area** of the **BeadStudio Histogram** window, select parameters that correspond to the region you want to view:
 - X Min
 - X Max
 - Y Min
 - Y Max
2. Click **Update**.

The histogram displays with the parameters you selected.

Autoscale is selected by default. When **Autoscale** is selected, the scale of the histogram automatically adjusts when you choose a different column. If you clear the **Autoscale** checkbox, the scale of the histogram does not adjust when you choose a different column.

Changing the Number of Bins

You can change the resolution of a histogram by changing the number of bins displayed in the histogram.

To change the number of bins displayed in a histogram:

- In the **Number of Bins** area, click and drag the slider.
 - Drag to the left to decrease the number of bins.
 - Drag to the right to increase the number of bins.

The number of bins displayed in the histogram updates dynamically, based on your selection.

Selecting Data

From within a histogram, you can visually select a range of data you are interested in viewing or analyzing.

To select data in a histogram, do one of the following:

- Double-click the histogram.
- In the **Selection** area, click .

A shaded selection area appears in the histogram (Figure 50).

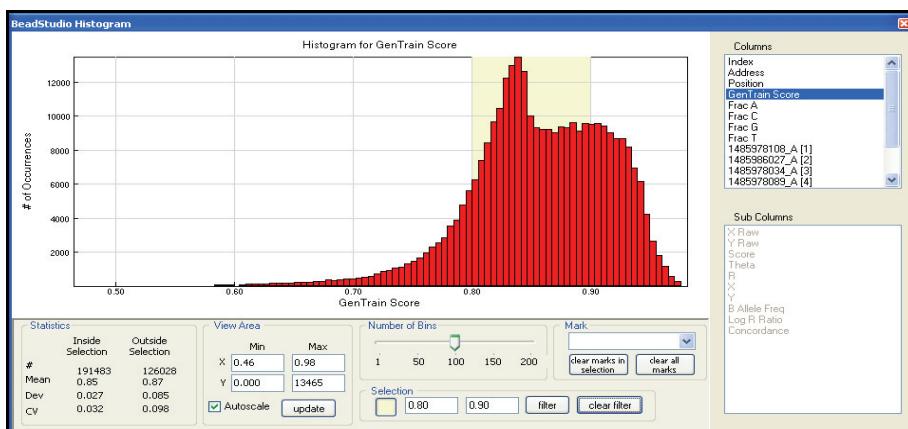


Figure 50 Histogram with Shaded Selection Area

To change the size of the shaded selection area, do one of the following:

- Drag the boundaries of the shaded selection area.
- a. Move the cursor over the edge of the shaded selection area until the cursor changes to this symbol: <-l->.

- b. Click and drag the left and right boundaries of the shaded selection area to the desired positions.
 The shaded selection area dynamically changes as you drag its left and right boundaries.

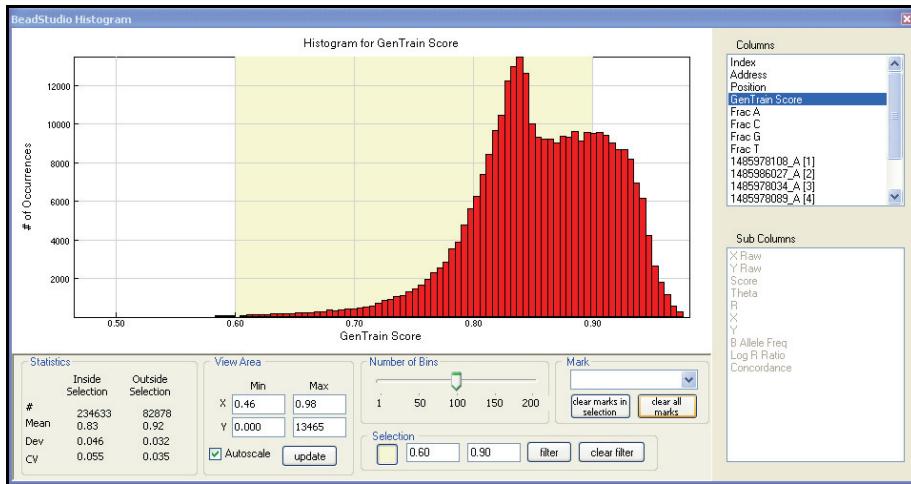


Figure 51 Resizing the Shaded Selection Area

- Type boundaries in the **Selection** settings area.
 - In the **Selection** area, type lower (left) and upper (right) boundaries to define the shaded selection area of the histogram.

The shaded selection area dynamically updates to the settings you specify.

Viewing Statistics

You can view table data statistics in the **Statistics** area of the **BeadStudio Histogram** window. Statistics dynamically update based on the shaded selection area of the histogram.

Inside Selection refers to data that fall inside the shaded selection area of the histogram.

Outside Selection refers to data that fall outside the shaded selection area of the histogram.

Statistics apply to the column currently plotted in the histogram.

Table 3 Histogram Statistics

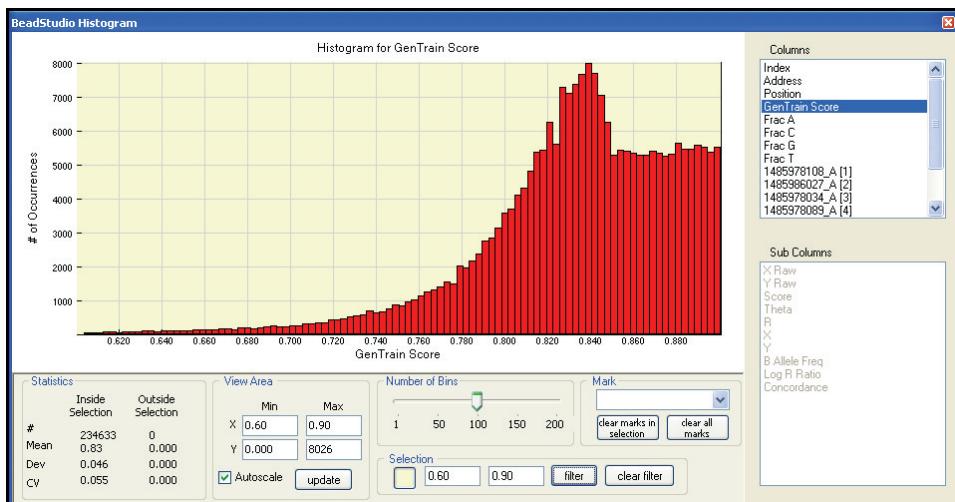
| Statistic | Description |
|-----------|--------------------------|
| # | Number of occurrences |
| Mean | Mean values |
| Dev | Standard deviation |
| CV | Coefficient of variation |

Filtering Data

To filter table data from within a histogram:

1. First select the data you want to filter by following the procedure *Selecting Data* on page 55.
2. In the **Selection** settings area of the **BeadStudio Histogram** window, click .

The table data is filtered according to the shaded selection area. The histogram displays only the filtered data (Figure 52).

**Figure 52** Filtering Selected Data

Clearing a Filter

To clear a filter from within a histogram:

- Click **clear filter**.

The filter is removed and the histogram appears as it did prior to filtering the data.

Marking a Selection

To mark a selection in a histogram, do the following:

1. In the **Mark** area of the **BeadStudio Histogram**, select **Configure Marks** from the dropdown menu.
The **Configure Marks** dialog box appears (Figure 53).

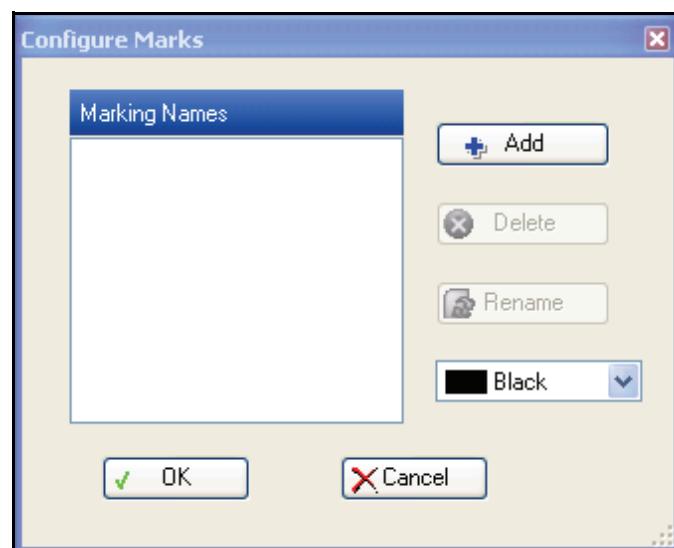


Figure 53 Configure Marks Dialog Box

2. Click **Add**.
3. The **Select Mark Name** dialog box appears (Figure 54).



Figure 54 Select Mark Name Dialog Box

4. Type a name for your mark in the text field.
5. Select a color for your mark from the dropdown menu.



Figure 55 Choosing a Mark Name and Color

6. Click **OK**.
7. In the **Configure Marks** dialog box, click **OK**.
The mark you created is displayed in the **Mark** area dropdown menu.
8. Select the mark you want to apply to your data from the **Mark** area dropdown menu.

After a few seconds, the marked data is displayed in the histogram with the properties of the mark you applied (Figure 56).

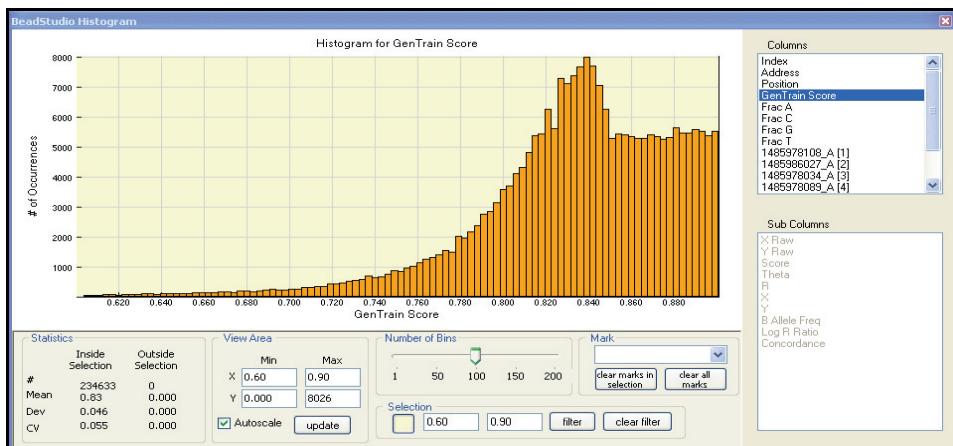


Figure 56 Marked Data Displayed in a Histogram

The data are also marked in the associated table (Figure 57).

| Index | Name | Address | Chr | GenTrain Score | Frac A | Frac C | Frac G | Frac T | GT |
|-------|------------|---------|-----|----------------|--------|--------|--------|--------|----|
| 1 | rs10000092 | 4150... | 4 | 0.8418 | 0.3922 | 0.1667 | 0.2059 | 0.2353 | AA |
| 2 | rs10000055 | 6560... | 18 | 0.8363 | 0.2549 | 0.2941 | 0.2157 | 0.2353 | AA |
| 3 | rs100016 | 7570... | 14 | 0.8489 | 0.2451 | 0.2157 | 0.1961 | 0.3431 | NC |
| 4 | rs10003241 | 3180... | 4 | 0.9203 | 0.3529 | 0.0882 | 0.1765 | 0.3824 | BB |
| 5 | rs10004197 | 4290... | 4 | 0.9070 | 0.3529 | 0.1961 | 0.1863 | 0.2647 | BB |
| 6 | rs1000449 | 130397 | 14 | 0.7796 | 0.4020 | 0.2157 | 0.2157 | 0.1667 | AA |
| 7 | rs10007472 | 6940... | 4 | 0.8176 | 0.3137 | 0.2157 | 0.2059 | 0.2647 | AA |
| 8 | rs1000772 | 7320... | 3 | 0.8590 | 0.1275 | 0.3137 | 0.2941 | 0.2647 | BB |
| 9 | rs10008418 | 4050... | 4 | 0.9181 | 0.3137 | 0.1078 | 0.2451 | 0.3333 | AA |
| 10 | rs1000858 | 1050... | 2 | 0.9470 | 0.3431 | 0.1961 | 0.2451 | 0.2157 | NC |
| 11 | rs10008860 | 6960... | 4 | 0.9184 | 0.1765 | 0.2353 | 0.3725 | 0.2157 | BB |
| 12 | rs1000940 | 3990... | 17 | 0.7872 | 0.1373 | 0.3039 | 0.2157 | 0.3431 | AA |
| 13 | rs10010359 | 1030... | 4 | 0.9369 | 0.1471 | 0.3039 | 0.1765 | 0.3725 | AA |
| 14 | rs1001193 | 360050 | 1 | 0.8695 | 0.2549 | 0.1765 | 0.1961 | 0.3725 | AA |
| 15 | rs10012204 | 6400... | 4 | 0.8719 | 0.3431 | 0.1471 | 0.2353 | 0.2745 | BB |
| 16 | rs1001452 | 7650... | 12 | 0.9645 | 0.4118 | 0.2255 | 0.1275 | 0.2353 | AA |
| 17 | rs1001587 | 670176 | 22 | 0.8321 | 0.2451 | 0.1961 | 0.3627 | 0.1961 | AB |
| 18 | rs1001628 | 2490... | 2 | 0.9058 | 0.3039 | 0.1569 | 0.0882 | 0.4510 | AA |
| 19 | rs10018142 | 780044 | 4 | 0.9187 | 0.2157 | 0.2647 | 0.2353 | 0.2843 | BB |
| 20 | rs10018513 | 1090... | 4 | 0.8825 | 0.3824 | 0.1275 | 0.2353 | 0.2549 | BB |
| 21 | rs10021084 | 4560... | 4 | 0.8225 | 0.0980 | 0.2549 | 0.3529 | 0.2941 | BB |
| 22 | rs1002182 | 1410... | 5 | 0.9446 | 0.2745 | 0.2059 | 0.2255 | 0.2941 | AB |
| 23 | rs1002244 | 5690... | 21 | 0.9349 | 0.3333 | 0.1275 | 0.1961 | 0.3431 | AA |
| 24 | rs10025341 | 4810... | 4 | 0.8324 | 0.2647 | 0.2941 | 0.1863 | 0.2549 | AA |
| 25 | rs10025951 | 4390... | 4 | 0.7343 | 0.1569 | 0.1863 | 0.1961 | 0.4608 | AA |
| 26 | rs10025977 | 4830... | 4 | 0.8697 | 0.2745 | 0.1961 | 0.2255 | 0.3039 | AA |
| 27 | rs1002607 | 4730... | 9 | 0.9375 | 0.3725 | 0.1765 | 0.1667 | 0.2843 | AA |
| 28 | rs10026892 | 4480... | 4 | 0.9271 | 0.3333 | 0.2353 | 0.1961 | 0.2353 | BB |
| 29 | rs10029070 | 2120... | 4 | 0.8216 | 0.4118 | 0.1078 | 0.2549 | 0.2255 | BB |
| 30 | rs10029245 | 3190... | 4 | 0.8854 | 0.3137 | 0.1275 | 0.2059 | 0.3529 | AA |
| 31 | rs1003081 | 2600... | 11 | 0.8289 | 0.2059 | 0.1863 | 0.2549 | 0.3529 | BB |
| 32 | rs10031159 | 4260... | 4 | 0.9133 | 0.2647 | 0.1471 | 0.1765 | 0.4118 | BB |

Figure 57 Marked Data Displayed in Associated Table

Line Plots

Line plots are also linked to data tables. Data appears in the same order in a line plot as it does in the associated table.

Use the **Line Plot** option to plot data from a single column, or to plot two columns simultaneously, in order to compare them.

Plotting a Single Column

To plot a single column in a line plot, do the following:

1. Click  **Line Plot** in the table to open the **BeadStudio Column Plot** window (Figure 58).

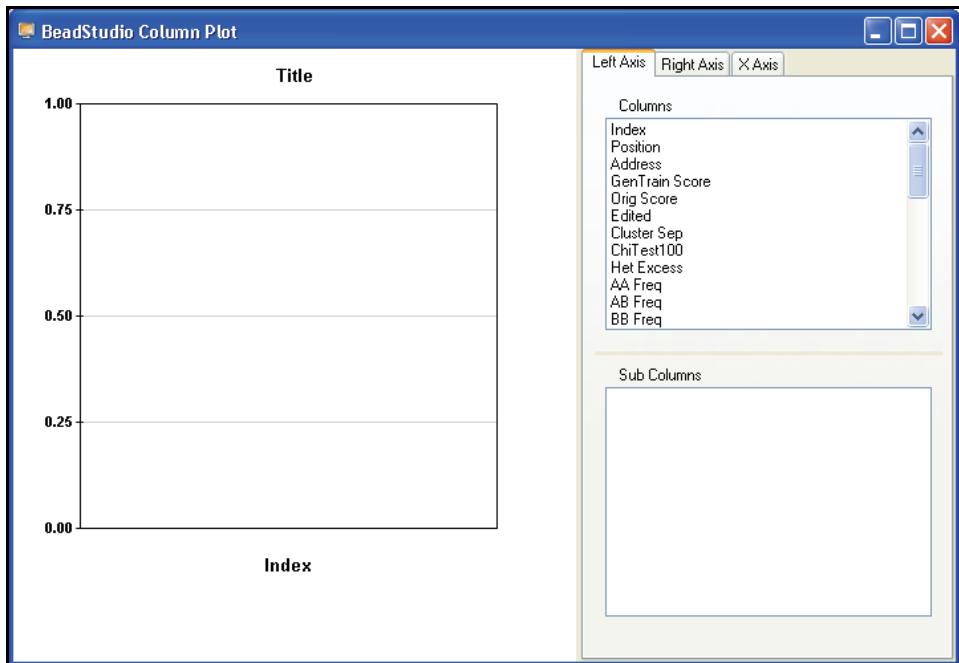


Figure 58 BeadStudio Column Plot Window

2. Choose a graphing option from the **Columns** area (and the **Subcolumns** area, if activated).

The line plot is dynamically populated with the option you chose (Figure 59).

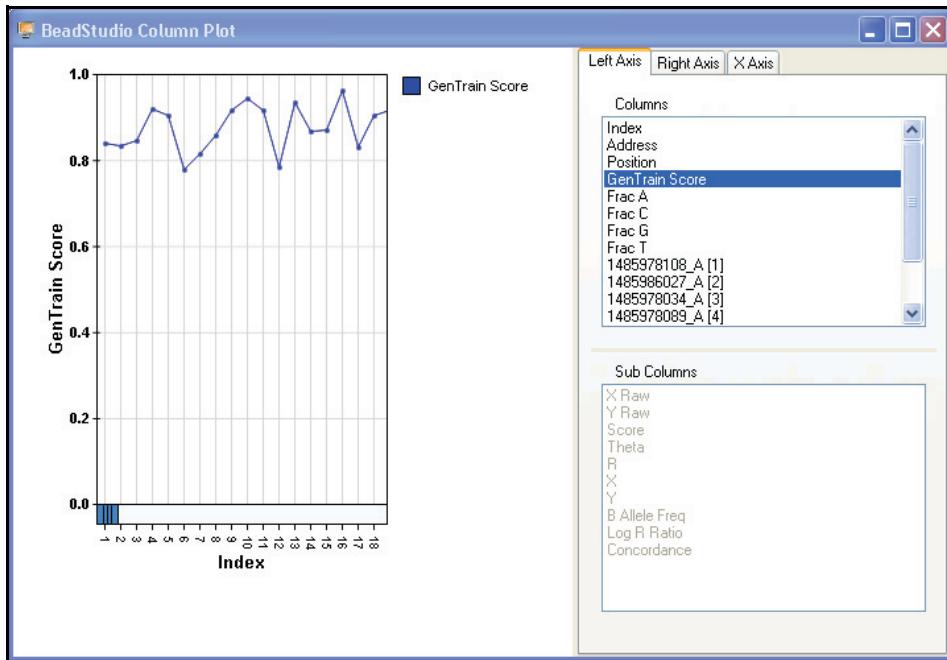


Figure 59 Plotting a Single Column in a Line Plot

Plotting Two Columns

To compare two columns in a line plot, do the following:

1. In the **Columns** area of the **Left Axis** tab (and the **Subcolumns** area, if activated), choose the first column you want to plot.
A line representing the first column appears in the line plot, and an item is added to the legend as shown in Figure 59.
2. Click to select the **Right Axis** tab.
3. In the **Columns** area of the Right Axis tab (and the **Subcolumns** area, if activated), choose the second column you want to plot.

A line representing the second item appears in the line plot, an item is added to the legend, and the right axis is populated (Figure 60).

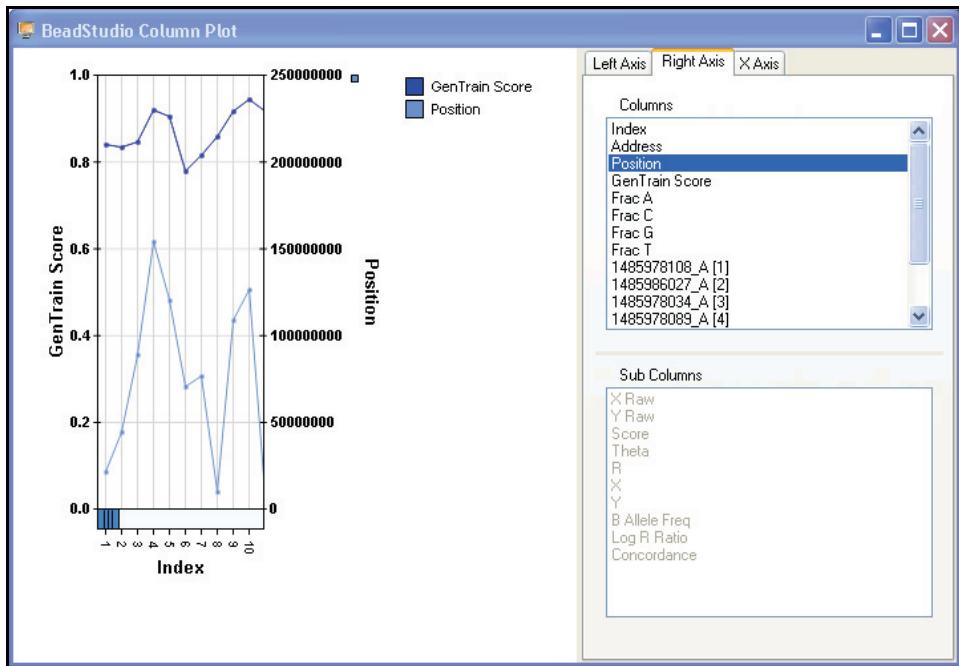


Figure 60 Plotting Two Columns in a Line Plot

Changing the X-Axis Label

You can also change the x-axis label of a line plot, which represents rows of data from the associated table.

To change the x-axis label of a line plot, do the following:

1. In the **BeadStudio Column Plot** window, click the **X-Axis** tab.
2. In the **Columns** area (and in the **Subcolumns** area, if activated), select a label.

The label you selected is applied to the x-axis of the line plot (Figure 61).

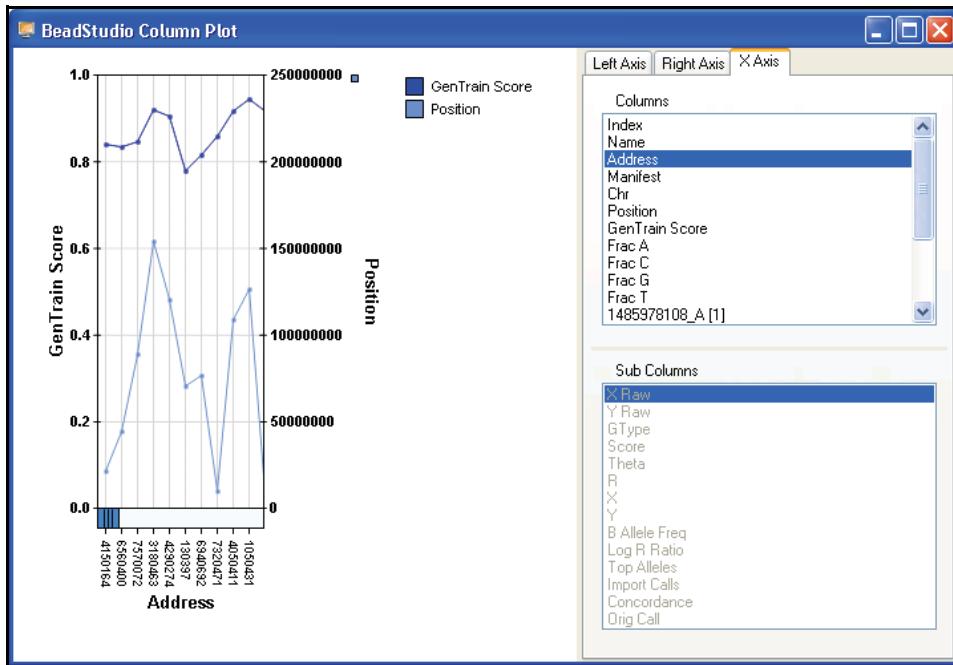


Figure 61 Line Plot with X-Axis Label "Address"

Changing the Plot Type

If you want to change the way your data is displayed from a line plot to a bar graph, do the following:

1. Right-click in a line plot.

The context menu appears (Figure 62).

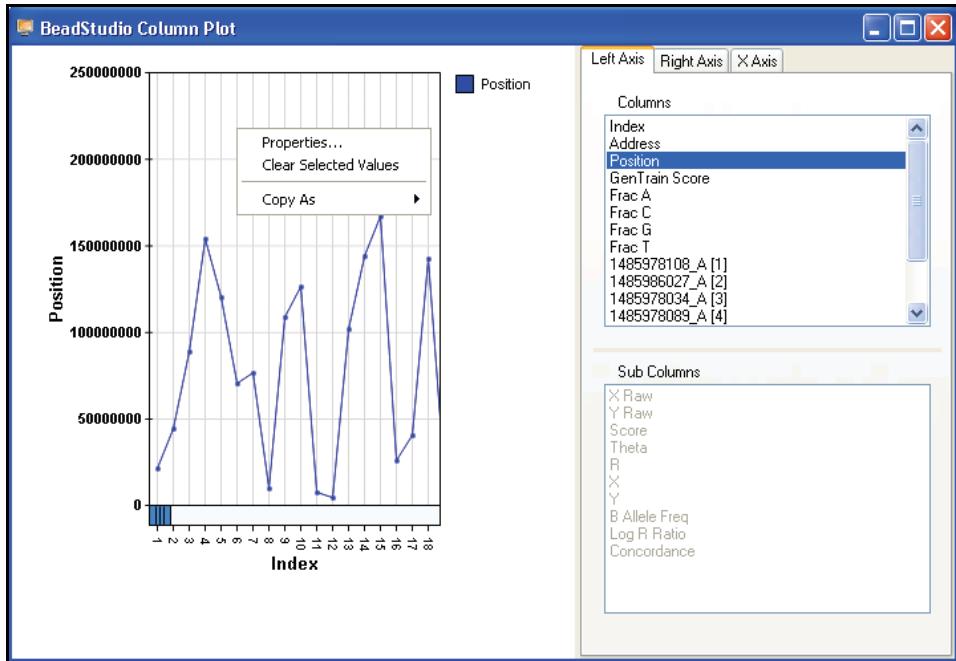


Figure 62 Line Plot Context Menu

2. Select **Properties** from the context menu.

3. The **Plot Settings** dialog box appears (Figure 63).

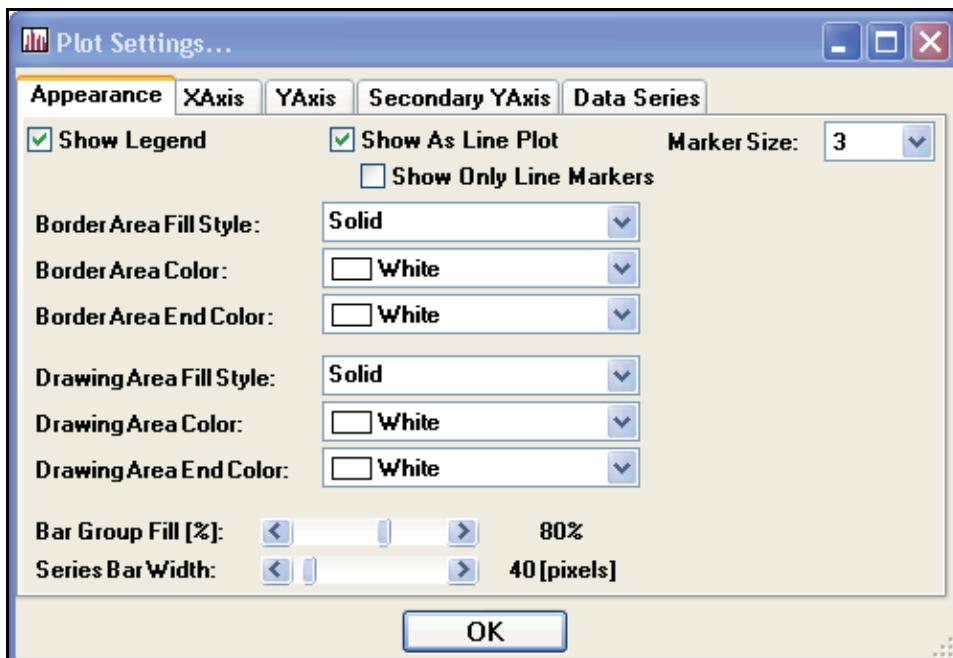


Figure 63 Plot Settings Dialog Box

4. Clear the **Show As Line Plot** checkbox.
5. [Optional] Select additional properties you would like to apply to the bar graph.
6. Click **OK**.

The line plot converts to a bar graph (Figure 64).

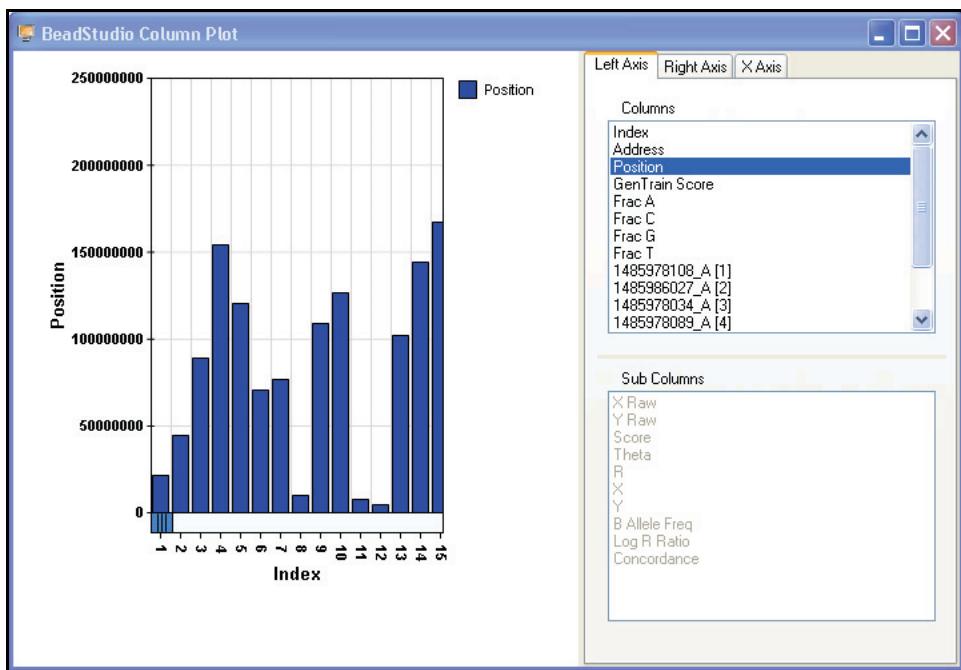


Figure 64 Bar Graph

Scatter Plots

Use the **Scatter Plot** option to see how two columns in the table are related.

1. Click  **Scatter Plots** in the table to open the **Plot Columns** dialog box with **Scatter Plots** selected (Figure 65).

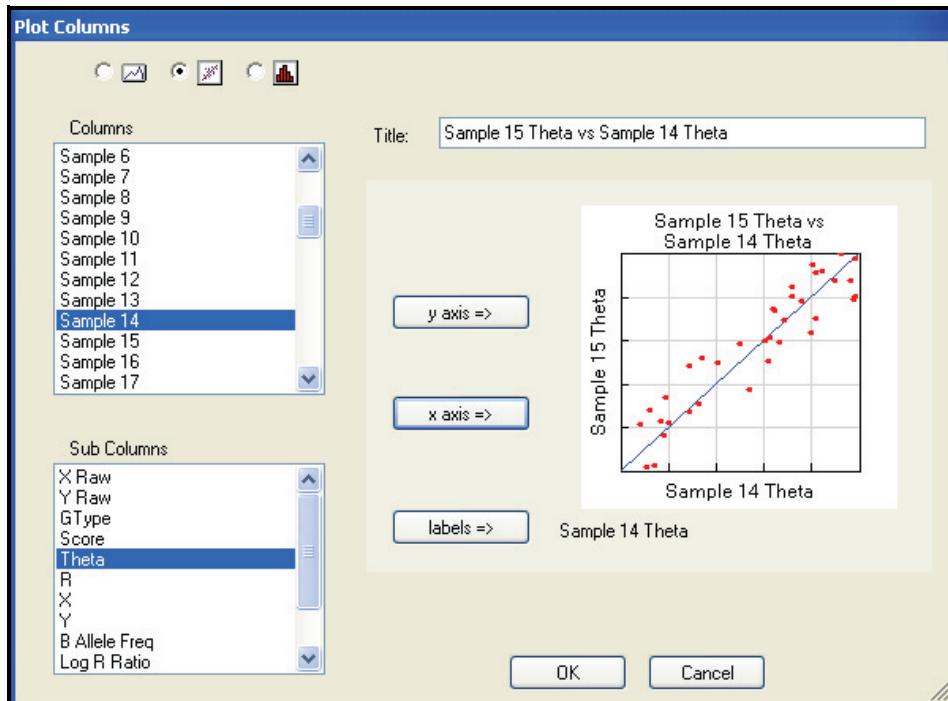


Figure 65 Plot Columns Dialog Box with Scatter Plot Selected

2. Choose graphing options from the **Columns** and **Subcolumns** listboxes and click **OK**.

The scatter plot appears with the parameters you selected (Figure 66).

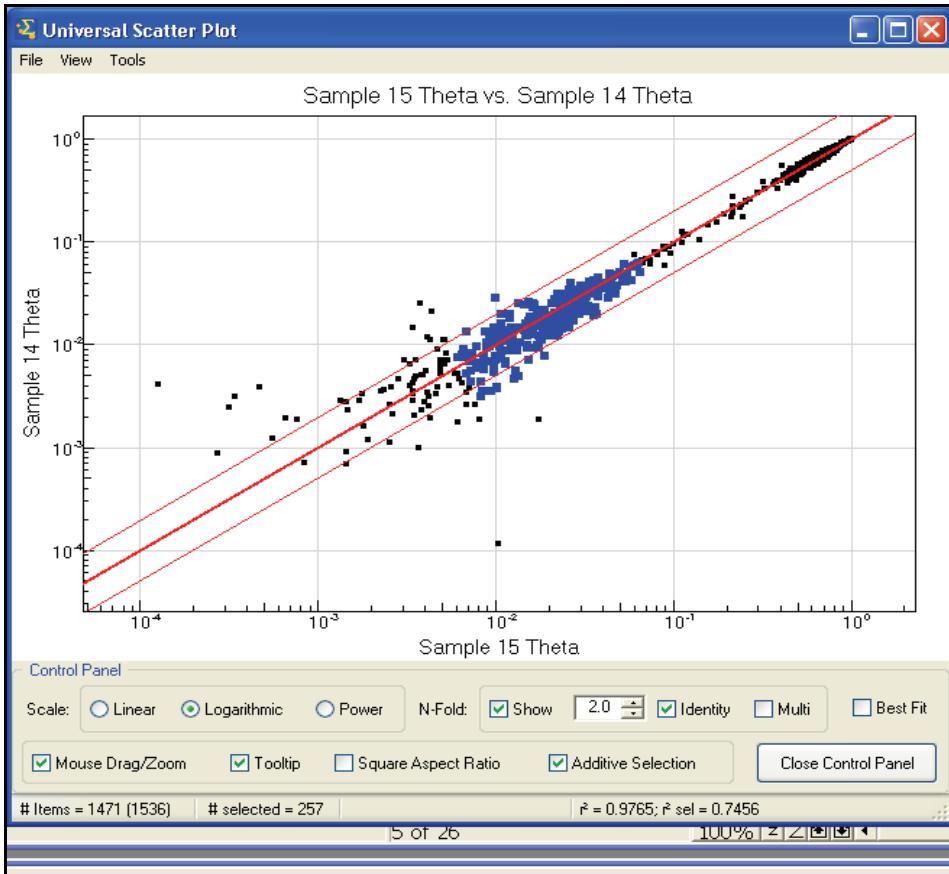


Figure 66 Universal Scatter Plot

Marking Samples

To mark samples in a scatter plot, do the following:

1. Right-click on a scatter plot to display its context menu.
2. Select **Mark Selected Items | <Add New>** (Figure 67).

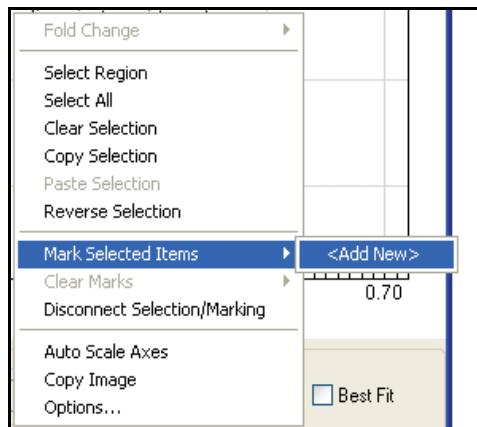


Figure 67 Scatter Plot Context Menu

The **Select Mark Name** dialog box appears (Figure 68).

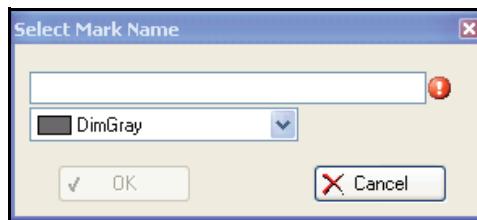


Figure 68 Select Mark Name Dialog Box

3. Type a name for your mark in the text field.
4. Select a color for your mark from the dropdown menu.



Figure 69 Selecting Mark Name and Color

The samples you marked in the scatter plot are displayed in the color you selected. The marked displayed in the associated table in the color you selected.

Heat Maps

Use the **Heat Map** option to plot data from any subcolumn across all columns. The heat map can be used only with tables that have columns with subcolumns.

Click  **Heat Map** in the toolbar of a table with subcolumns to open the **Plot Sample Subcolumns in a Heat Map** dialog box.



NOTE

Heat maps are most useful when your data has been clustered.

Populating the Heat Map with Data

To populate the heat map with data:

1. Click  **Heat Map**.

The **Plot Sample Subcolumns in a Heat Map** dialog box appears (Figure 70).

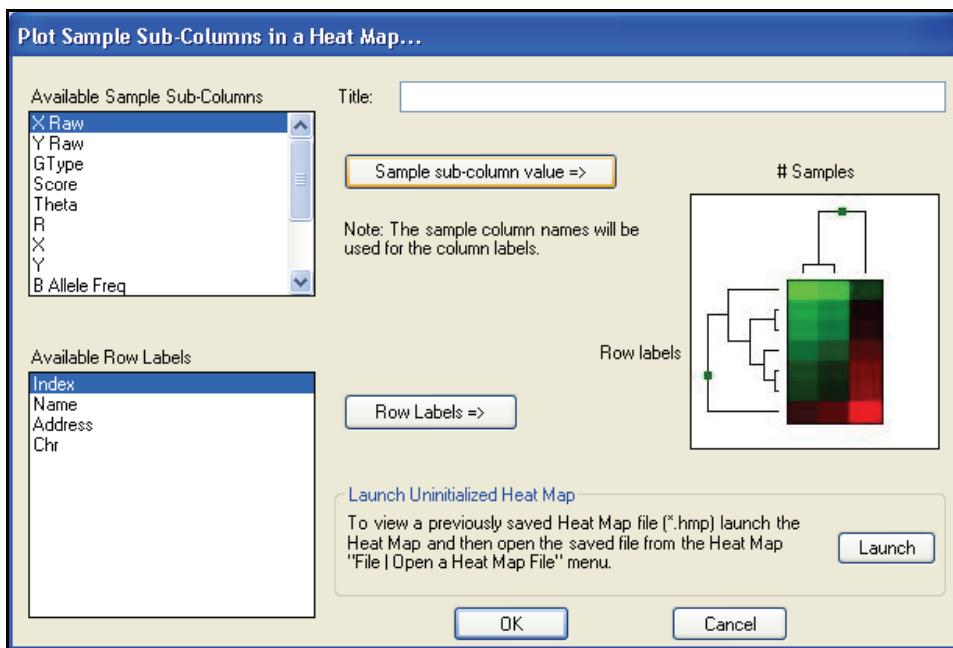


Figure 70 Plot Sample Subcolumns in a Heat Map Dialog Box

2. In the **Title** text field, enter a title for your heat map.
 3. In the **Available Sample Subcolumns** listbox, select the data series you wish to map in a heat map.
 4. Click **Sample Subcolumn Value**.
The data series name you select appears at the top of the heat map image to the right.
 5. In the **Available Row Labels** listbox, select a column that you want to use as the row labels of the heat map.
 6. Click **Row Labels**.
 7. Click **OK**.
- The heat map is generated with the row labels and data series you selected (Figure 71).

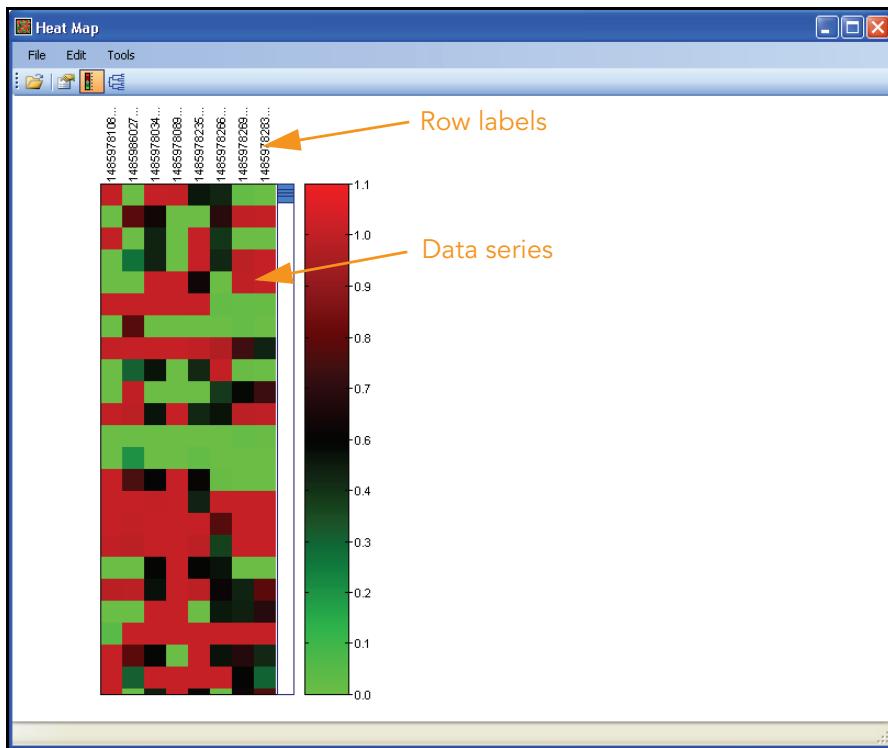


Figure 71 Heat Map with Row Labels and Data Series

Clustering Heat Map Data



WARNING

Clustering by rows is very process-intensive and can take an extremely long time. You are advised to filter the number of rows to a few thousand or fewer before clustering by row.

To cluster heat map data:

1. In the **Heat Map** window, click  **Cluster Heat Map Rows and/or Columns**.

The **Cluster Options** dialog box appears (Figure 72),.

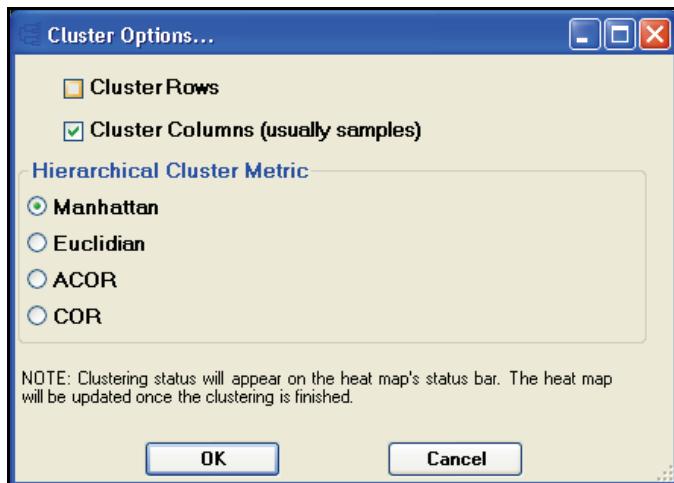


Figure 72 Cluster Options Dialog Box

2. In the **Cluster Options** dialog box, select one or both of the following:
 - **Cluster Rows**
 - **Cluster Columns (Usually Samples)**
3. Select one of the following **Hierarchical Cluster Metric** options:
 - **Manhattan**—Computes the distance between two points if a grid-like path is followed.
 - **Euclidian**—Computes the shortest distance between two points.

- **ACOR** (Absolute Correlation)—Computes the Pearson correlation using a $1 - |r|$ distance measure.
- **COR** (Correlation)—Computes the Pearson correlation using a $1 - r$ distance measure.

**NOTE**

Generally, Illumina recommends using multiple clustering methods to validate results. Groupings with a true biological basis will usually replicate regardless of the algorithm used.

4. Click OK.

The status bar at the bottom of the window displays the progress of the cluster analysis.

When the data is finished clustering, the heat map automatically displays the hierarchical clusters (Figure 75).

**Similarities
and Distances**

There are several ways to compute the similarity of two series of numbers. The most commonly used similarity metric is the Pearson correlation. The Pearson correlation coefficient between any two series of numbers $X = \{X_1, X_2, \dots, X_N\}$ and $Y = \{Y_1, Y_2, \dots, Y_N\}$ is defined as:

$$r = \frac{1}{N} \sum i = 1, N \left(\frac{X_i - \bar{X}}{\sigma_X} \right) \left(\frac{Y_i - \bar{Y}}{\sigma_Y} \right)$$

Distance is then defined as $1 - r$ for Correlation and $1 - |r|$ for Absolute Correlation. BeadStudio also uses Manhattan $(\Sigma |X_1 - Y_1|)$ and squared Euclidian $(\Sigma (X_1 - Y_1)^2)$ distances.

BeadStudio presents the clustering information in the form of a dendrogram, a tree-like structure whose branches correspond to rows and/or columns of the table. The distance on the X-axis establishes the similarity relationships among the genes or samples. For example, if the dendrogram plots the similarity of samples based on gene expression, samples C and D are very similar to each other, less similar to B, and even less similar to A (Figure 73).

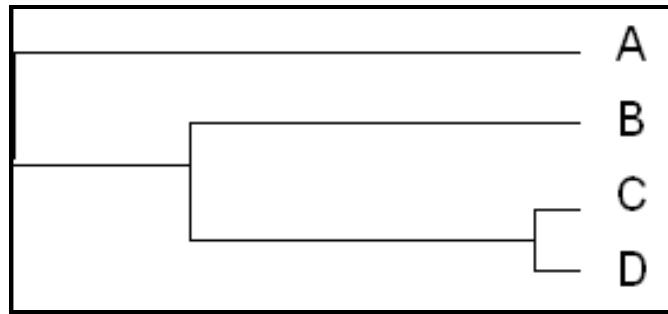


Figure 73 Dendrogram, Similarity Example

After clustering, nodes are reordered starting near the top to ensure that node "ar" is closer to "B" than node "al", and node "bl" is closer to "A" than node "br" (Figure 74).

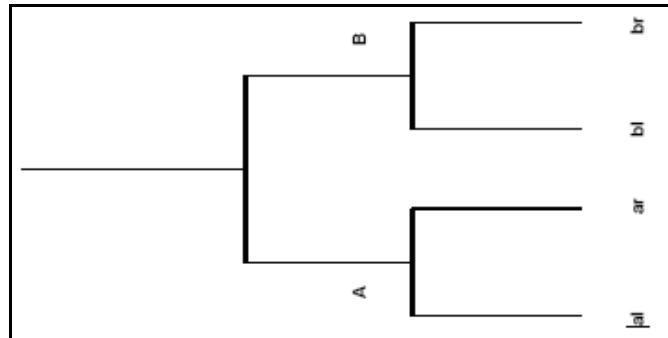


Figure 74 Dendrogram, Showing Nodes

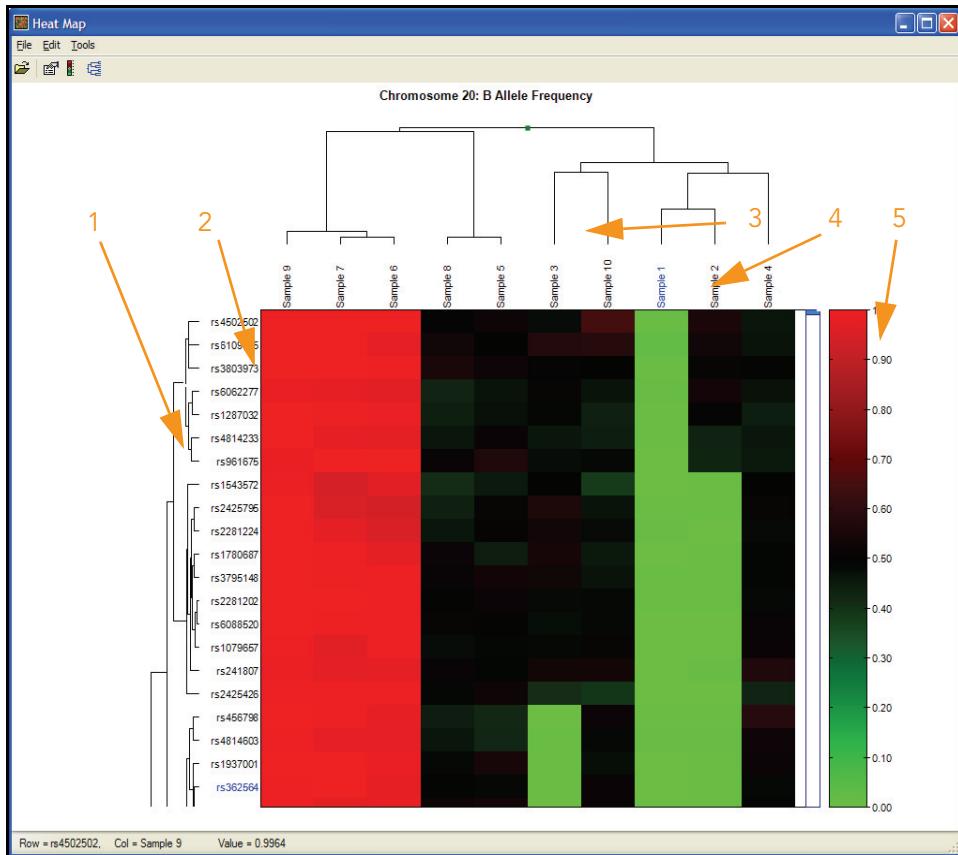


Figure 75 Heat Map, Display and Scrolling Properties

Table 4 Heat Map Elements

| Number | Element |
|--------|---------------------|
| 1 | Row tree cluster |
| 2 | Row label area |
| 3 | Column tree cluster |
| 4 | Column label area |
| 5 | Heat map scroll bar |

Resizing the Row Tree Cluster

The row tree cluster is shown in Figure 75, #1.

To resize the row tree cluster:

1. Press and hold the **Shift** key.
2. Click and hold the left mouse button anywhere in the row tree cluster area.
3. Move your mouse slowly left or right to resize the row tree cluster area.

Resizing Row Labels

The row label area is shown in Figure 75, #2.

To change the width of the row label area:

1. Press and hold the **Shift** key.
2. Click and hold the left mouse button anywhere in the row label area.
3. Move your mouse slowly left or right to resize the row label area.
4. To change the height of the row label area, do one of the following:

Using the mouse wheel:

- a. Position the cursor over the row label area.
- b. Move the mouse wheel up or down.

Using the mouse button:

- a. Position the cursor over the row label area.
- b. Press the left mouse button and drag up or down to resize.

Resizing the Column Tree Cluster

The column tree cluster is shown in Figure 75, #3.

To resize the column tree cluster:

1. Press and hold the **Shift** key.
2. Click and hold the left mouse button anywhere in the row tree cluster area.
3. Move your mouse slowly up or down to resize the column tree cluster area.

Resizing Column Labels

The column label area is shown in Figure 75, #4.

To change the height of column labels:

1. Press and hold the **Shift** key.
2. Click and hold the left mouse button anywhere in the column label area.
3. Move your mouse slowly up or down to resize the column label area.

To change the width of column labels, do one of the following:

- ▶ Position the cursor over the column label area, and move the mouse wheel up or down.
- ▶ Position the cursor over the column label area, press the left mouse button, and drag up or down to resize.

Scrolling the Heat Map Area

The heat map scroll bar is shown in Figure 75, #5.

To scroll the heat map area, do one of the following:

- ▶ Position the cursor over the heat map, and use the mouse wheel to scroll up or down.
- ▶ Click the scroll bar and hold, then drag the scroll bar up or down.

Opening a Heat Map File

1. Select **File | Open Heat Map File**.

The **Open Heat Map Data File** dialog box appears.

2. Browse for and select a previously-saved heat map file.

3. Click **Open**.

The heat map file appears.

Saving a Heat Map

1. Select **File | Save As**.

The **Save Heat Map to File** dialog box appears.

2. Browse to the location you want to save your heat map.

3. Type a name for your heat map in the **File Name** text field.

4. Click **Save**.

Your heat map is saved in the location you specified.

Exiting the Heat Map Window

To exit the heat map window:

1. Select **File | Exit**.

If you have clustered your data during this session, you are prompted to save the clustered heat map to a file, so that you do not have to recluster again when you use this heat map in future sessions.

2. Select **File | Exit**.

The **Unsaved Data** dialog box appears.

3. Click **Yes** to save the clustered heat map to a file.



NOTE

Because the clustering process can be time-intensive for large data sets, saving your clustered heat map will save you time later.

Editing Heat Map Properties

You can change the visual properties of the heat map to suit your preferences. You can the following elements of your heat map:

- ▶ Title
- ▶ Legend
- ▶ Row/column properties
- ▶ Scroll bar properties

Title

To change the heat map title:

1. Do one of the following:

- ▶ Select **Edit | Properties**.
- ▶ Click  **Edit Heat Map Properties**.

The **Heat Map Properties** dialog box appears (Figure 76).

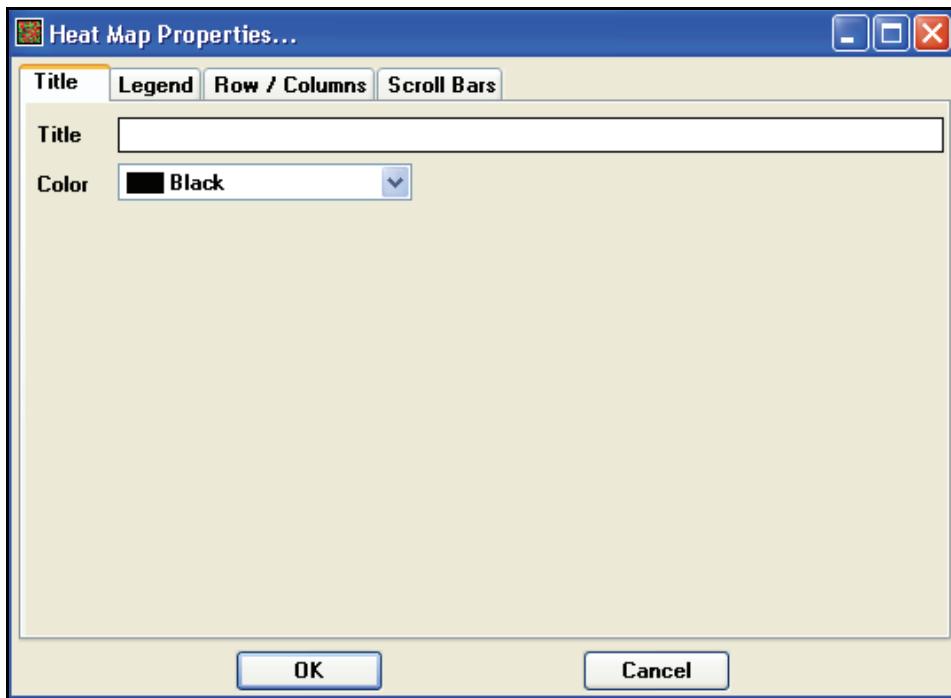


Figure 76 Heat Map Properties Dialog Box, Title Tab

2. In the **Title** tab, enter a title for your heat map in the **Title** text field.
3. **[Optional]** Choose a color for the heat map title by selecting a color from the **Color** pulldown menu.
4. Click **OK**.

The heat map appears with the new title centered above it.

Legend

Changing the Scale of the Legend Axis

To change the scale of the legend axis:

1. Click the **Legend** tab (Figure 77).

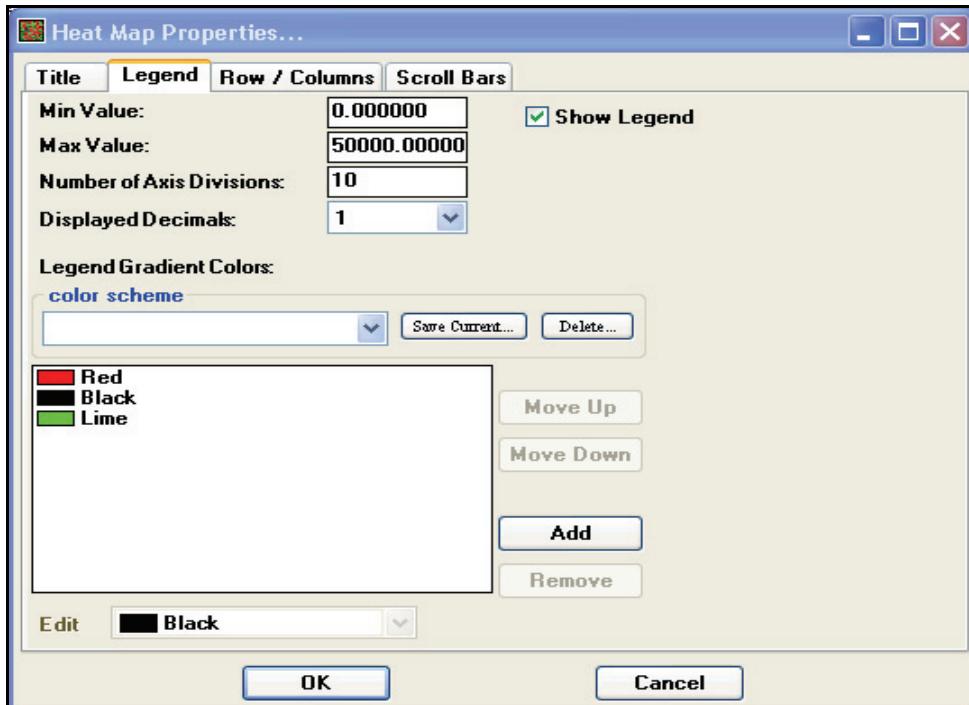


Figure 77 Heat Map Properties Dialog Box, Legend Tab

2. Enter a **Min Value**, **Max Value**, **Number of Axis Divisions**, and **Displayed Decimals**.

Showing/Hiding the Legend

To show the legend with the heat map, select the **Show Legend** checkbox (Figure 77). To hide the legend, clear the **Show Legend** checkbox.

Changing the Legend Gradient Colors

The default legend gradient colors are Red, Black, and Lime. To change the legend gradient colors, do any of the following:

To choose a color scheme:

1. Make a selection using the **Color Scheme** pulldown menu (Figure 77).

2. Click **OK**.

The heat map displays with the color scheme you selected.

To change the assignment of individual colors:

1. Click a color in the **Legend Gradient Colors** area.
 2. Click **Move Up** or **Move Down** to reassign the color.
 3. Click **OK**.
4. The heat map displays with the color(s) you selected.

To add a color to the heat map:

1. Click **Add**.
The color dialog box appears.
2. Choose a color from the dropdown menu.
3. Click **OK**.
The heat map displays with the color you selected.

To remove a color from the heat map:

1. Click a color in the **Legend Gradient Colors** area.
2. Click **Remove**.
The heat map displays without the color you removed.

Row/Column Properties

You can change heat map row and column properties (Figure 78) to suit your preferences.

Showing/Hiding Labels

- To show row labels, select the **Show Labels** checkbox in the **Rows** area.

- ▶ To hide row labels, clear the **Show Labels** checkbox in the **Rows** area.
- ▶ To show column labels, select the **Show Labels** checkbox in the **Columns** area.
- ▶ To hide column labels, clear the **Show Labels** checkbox in the **Columns** area.

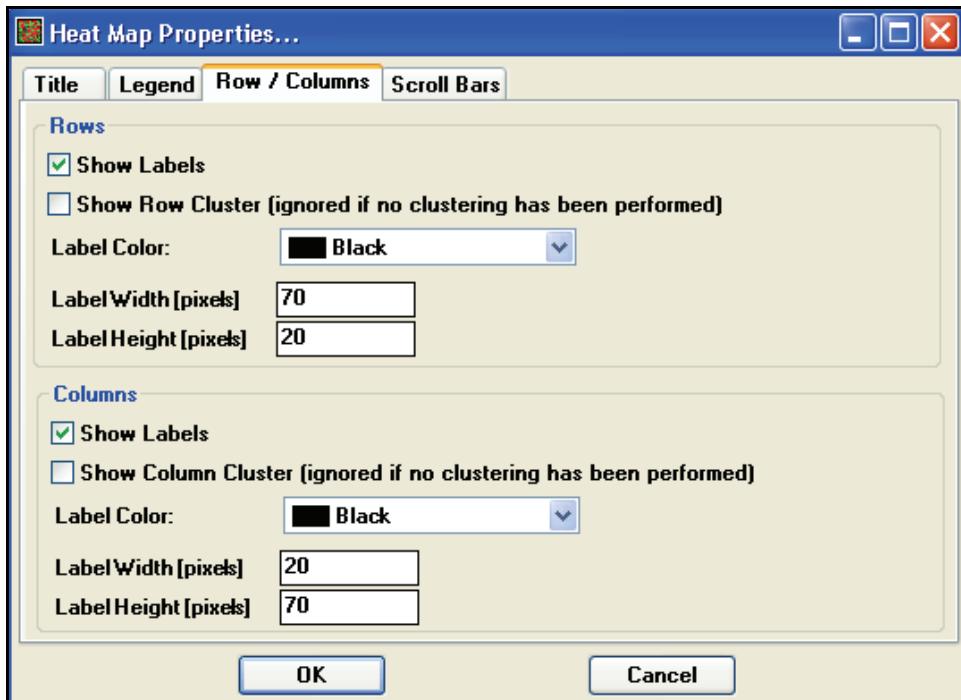


Figure 78 Heat Map Properties Dialog Box, Row / Columns Tab

Showing/Hiding Clusters

- ▶ To show the row cluster, select the **Show Row Cluster** checkbox.
- ▶ To hide the row cluster, clear the **Show Row Cluster** checkbox.
- ▶ To show the column cluster, select the **Show Column Cluster** checkbox.
- ▶ To hide the column cluster, clear the **Show Column Cluster** checkbox.

Changing the Label Color

To change the label color for rows or columns:

- ▶ Select a new color from the corresponding **Label Color** dropdown menu.

Changing the Label Height/Width

To change the label height or width for rows or columns:

- ▶ Enter a new height in the corresponding **Label Height** text field.
- ▶ Enter a new width in the corresponding **Label Width** text field.

Scroll Bar Properties

You can change the properties of the scroll bar to suit your preferences.

To change scroll bar properties (Figure 79), select new colors using the **Scroll Area Color**, **Scroll Bar Color**, and **Scroll Border Color** dropdown menus.

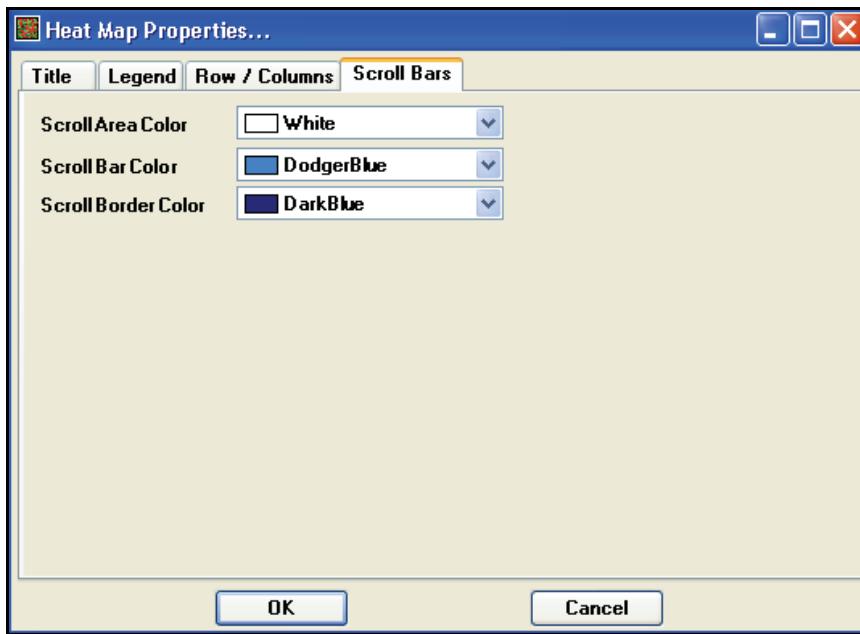


Figure 79 Heat Map Properties Dialog Box, Scroll Bars Tab

Creating a Presentation Image

To create a presentation image:

1. Select **Tools | Generate Presentation Image**.

The **Presentation Image Setup** dialog box appears (Figure 80).

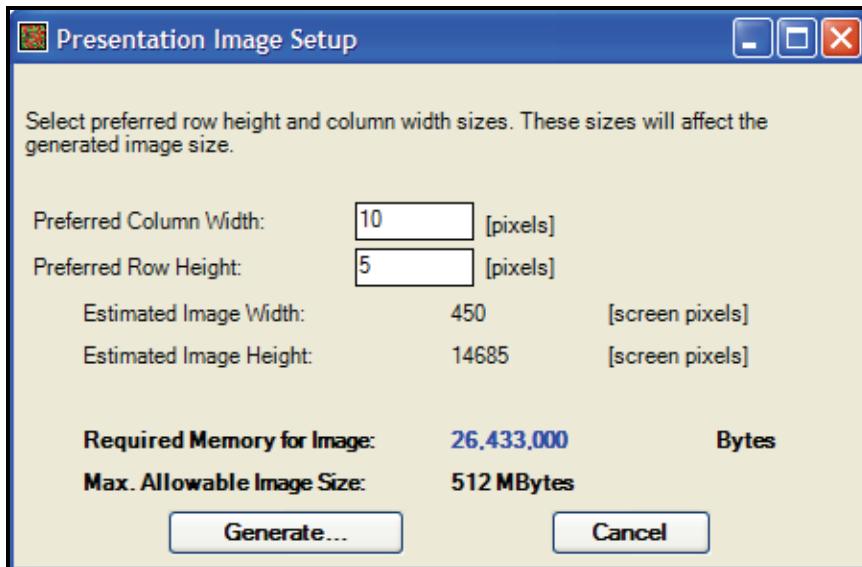


Figure 80 Presentation Image Setup Dialog Box

2. Enter a **Preferred Column Width** and **Preferred Row Height** in pixels.
3. Look at the **Required Memory for Image** and decide whether you want an image file of this size.
If the image file size is too large, adjust the number of pixels for the row height and column width.
4. Click **Generate**.
The **Image Generation Progress** status bar displays the status of the image generation.
5. The **Presentation View** window appears with a static heat map image that can be viewed in its entirety and saved to a file (Figure 81).

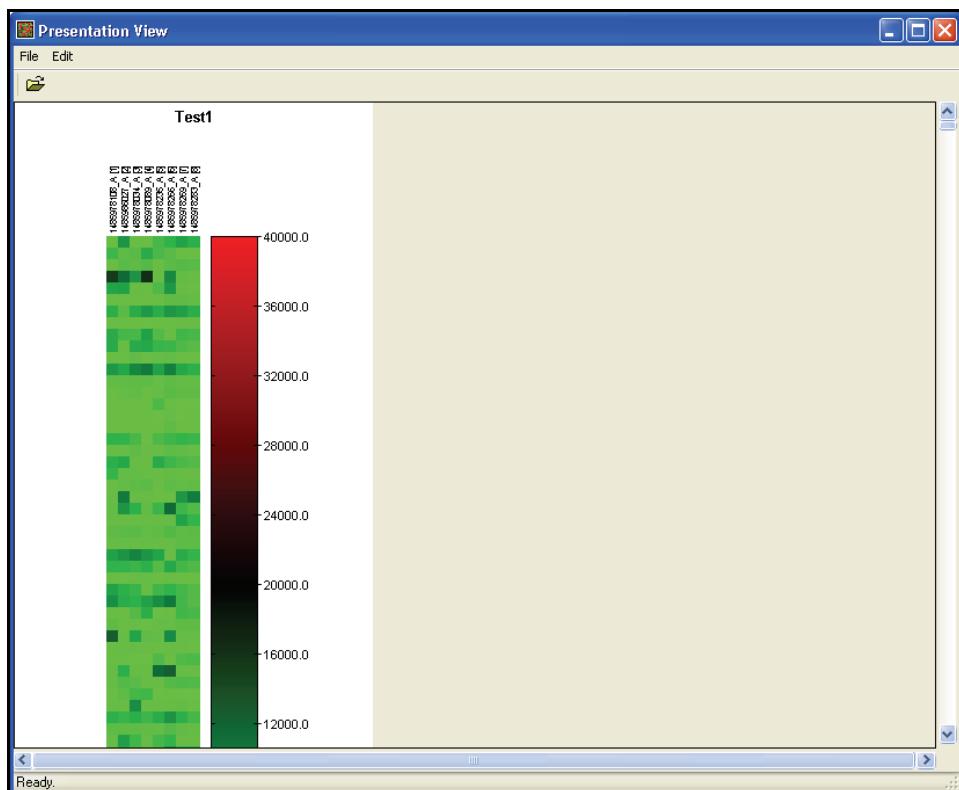


Figure 81 Heat Map, Presentation View

Chapter 5

Visualization Tools

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Introduction

This chapter describes how to use BeadStudio visualization tools.

The Illumina Genome Viewer (IGV) is a tool that allows you to visualize data on a genome-wide scale. You can access additional analysis tools directly through the IGV, including:

- ▶ Illumina Chromosome Browser (ICB)
- ▶ Illumina Sequence Viewer (ISV)
- ▶ Chromosome Zoom Plot
- ▶ Chromosome Heat Map
- ▶ Bookmark Viewer

Using the Illumina Genome Viewer

To launch the IGV:

- ▶ Select **Analysis | Show Genome Viewer** (Figure 82).

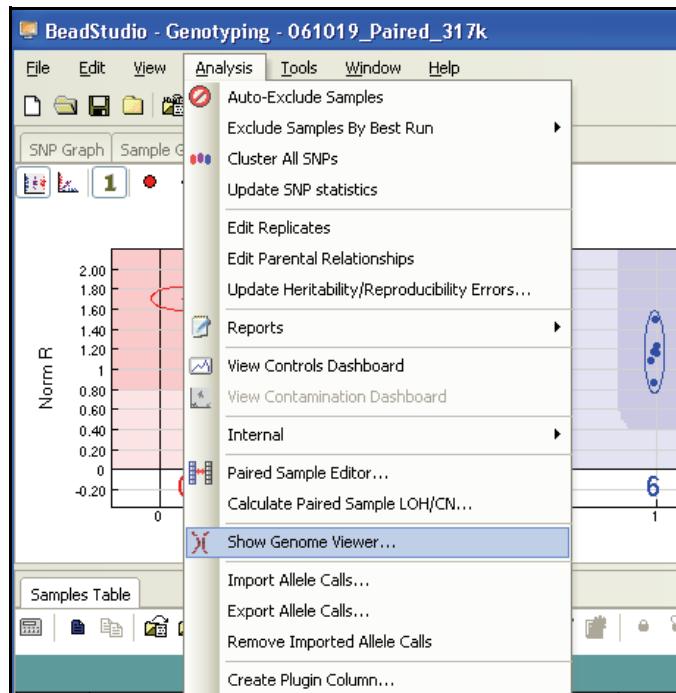


Figure 82 Launching the IGV

The IGV main window appears (Figure 83).

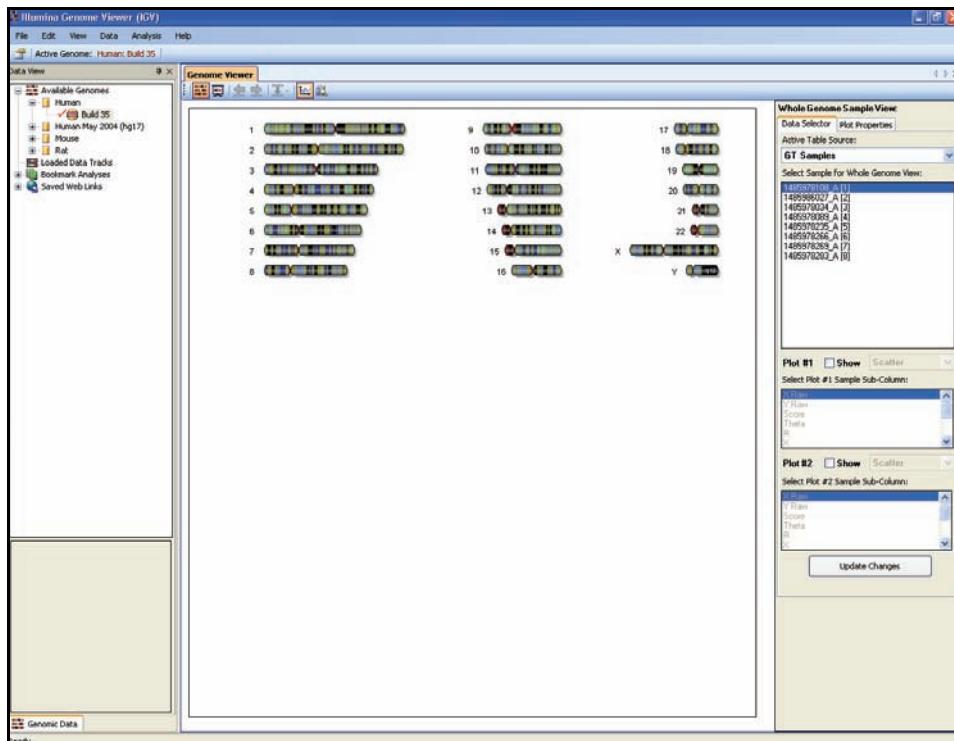


Figure 83 IGV Main Window, Whole Genome View

The IGV has two viewing modes:

- ▶ Whole Genome View Mode
- ▶ Chromosome Slideshow Mode

You can display a data plot in the IGV in either of these modes.

In Whole Genome View mode, the IGV can display up to two plots at a time for each chromosome. When you open the IGV for the first time, no data is displayed.

To display a plot in Whole Genome View mode:

1. In the IGV, click  **Whole Genome View Mode**.
2. In the **Whole Genome Sample View** area, click the **Data Selector** tab.

3. Select the checkbox for Plot #1 and/or Plot #2.
4. In the **Select Plot #1 Sample Subcolumn** area, select the data parameter you want to plot.
5. Click **Update Changes**.



For high-density genotyping BeadChips, it may take some time to process the data and display the plots in Whole Genome View Mode.

The Whole Genome View Mode data plot appears (Figure 84).

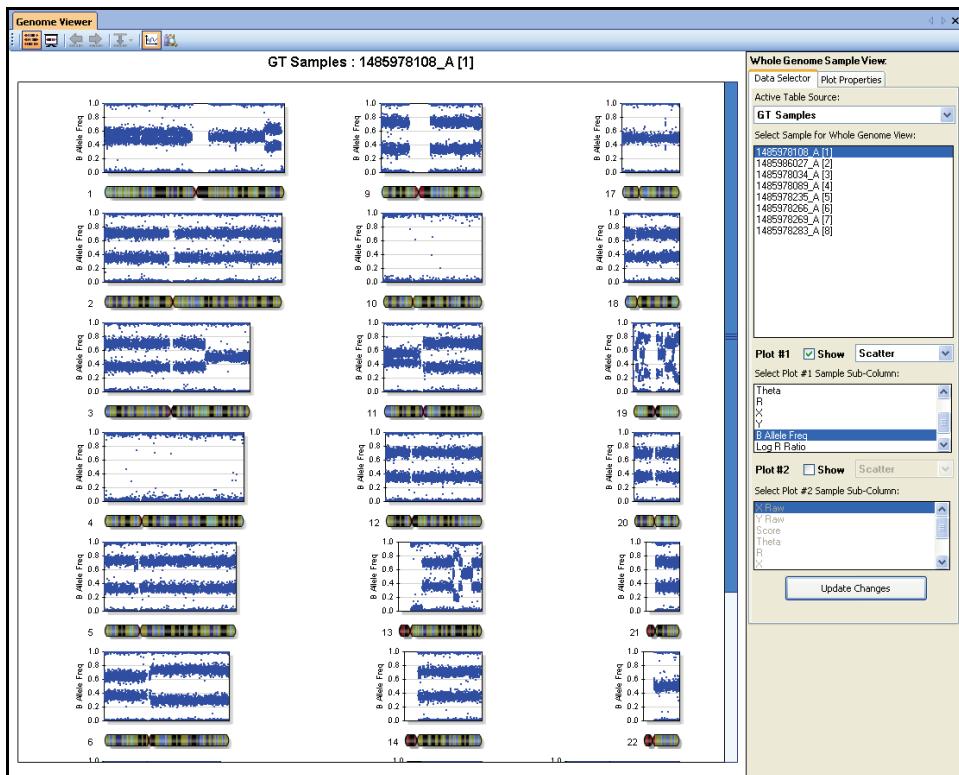


Figure 84 Data Plot Shown in Whole Genome View Mode

In Chromosome Slideshow Mode, the IGV can display up to four plots at a time, of a single data series each, over any chromosome.

To display a data plot in Chromosome Slideshow Mode:

1. In the IGV, click  **Chromosome Slide Show Mode**.
2. In the **Plot Selector** panel, select the **Plot Number** and **Show** checkboxes for the plots you want to display.
The plots appear in the IGV main window.
3. In the **Select Sample** area, choose the samples for which you want to display data.
4. **[Optional] Select the Set All Plots Shown to Selected Subcolumn** checkbox.
5. In the **Select Subcolumn** area, choose the sample subcolumn you want to plot.

The plot appears with the data parameters you have chosen (Figure 85).

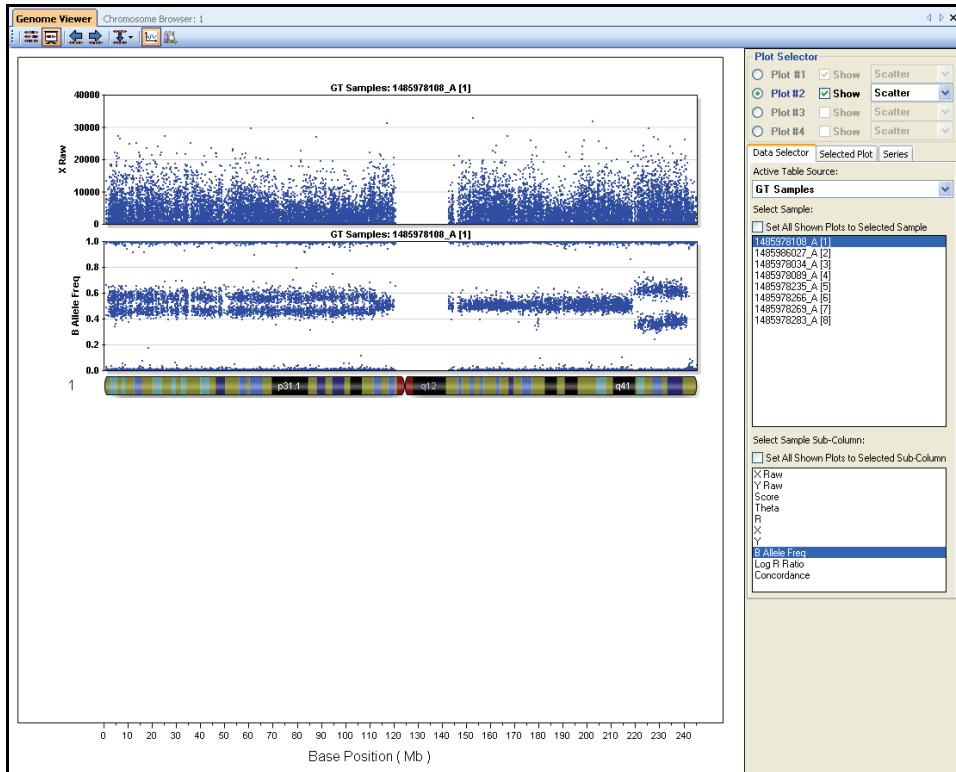


Figure 85 Data Plot Shown in Chromosome Slide Show Mode

In addition to using the IGV, you can browse your data at the chromosomal level using the ICB and/or the Zoom Plot.

To launch the ICB:

- Double-click a chromosome in the IGV (Figure 87, #1).

The ICB appears. However, when it first opens, there is no data visible. To proceed, you will need to display a data plot.

To display a data plot in the ICB:

1. Click the **Plot Settings** tab.
2. Select the **Display Plot** checkbox.

The data plot appears above the chromosome in the ICB.

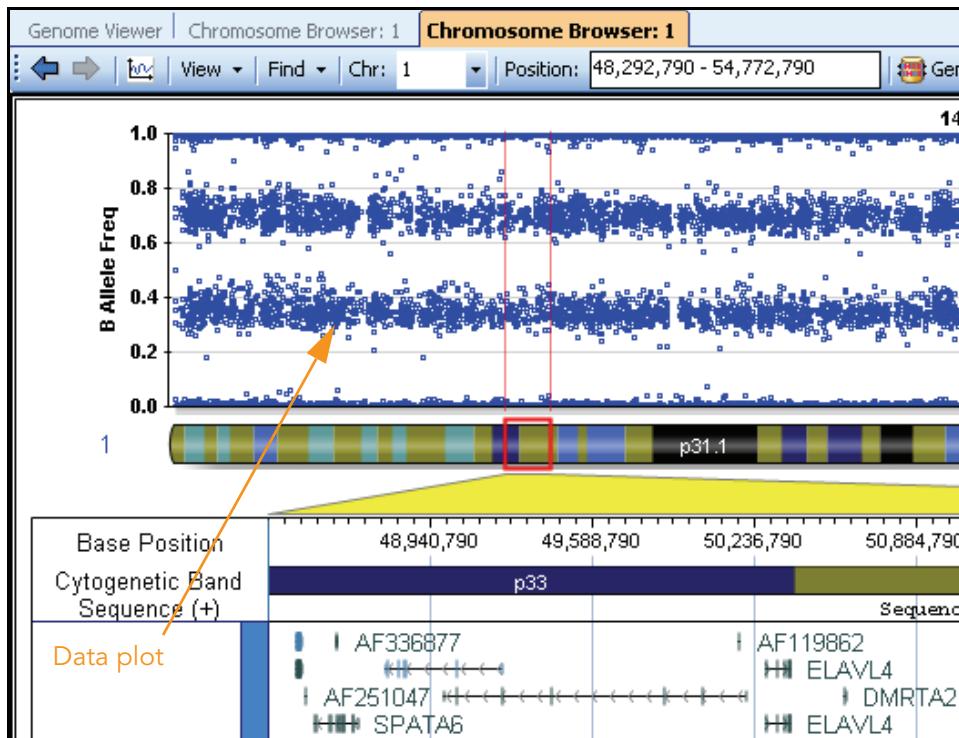


Figure 86 Data Plot Displayed Above the Chromosome

For more information about the ICB, see *Using the Illumina Chromosome Browser* on page 102.

The zoom plot allows you to zoom into a particular chromosomal region to view data in finer detail relative to known chromosomal annotation.

To launch the zoom plot:

1. Return to the IGV main window.
2. Double-click any plot in the IGV (Figure 87).

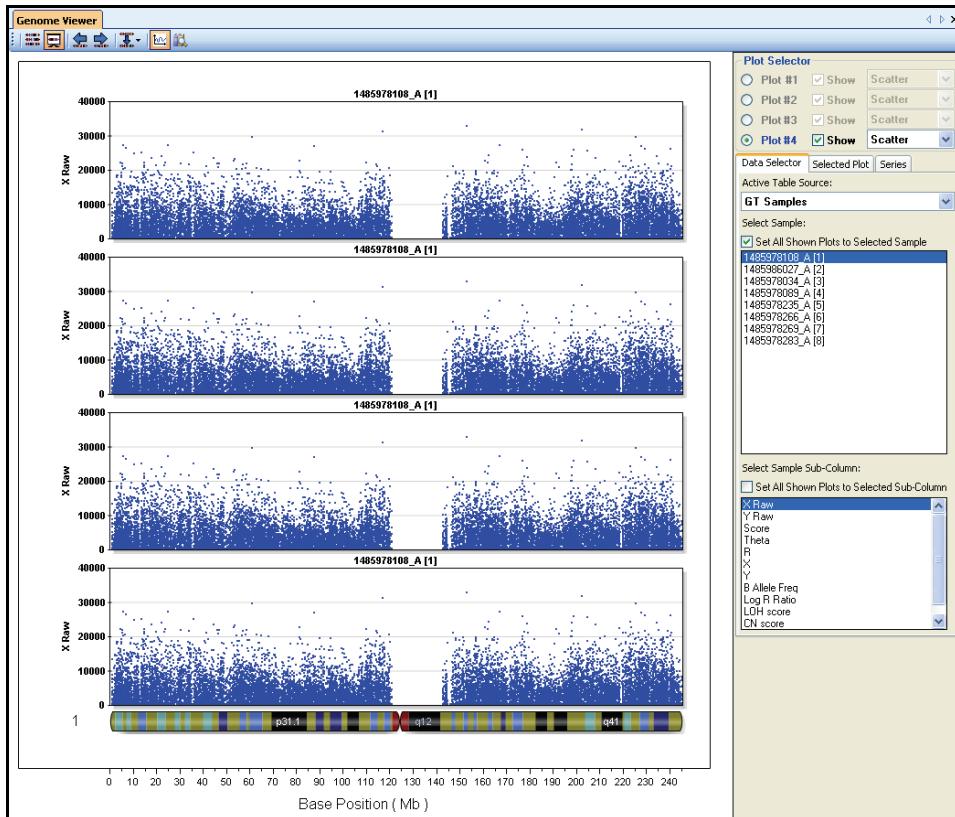


Figure 87 Data Visualization at the Chromosomal Level

The plot you clicked is launched as a zoom plot (Figure 88).

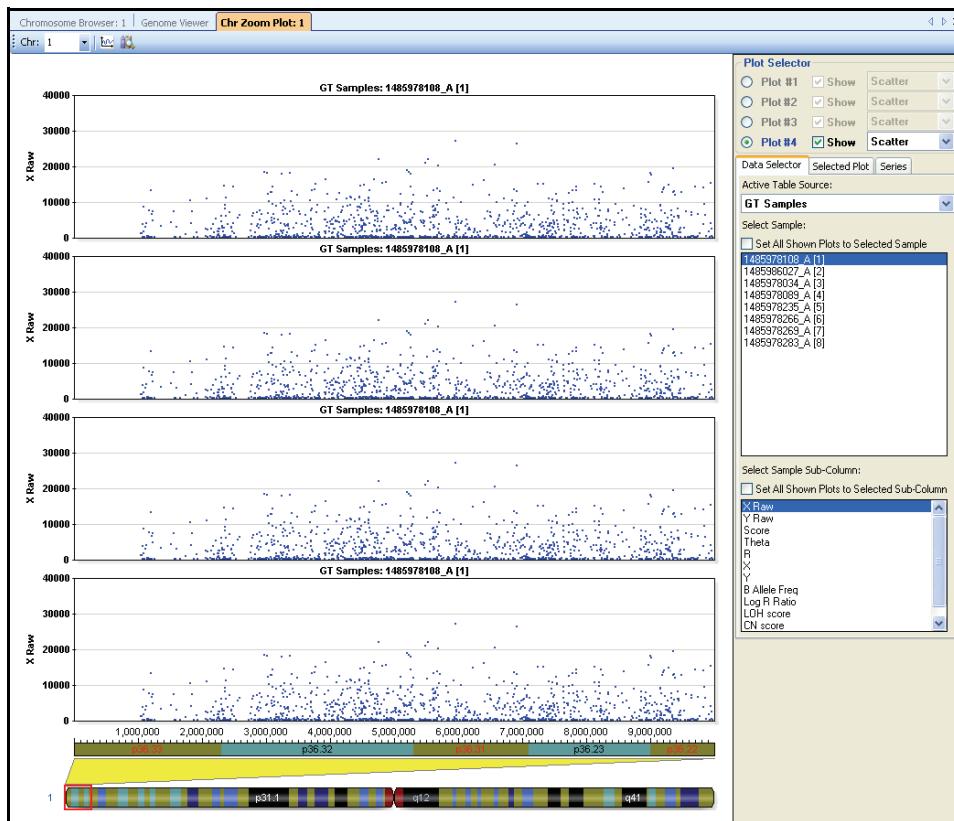


Figure 88 Zoom Plot

Getting Data Files

The IGV is installed with genomes from the UCSC GoldenPath database.

If you want to analyze genomes or builds other than those installed with the IGV, you can download additional genome annotation files by following these steps:

1. Go to <http://hgdownload.cse.ucsc.edu/downloads.html>.
2. Click the species of the genome you want to download.
3. In your BeadStudio-installed directory structure, navigate to the folder that contains your Genome Viewer genome files.

The default location is:

C:\Program Files\BeadStudio\Modules\GenomeViewer\Genomes.

4. Create a new folder under the path in Step 3. The folder name should be descriptive of the genome and build; for example, "Human 2004 (hg17)" (Figure 89).

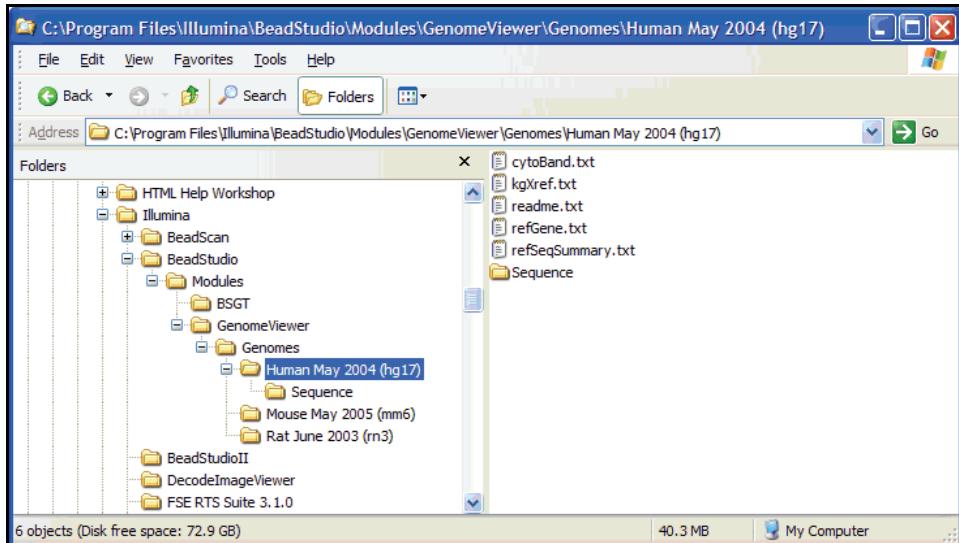


Figure 89 IGV, Genome Directory Structure

5. In your internet browser, click **Annotation Database**.



Annotation Database should be one of the top few clickable links in each list.

A text-based web page appears.

6. Scroll down, beyond the text, to the download list.
7. Download the following required files from the download list and save them to the folder you created in Step 3.
 - cytoband.txt
 - refGene.txt
 - kgXref.txt

8. [Optional] Download the following files and save them in the same location:
 - refSeqSummary.txt
 - FASTA formatted sequence files in Sequence sub-folder (Figure 90)

If you want to load a FASTA format sequence file (e.g., chr1.fa.zip) in your web browser, click the **Data set by chromosome** link to go to that page. Download and unzip the file to a subfolder named "Sequence" (Figure 90).

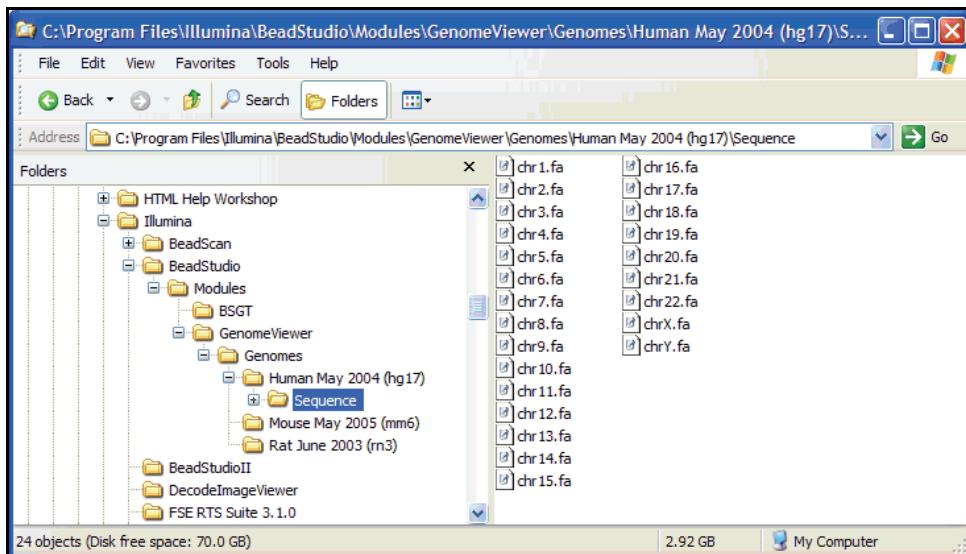


Figure 90 IGV, Optional Genome Sequence File Sub-Folder

9. Unzip the files you downloaded in Step 7.
10. To make the newly downloaded genome visible to the IGV, exit the BeadStudio application and restart it.
11. To view a newly-added genome in the ICB, go to **Edit | Preferences**.
The **Preferences** dialog box appears.
12. Select the genome you want to view in the **ChangeActiveGenome** listbox.
13. Click **OK**.

The genome you selected appears in the IGV main window.

Working with the IGV Toolbar

This section includes a brief description of each IGV toolbar function. Figure 91 shows the IGV toolbar, and Table 5 lists the IGV toolbar buttons and their functions.



Figure 91 Illumina Genome Viewer Toolbar

Table 5 IGV Toolbar Buttons

| Button | Name | Function |
|--------|-------------------------------|--|
| | Whole Genome View Mode | Shows all genome chromosomes with all enabled plots shown above each chromosome. Whole Genome View Mode is the default IGV mode. |
| | Chromosome Slide Show Mode | Enables the slide show mode which allows the user to view from one to four chromosomes at the same time. |
| | View Previous Slide | Displays the previous chromosome slide. This button is enabled only when Chromosome Slide Show Mode is active. |
| | View Next Slide | Displays the next chromosome slide. This button is enabled only when Chromosome Slide Show Mode is active. |
| | Jump to a Specific Chromosome | Displays a menu that allows you to jump to a particular chromosome slide. |
| | Show Plot Selector Panel | Toggles between showing and hiding the Plot Selector panel. This button is only enabled if Chromosome Slide Show Mode is active. |
| | Show/Hide Bookmarks | Toggles between showing and hiding bookmark files you can select to display with the current plot(s). |

Working with Menus

Table 6 describes the IGV menu selections and their functions.

Table 6 Illumina Genome Viewer Menus

| Selection | Description |
|------------------------|---|
| File Menu | |
| Exit | Closes the IGV window. |
| Edit Menu | |
| Preferences | Displays a dialog box from which you can edit the genome, viewing area, and Giesma stain properties. |
| View Menu | |
| Chromosome Browser | Launches the ICB in the main window, on top of the IGV. |
| Chromosome Zoom Plot | Displays zoom plots for the currently-displayed chromosome. |
| Chromosome Heat Map | Displays a chromosome heat map for the current chromosome. |
| Sequence Viewer | Launches the ISV in the main window, on top of the IGV. |
| Bookmark Viewer | Displays bookmark files for the current chromosome. |
| Genome Viewer | Toggles the IGV on and off. |
| Workspace | Toggles the Data View and Plot Selector panel on and off. |
| Data Menu | |
| Add/Remove Data Tracks | Allows you to add or remove data tracks for use with the ICB |
| Analysis Menu | |
| Auto-Bookmark | Opens the Auto-Bookmark Analysis window, from which you can select Auto-Bookmark Analysis algorithms to apply, and save your selections with comments as a new analysis. |

Table 6 Illumina Genome Viewer Menus (continued)

| Selection | Description |
|-----------|---|
| Help Menu | |
| About | Displays copyright information for the IGV. |

Plotting Sample Columns When you launch the Illumina Genome Viewer from the **Analysis** menu, it has access to **Full Data Table** sample columns. The table samples and sample subcolumns appear in the **Plot Selector** panel to the right of the IGV main window. Figure 92 shows a close-up view of the **Plot Selector** panel.

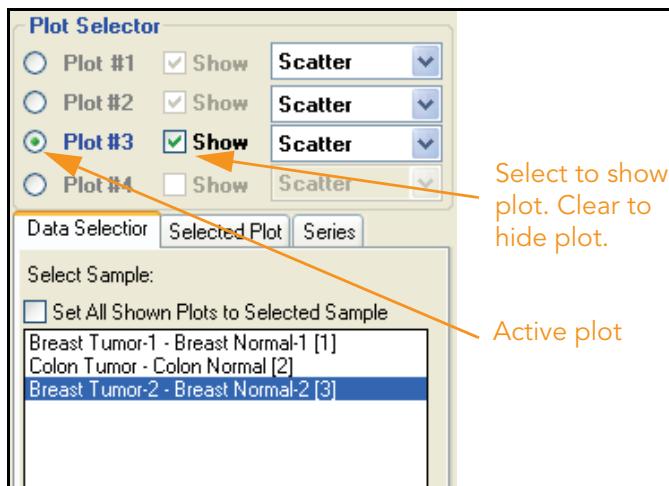


Figure 92 Plot Selector Panel

You can choose a plot to edit by selecting the **Plot 1** to **Plot 4** radio buttons. The **Show** checkboxes toggle the plot view.

To select the sample as the data source for all displayed plots, select **Set All Shown Plots to Selected Sample**. Use this feature to look at different subcolumns for the same sample, and to rapidly view the same subcolumns for different samples by changing the selected sample in the **Select Sample** listbox.

To plot the item in the **Select Sample Sub-Column** listbox in all visible plots, select **Set All Shown Plots to Selected Sub-Column**. Use this feature when you want to look at the same sample subcolumn for different samples on different plots.

Using the Illumina Chromosome Browser

The Illumina Chromosome Browser (ICB) allows you to explore data by chromosome or by gene. The following sections describe how to work with the ICB.

Launching the ICB

Launch the ICB by double-clicking on a chromosome in the Illumina Genome Viewer (Figure 93, #1).

Navigating the ICB

Figure 93 shows the ICB toolbar and menus.

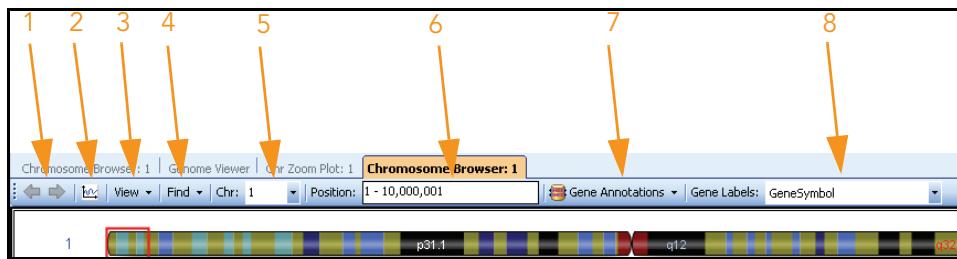


Figure 93 ICB Toolbar

Table 7 ICB Toolbar Elements

| Number | Element | Description |
|--------|-------------------------------------|--|
| 1 | Navigate Backward/ Navigate Forward | Behave much like Back/Forward in an internet browser; take you backward to and forward from previous views. |
| 2 | Toggle Plot Display | Displays a plot of data above the chromosome. The plot series data can be selected from samples in the Plot Settings tab at the bottom of the form. |

Table 7 ICB Toolbar Elements (continued)

| Number | Element | Description |
|--------|------------------|--|
| 3 | View | Allows you to select multiple views, including: <ul style="list-style-type: none"> • Dense Gene View • CpG Islands • Manifest Probes SNPs • Custom Tracks • Bookmark Viewer |
| 4 | Find | Allows you to select Find Genes or Find Probes . |
| 5 | Chr | Shows the current chromosome displayed for the current genome. You can choose a different chromosome to view from the dropdown menu. |
| 6 | Position | Shows the base pair range of the current view in physical coordinates. The current view is shown visually by the red rectangle drawn on the chromosome. You can enter a new range here. Press Enter to accept the newly entered base pair range. |
| 7 | Gene Annotations | Choose one of the following: <ul style="list-style-type: none"> • Edit—displays the Gene Annotation window, which allows you to import custom gene annotations to view in the ICB. • RefSeq—displays RefSeq information in the ICB in blue text. • Swiss-Prot/UniProt—displays Swiss-Prot/UniProt information in the ICB in black text. |
| 8 | Gene Labels | Toggles how the gene IDs are shown in the gene region. Genes can be viewed using the following identifiers: <ul style="list-style-type: none"> • KnownGeneID • mRNAID • Swiss-Prot Accession • Swiss-Prot Display ID • Gene Symbol • RefSeq ID • NCBI Protein Accession • None |

Using the ICB Context Menu In addition to menus and toolbar options, the ICB has a context menu with additional features that are useful for working with your data in the ICB.

Table 8 ICB Context Menu Options & Descriptions

| Menu Option | Description |
|-----------------|---|
| Zoom In | Allows you to see a finer resolution of the current view. |
| Zoom Out | Allows you to widen the current view. |
| Copy As | Allows you to copy the image in one of the following file formats: BMP, JPEG, TIFF, PNG, GIF. |
| Clear History | Clears the clipboard of information you have copied. |
| Add Bookmark | Displays the Add Bookmark window, from which you can add a bookmark to an existing bookmark list, or create a new bookmark list. |
| Delete Bookmark | Deletes a bookmark. |
| Bookmark Viewer | Displays the Bookmark Analyses window, from which you can do the following: <ul style="list-style-type: none"> • Import a bookmark analysis file • Save a selected bookmark analysis • Delete a selected bookmark analysis • Add, edit, delete, display, or hide bookmarks in a bookmark analysis file |

Changing the View Region To change the **View Region** (defined by the red rectangle), do one of the following:

1. Position the cursor inside the red rectangle.
 2. Click and hold the left mouse button and drag the red rectangle to the area you want to view.
or:
 1. Position the cursor inside the **Base Position** axis row.
 2. Click and hold the left mouse button and drag the **Base Position** axis, or scroll using the mouse wheel.
- This moves the **View Region** by small increments.

To move *and* resize the **View Region**, do any of the following:

- ▶ Double-click on a cytogenetic band on the displayed chromosome.
The **View Region** is fitted to the cytoband.
- ▶ Double-click a gene of interest.
This changes the **View Region** size to fit the gene.
- ▶ Double-click a SNP in the SNPs table row.
Annotation data for this SNP is displayed.

Viewing Gene Information

The gene you select appears in red in the ICB (Figure 94). Information about the selected gene is shown in the **Gene ID** and **Gene Details** tabs below the ICB main window.

To zoom the current view to fit the whole gene:

- ▶ Double-click a gene.
The **Gene ID** tab shows all available gene cross-reference names from various databases. A brief description and RefSeq Summary (if available) are also shown.

To open your default internet browser to the NCBI definition of that particular RefSeq gene:

- ▶ Click the RefSeq ID label (shown in blue in the Gene ID tabbed window of Figure 94).
The **Gene Details** tab shows all gene exons, gene strand type, transcription start/end positions, and coding region start/end positions for the currently selected gene.
- ▶ To cause the view to fit a selected exon:
Double-click any exon shown in the exon listbox.

Figure 94 shows the view zoomed in for exon #2 of the currently-selected gene. If sequence files are available in the appropriate genome folders (and if the sequence will fit into the current view) the positive strand genomic sequence is displayed.

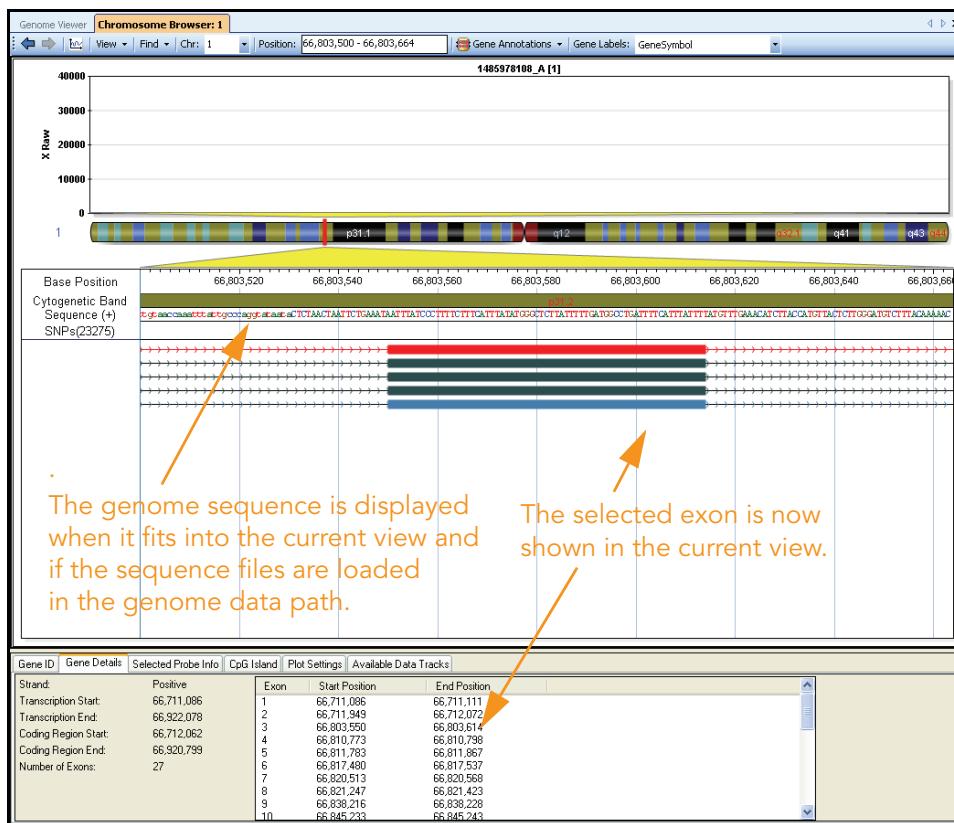


Figure 94 Viewing Gene Exons in Detail

Plotting Sample Columns

To plot sample columns from the **Full Data Table** in the ICB:

- Click  **Toggle Plot Display**.

The plot toggles to display mode and the **Plot Settings** tab at the bottom of the screen becomes active (Figure 95).

To hide the plot:

- Click  **Toggle Plot Display** to toggle off.

You can select the sample and sample column to be plotted. The plot automatically updates when an item is selected in either the **Select Sample** or **Select Sample Sub-Column** listbox. You can set the plot type, Y-axis properties, and series properties in the **Plot Settings** tab.

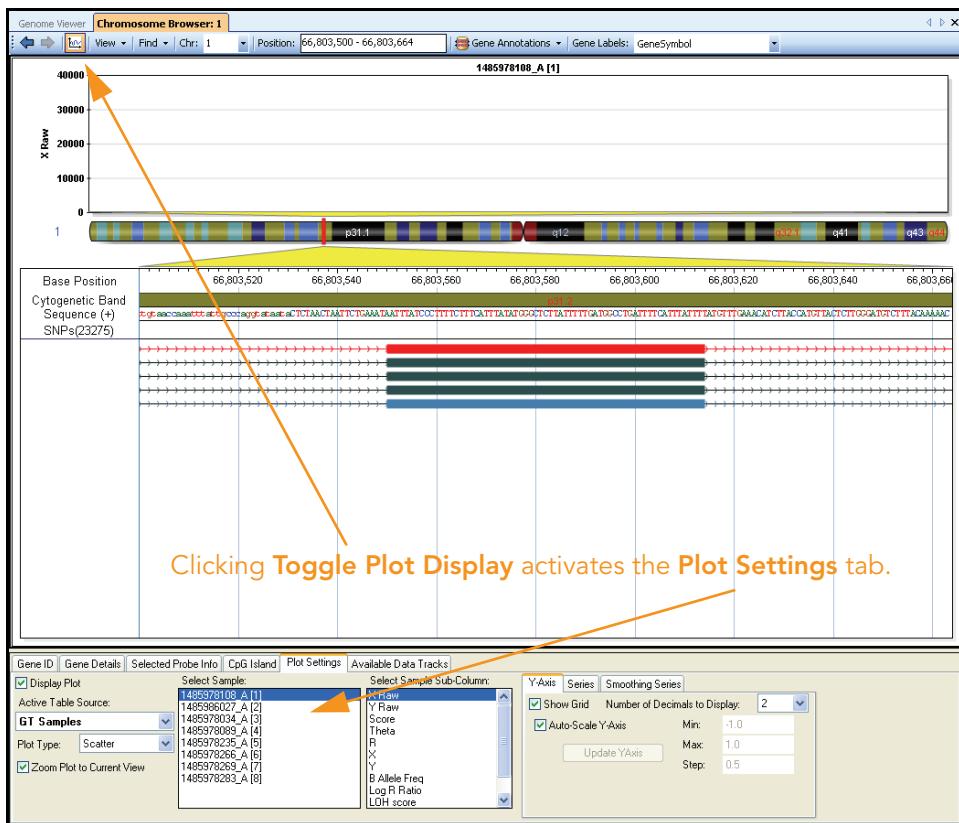


Figure 95 Plotting Sample Data in the ICB

Viewing Project Manifest SNPs

To display SNPs in the current project manifest:

- ▶ Select **View | Load Manifest | SNPs.**

The SNPs in the current manifest are displayed (Figure 96).



NOTE

If SNPs have been mapped to a different genome build than that loaded in the IGV, probes may not match the chromosome position displayed.

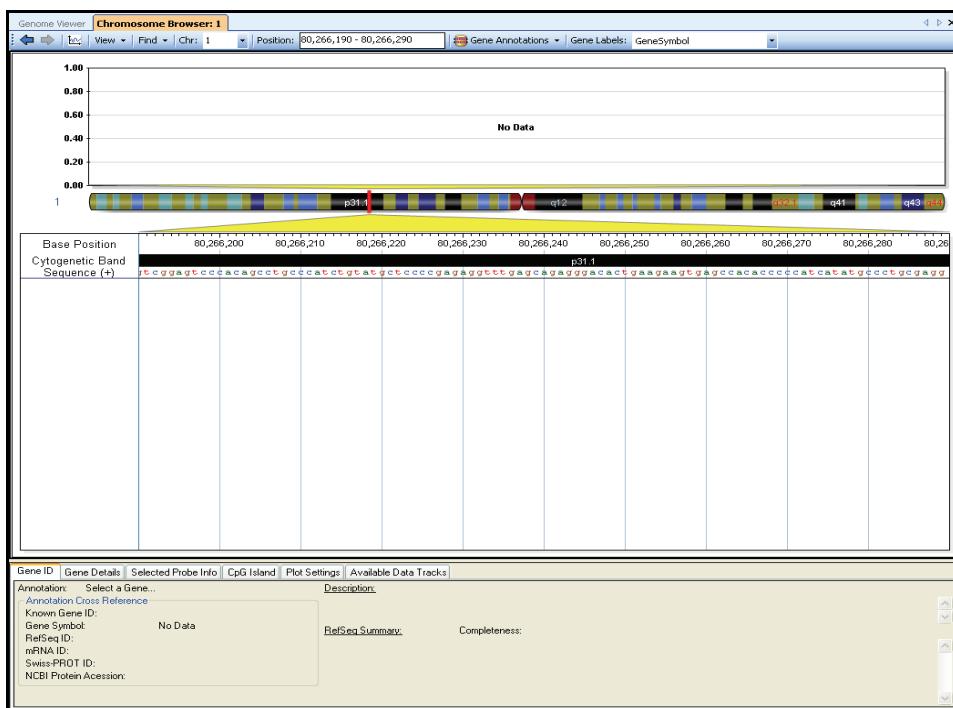


Figure 96 Loading Project Manifest SNPs into the ICB

SNPs are represented in the ICB by small, black, vertical bars.

- ▶ To activate the **SNP Info** tabbed window, click a SNP. The **SNP Info** tabbed window shows the RefSNP ID, allele, chromosome position, and top genomic sequence for the SNP.
- ▶ To zoom the view of the selected SNP, double-click a SNP.

Figure 97 shows manifest SNPs loaded after selecting **View | Load Manifest | SNPs.**

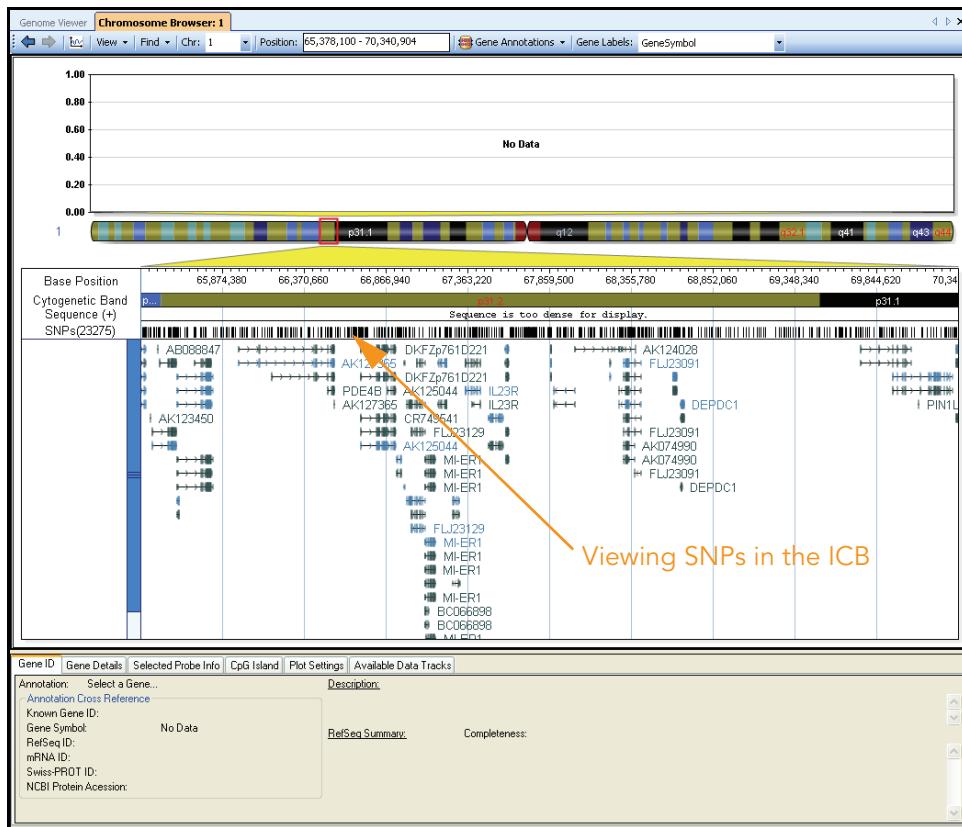


Figure 97 Viewing GT Manifest SNPs in the ICB

A vertical blue line overlays the SNP, showing any genes with which the SNP intersects (Figure 98).

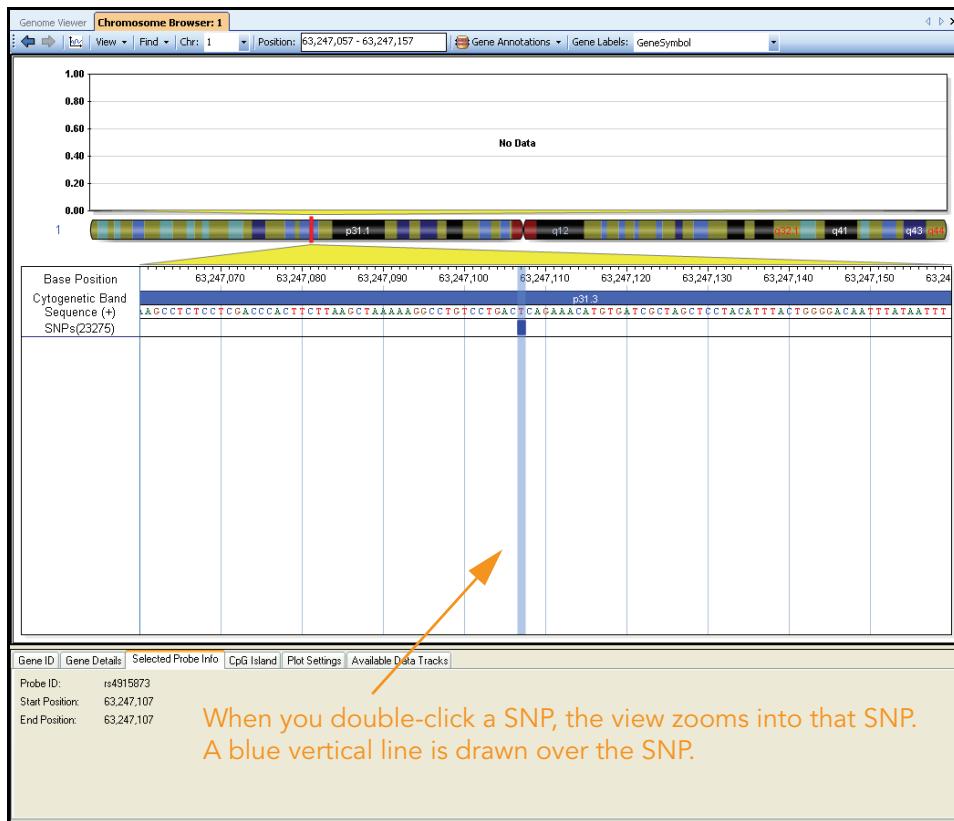


Figure 98 Zoomed View (Double-Clicking a SNP)

Loading Data Tracks

Data tracks are user-defined files containing information that can be plotted in the ICB. To learn how to load your own data tracks, use the standard linkage disequilibrium data tracks installed automatically with BeadStudio.

Loading Illumina-Provided Data Tracks

To load standard linkage disequilibrium data tracks provided with BeadStudio, do the following:

1. In the IGV, open the ICB.
2. Do one of the following:
 - a. In the **Data View** area, right-click **Loaded Data Tracks**.
 - b. Go to **Data | Add/Remove Data Tracks**.

The **Add/Remove Data Tracks** dialog box appears (Figure 99).

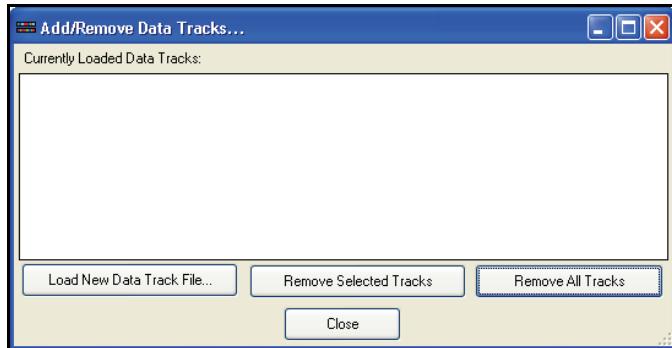


Figure 99 Add/Remove Data Tracks Dialog Box

3. Click **Load New Data Track File**.
4. Browse to the location of the standard linkage disequilibrium data tracks that were automatically installed with BeadStudio:
c:\Program Files\Illumina\BeadStudio
2.0\Modules\GenomeViewer\Data Tracks\Smith_LD
5. To select one of the available files to load, do one of the following:
 - a. Double-click one of the available files: 10kb.txt, 30kb.txt, 100kb.txt, 500kb.txt, or 1000kb.txt.
 - b. Select one of the available files and click **Open**.

The available data tracks appear in the **Add/Remove Data Tracks** dialog box.

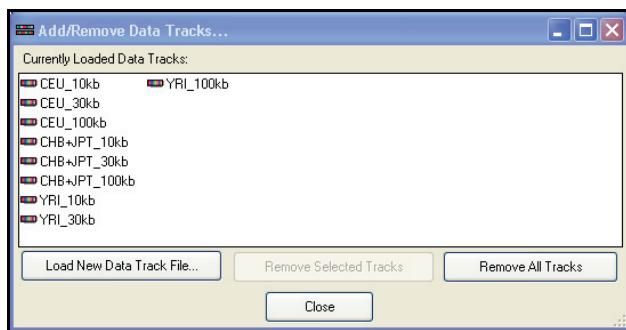


Figure 100 Selected Data Tracks

The loaded data tracks are also listed in the IGV, and graphically displayed in the ICB (Figure 101).

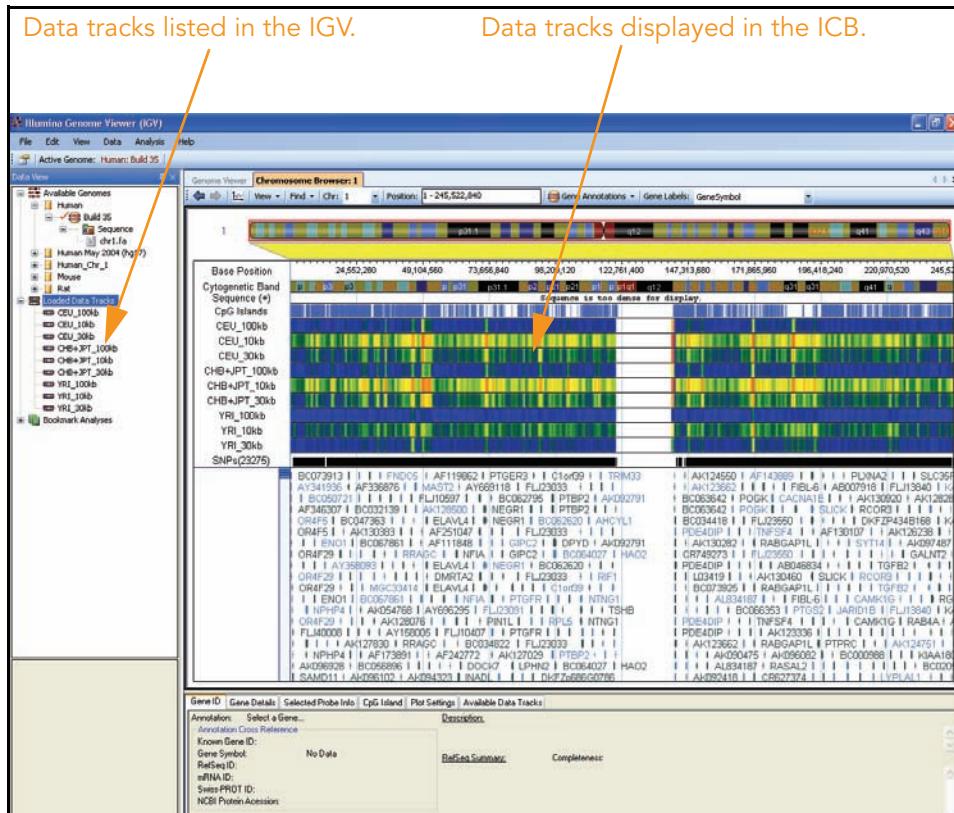


Figure 101 Data Tracks Displayed in the Illumina Chromosome Browser

- Click **Close** to close the **Add/Remove Data Tracks** dialog box.

Creating and Loading Your Own Data Tracks

To create your own data tracks to view in the ICB, do the following:

- Create a new tab-delimited text file using Excel or another application.
- To create a data track file, add data to the file using the following guidelines:

- All fields must be tab-delimited.
- The first line of the file must contain the column labels. The filename of the data track will be displayed as the label in the IGV.
- Fields that contain “NA” or “NaN” will be treated as “not a number” by the system. This is not case sensitive. “na” or “NAN” will be treated in the same manner.

Table 9 is an example of a user-created data track file. In this example, three data tracks are shown (CEU_10kb, JC_10kb, and YRI_10kb). However, you can create a file with more data tracks.

Table 9 Sample User-Created Data Track File

| chrom | chromStart | chromEnd | CEU_10kb | JC_10kb | YRI_10kb |
|-------|------------|----------|----------|---------|----------|
| chr1 | 900000 | 1000000 | 0.247 | 0.462 | 0.125 |
| chr1 | 950000 | 1050000 | 0.197 | 0.271 | 0.076 |
| chr1 | 1000000 | 1100000 | 0.202 | 0.247 | 0.09 |
| chr1 | 1500000 | 1150000 | 0.282 | 0.381 | 0.113 |
| ... | | | | | |
| chrX | 56850000 | 56950000 | 0.95 | 0.988 | 0.971 |
| chrX | 56900000 | 57000000 | 0.926 | 0.984 | 0.965 |
| chrX | 56950000 | 57050000 | 0.91 | NA | 0.953 |
| chrX | 57000000 | 57100000 | 0.908 | NA | 0.941 |
| chrX | 57050000 | 57150000 | 0.908 | NA | 0.93 |
| chrX | 57100000 | 57200000 | NA | NA | NA |

3. Save the data track file.

Using the Illumina Sequence Viewer

To use the Illumina Sequence Viewer (ISV):

1. In the IGV main menu, go to **View | Sequence Viewer**.

The Illumina Sequence Viewer appears (Figure 102).

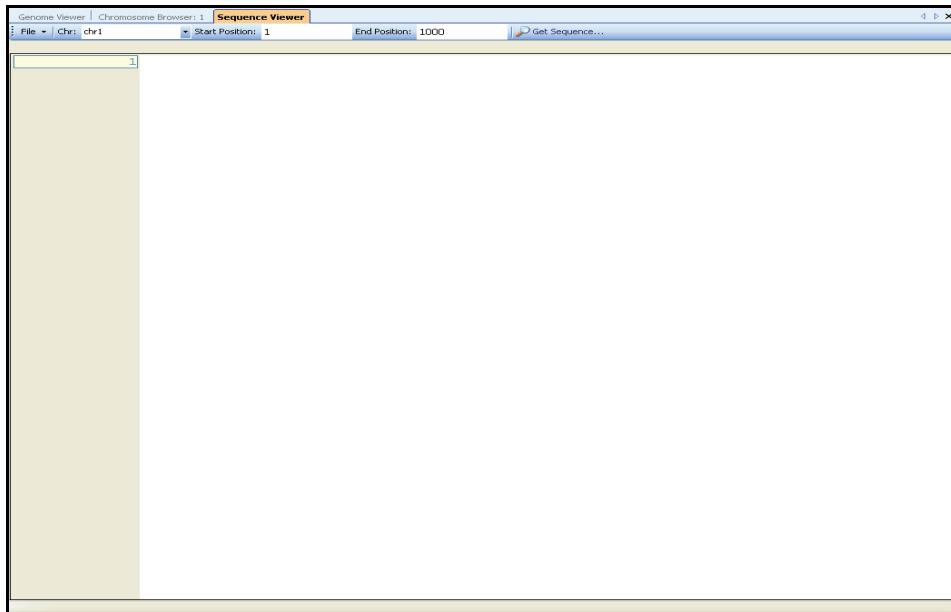


Figure 102 Illumina Sequence Viewer

To view a specific sequence in the ISV:

1. Type the starting base position number of the sequence in the **Start** text field.
2. Type the ending base position number of the sequence in the **End** text field.
3. Click **Get Sequence**.

The sequence you selected appears in the ISV (Figure 103).

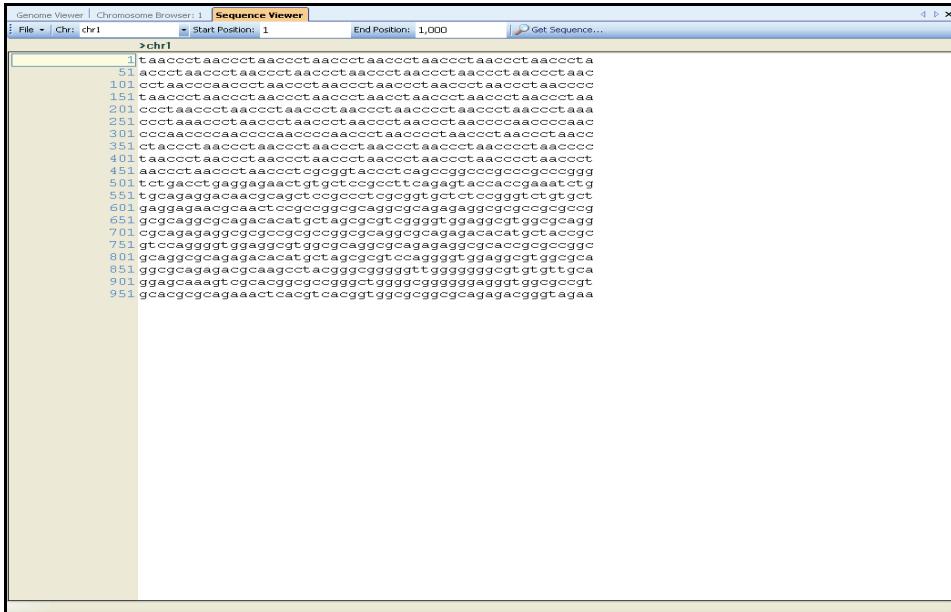


Figure 103 Illumina Sequence Viewer Displaying a Selected Sequence

Using the Chromosome Heat Map

The **Chromosome Heat Map** feature is similar to the heat map feature in the BeadStudio main window; however, it also includes features that are helpful for analyzing large data sets. The Chromosome Heat Map displays data below the length of individual chromosomes, and allows you to scroll through the heat map to see where data is clustered in relation to positions along the chromosome. You can also change the type of data being clustered for each chromosome.

In the Chromosome Heat Map, rows (SNPs) are represented on the X-axis and columns (samples) are represented on the Y-axis. Rows are automatically ordered by chromosome and chromosomal position.

You can use the Chromosome Heat Map to perform the following functions:

- ▶ **Data clustering**—to look for samples with similar behavior.
- ▶ **Data binning**—to reduce the number of data points.
- ▶ **Data smoothing**—to reduce noise and examine trends and patterns.

To use the Chromosome Heat Map feature:

1. In the IGV, go to **View | Chromosome Heat Map**.

The **Chromosome Heat Map** window appears (Figure 104).

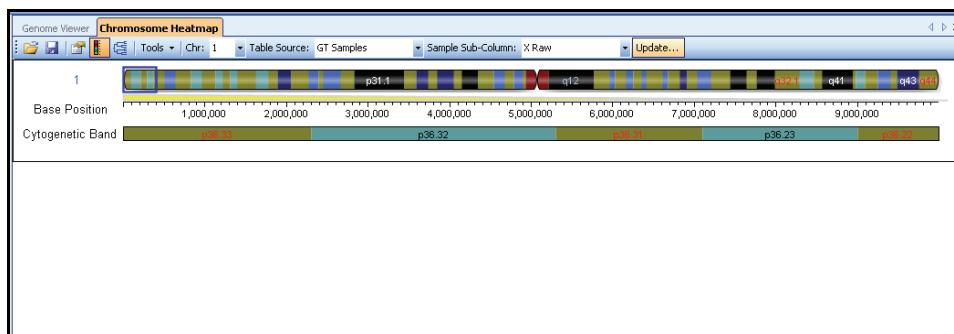


Figure 104 Chromosome Heat Map

2. To populate the Chromosome Heat Map with data, choose the following in the toolbar above the chromosome:
 - A chromosome
 - A table source
 - A sample subcolumn

3. Click **Update**.

The Chromosome Heat Map appears (Figure 105).



Figure 105 Chromosome Heat Map

4. **[Optional]** To adjust the properties of the Chromosome Heat Map, including title, colors, labels, etc.:
 - a. Right-click on the Chromosome Heat Map. The **Properties** dialog box appears.
 - b. Click the various tabs to select new properties.
 - c. Click **OK**.
 The Chromosome Heat Map is displayed with the properties you selected.

Clustering Data

To cluster data within the Chromosome Heat Map:

1. In the **Heat Map** window, click **Cluster heat map rows and/or columns**.

The **Cluster Options** dialog box appears (Figure 106).

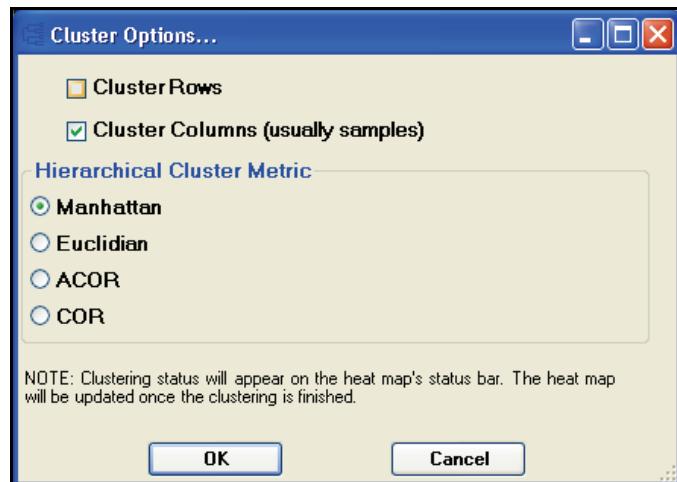


Figure 106 Cluster Options Dialog Box

2. In the **Cluster Options** dialog box, select one or both of the following:
 - **Cluster Rows**
 - **Cluster Columns (usually samples)**
3. Select one of the following **Hierarchical Cluster Metric** options:
 - **Manhattan**—Computes the distance between two points if a grid-like path is followed.
 - **Euclidian**—Computes the shortest distance between two points.
 - **ACOR** (Absolute Correlation)—Computes the Pearson correlation using a $1 - |r|$ distance measure.
 - **COR** (Correlation)—Computes the Pearson correlation using a $1 - r$ distance measure.
4. Click **OK**.

The status bar at the bottom of the window displays the progress of the cluster analysis.
When the data is finished clustering, the heat map automatically displays the hierarchical clusters.

Binning Data

To bin data within the Chromosome Heat Map:

1. In the Chromosome Heat Map, select **Tools | Bin Current Data.**

The **Data Binning Options** dialog box appears (Figure 107).

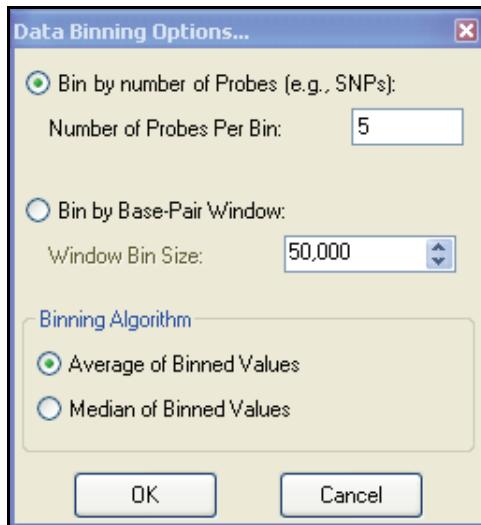


Figure 107 Data Binning Options Dialog Box

2. Choose a binning option:

- **Bin by Number of Probes**
- **Bin by Base Pair Window**

3. Choose a binning algorithm:

- **Average of Binned Values**
- **Median of Binned Values**

4. Click **OK**.

The data is binned and displayed using the parameters you selected.



NOTE

Once you have selected your binning parameters, the binning option in the **Tools** menu is disabled. If you want to re-bin using different parameters, select **Update** from the toolbar menu and follow the binning data procedure above.

Smoothing Data

To smooth sample data within the Chromosome Heat Map:

1. In the Chromosome Heat Map, select **Tools | Smooth Sample Data**.

The **Smooth Sample Data Options** dialog box appears (Figure 108).

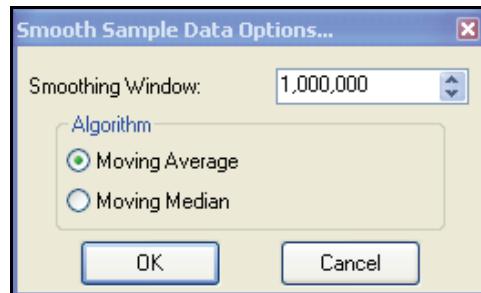


Figure 108 Smooth Sample Data Options Dialog Box

2. Choose a size for the smoothing window.

The units are base pairs.

3. Choose a smoothing algorithm:

- **Moving Average**
- **Moving Median**

4. Click **OK**.

The data is smoothed and displayed in the Chromosome Heat Map using the parameters you selected.



NOTE

Once you have selected your smoothing parameters, the smoothing option in the **Tools** menu is disabled. If you want to re-smooth using different parameters, select **Update** from the toolbar menu and follow the smoothing data procedure above.

Using the Bookmarking Tool

The BeadStudio bookmarking tool allows you to annotate regions of interest in the ICB.

Bookmarks are user-defined marks that include information such as:

- ▶ **Bookmark List**—name of the list to which a particular bookmark belongs.
 - ▶ **Table Source**—table data source with which this bookmark is associated.
- Some modules have more than one data table. For example, the BeadStudio Genotyping Module has a GT Samples table and a Paired Samples table.
- ▶ **Sample ID**—identifier of the particular sample in a particular table data source that this bookmark is associated with.
 - ▶ **Bookmark Type**—classification of the bookmark; for example, a region with LOH or another chromosomal aberration.
 - ▶ **Base Start Position**—base number of the starting point of the bookmarked region.
 - ▶ **Base End Position**—base number of the ending point of the bookmarked region.

To use the BeadStudio bookmarking tool:

1. Right-click anywhere in the ICB main window.

The context menu appears (Figure 109).

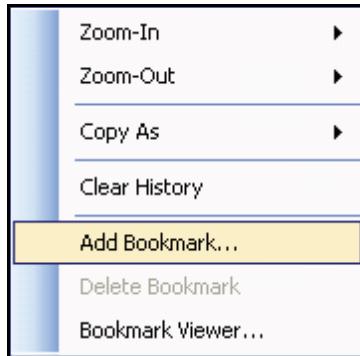


Figure 109 ICB Context Menu

2. Select **Add Bookmark**.

The **Add Bookmark** window appears (Figure 110).

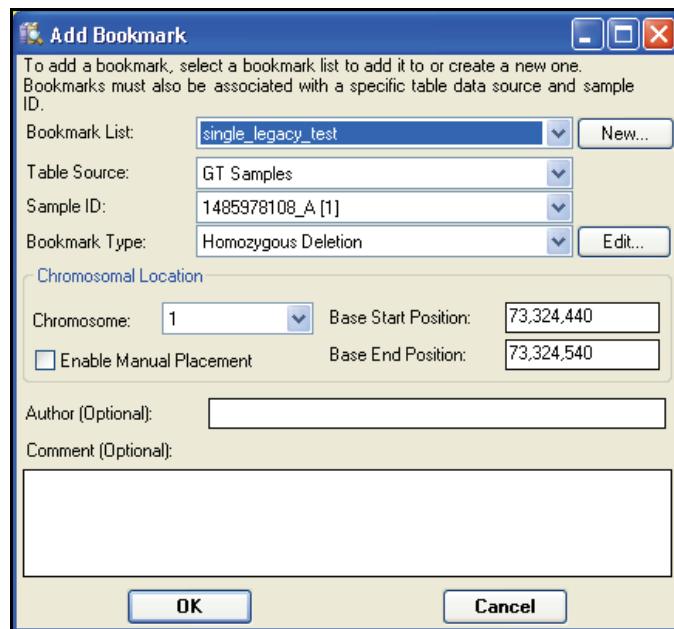


Figure 110 Add Bookmark Window

3. Using the dropdown menus, do the following:
 - a. Assign the bookmark to a **Bookmark List**.
 - b. Associate the bookmark with a **Table Source**.
 - c. Associate the bookmark with a **Sample ID**.
 - d. Assign a **Bookmark Type**.
4. **[Optional]** Create a new bookmark. See *Saving a Bookmark* on page 126.
5. **[Optional]** Create a new bookmark list. See *Creating a Bookmark List* on page 128.
6. **[Optional]** Edit bookmark characteristics. See *Editing Bookmark Characteristics* on page 130.
7. Click **OK**.

The bookmark you created is added to the bookmark file you designated.

Once you create and display a bookmark, it is superimposed on data plots within the ICB (Figure 111), and saved with your project. This allows you to refer to the bookmarked region again later.

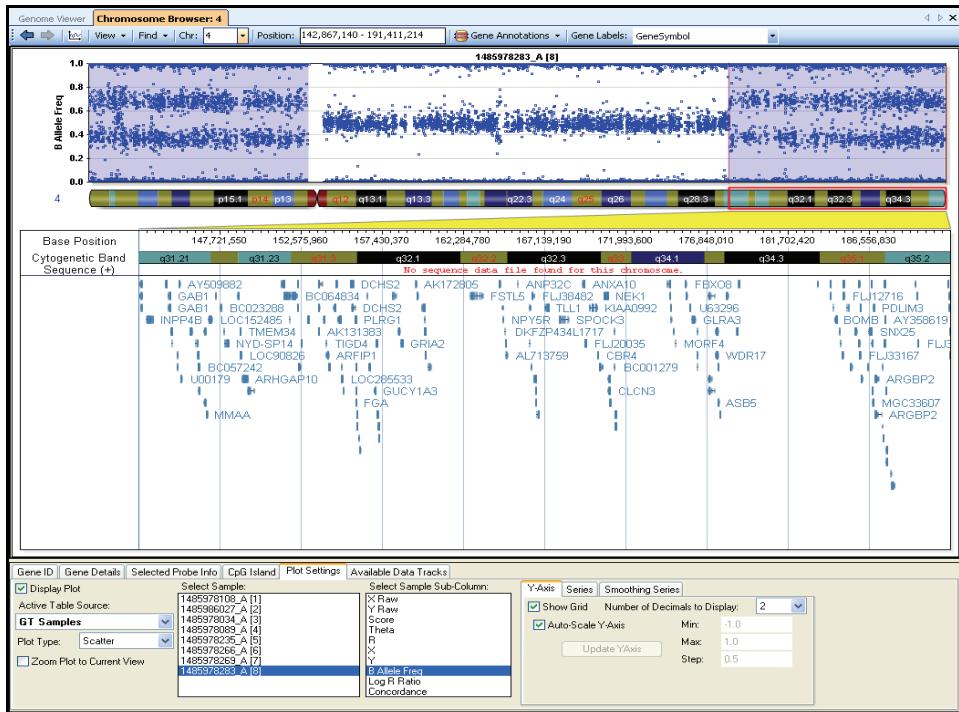


Figure 111 Bookmarks Displayed in the ICB

Bookmarks are flexible in that they can be moved to other regions in the ICB. In addition, you can export bookmarks from BeadStudio and later import them into another BeadStudio workspace. Using the Reports function, you can also export a summary of the data contained within the bookmarks.

Drawing a Bookmark

Bookmarks are marks that you draw over a plot of data along a user-defined length of a chromosome shown in the ICB. You can use a bookmark to annotate a chromosomal aberration that you want to analyze.

To draw a bookmark, perform the following tasks:

1. In the ICB, click the **Plot Settings** tab to activate it.
2. In the **Plot Settings** tab, select the **Display Plot** checkbox (Figure 112).

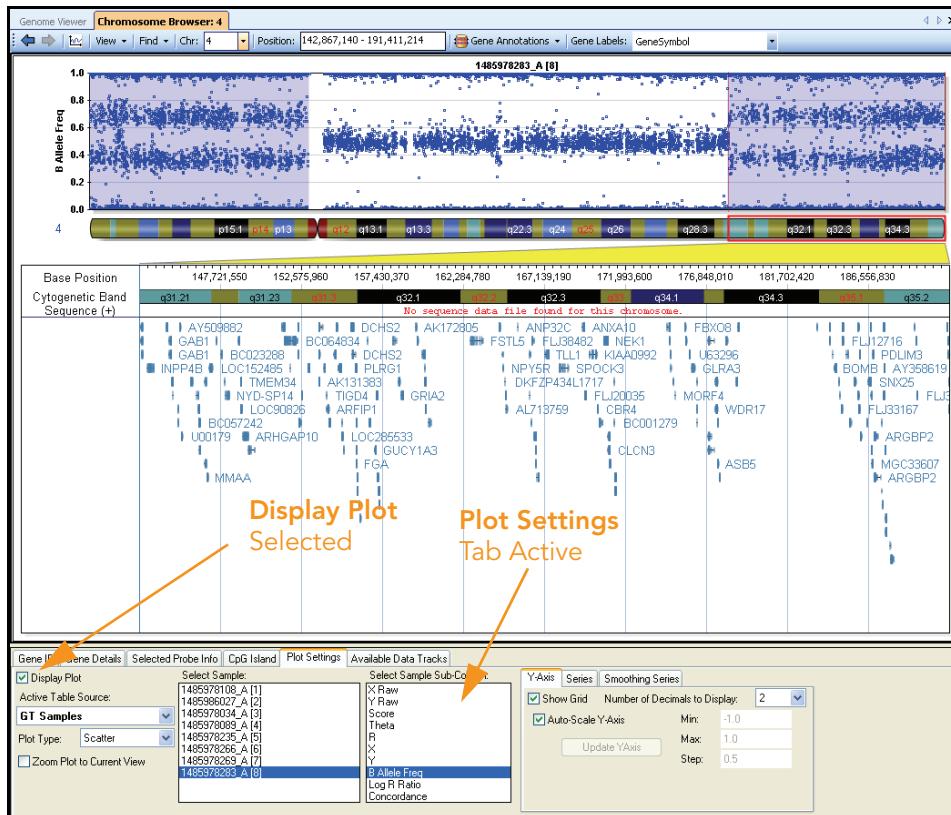


Figure 112 Drawing Bookmarks

3. Move the View Region defined by the red rectangle to a region of interest by positioning the cursor directly over the center of the region and dragging it to the left or right (Figure 113).

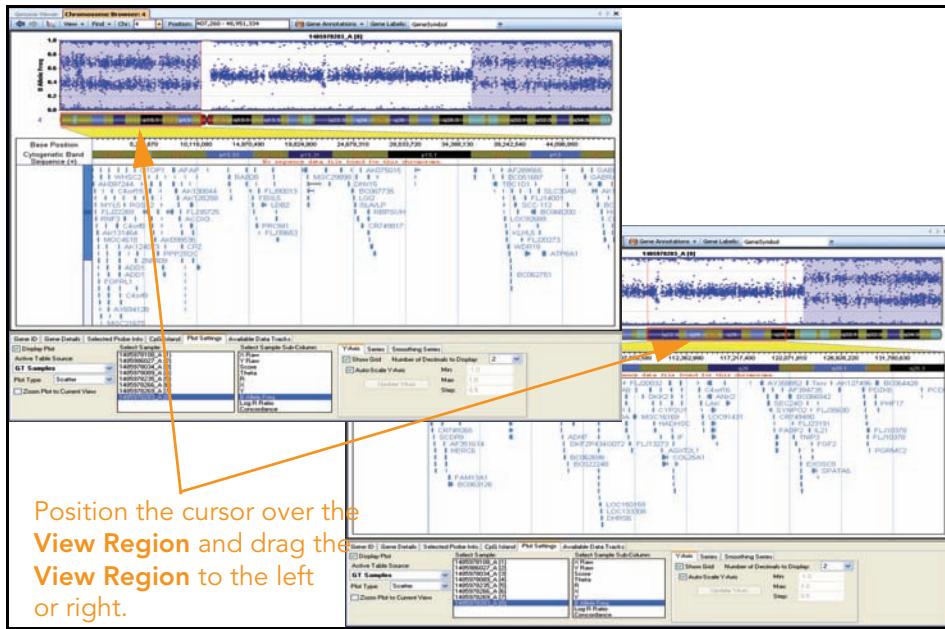


Figure 113 Moving the View Region

4. Resize the View Region by performing the following steps:
 - a. Select the left or right edge of the View Region represented by the red rectangle.
 - b. Watch for the cursor to change to this shape: <-->. When the cursor changes to <-->, you can change the size of the View Region.
 - c. Drag the edge of the View Region until the region is the size you want it to be (Figure 114).

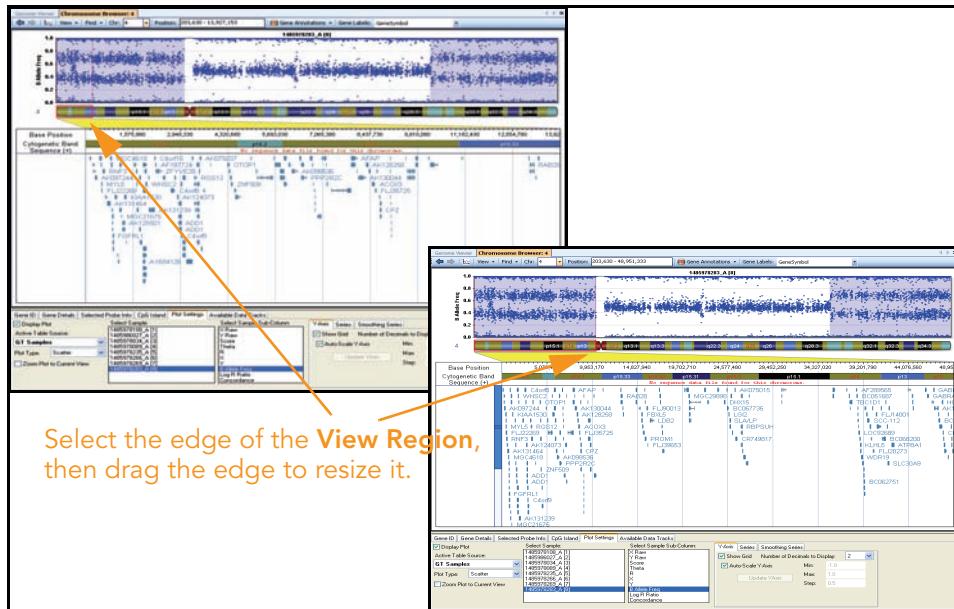


Figure 114 Resizing the View Region

You have successfully drawn a bookmark. Now you can save the bookmark so that it can be used again later.

Saving a Bookmark

When you identify a region of interest, you can create and save a bookmark, or shaded region, which is superimposed over the corresponding data. Saving a bookmark allows you to mark a region of interest so that you can easily return to it later.

To save a bookmark:

1. Go to the Bookmark Viewer by doing one of the following:
 - Right-click and select **Bookmark Viewer** from the ICB context menu and click **Add...**.
 - Right-click and select **Add Bookmark**.

The **Add Bookmark** dialog box appears (Figure 115).

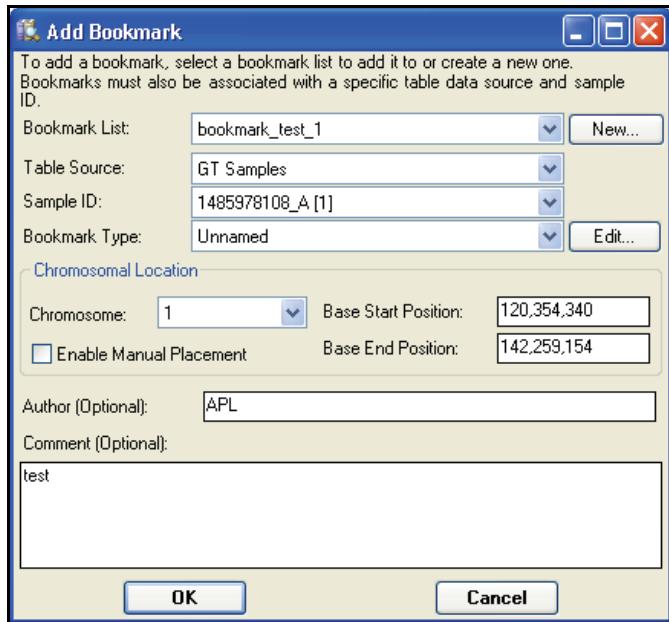


Figure 115 Add Bookmark Dialog Box

2. In the **Add Bookmark** dialog box, edit the characteristics of the bookmark, including the following:
 - Bookmark List
 - Table Source
 - Sample ID
 - Bookmark Type
 - Chromosome Number
 - Chromosomal Location
 - Author
 - Comment
3. Click **OK**.
A bookmark is created in the current position of the View Region.

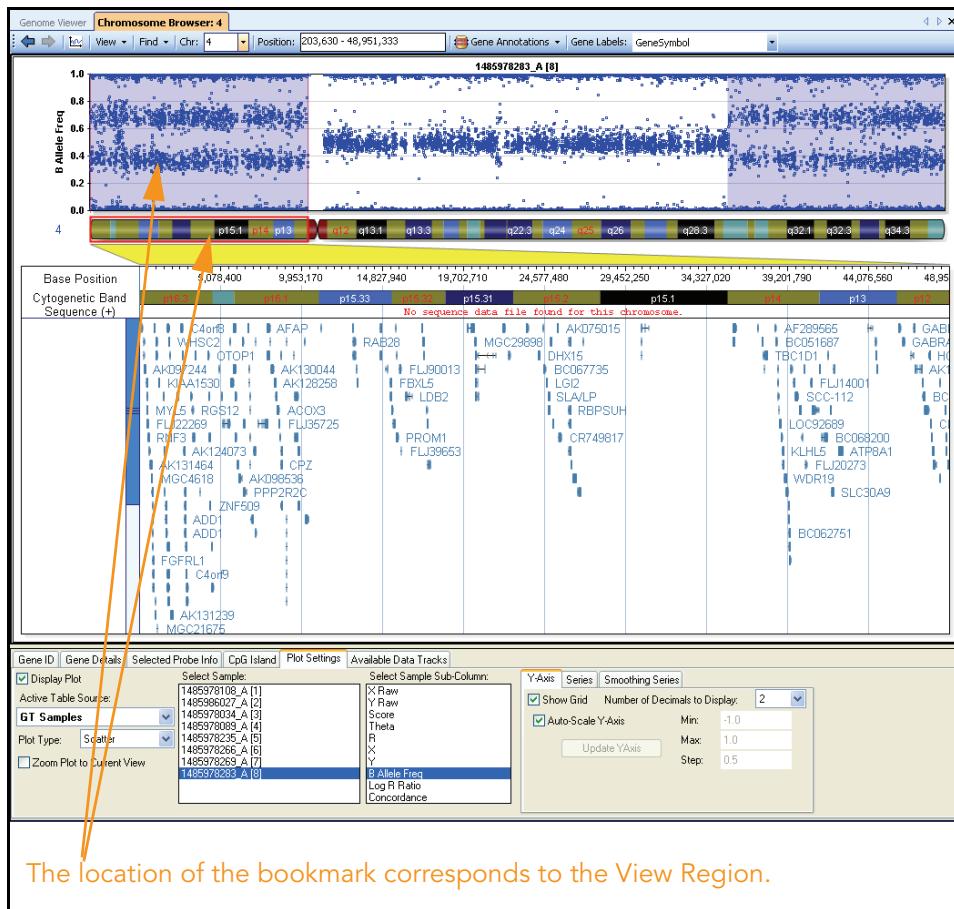


Figure 116 Bookmark Created at View Region

After you have created a bookmark, you can edit the characteristics of the bookmark at any time.

Creating a Bookmark List

Perform the following steps to create a bookmark list:

1. In the **Add Bookmark** window, select **New**.

The **New Bookmark List** dialog box appears (Figure 117).

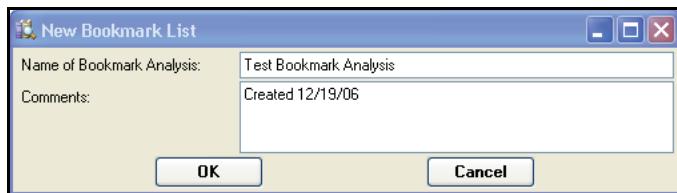


Figure 117 New Bookmark List Dialog Box

2. Enter a name for your new bookmark in the **Name of Bookmark Analysis** text field.
3. **[Optional]** Enter comments in the **Comments** text field.
4. Click **OK**.

The new bookmark list is added to the selections in the **Bookmark List** dropdown menu in the **Add Bookmark** dialog box.

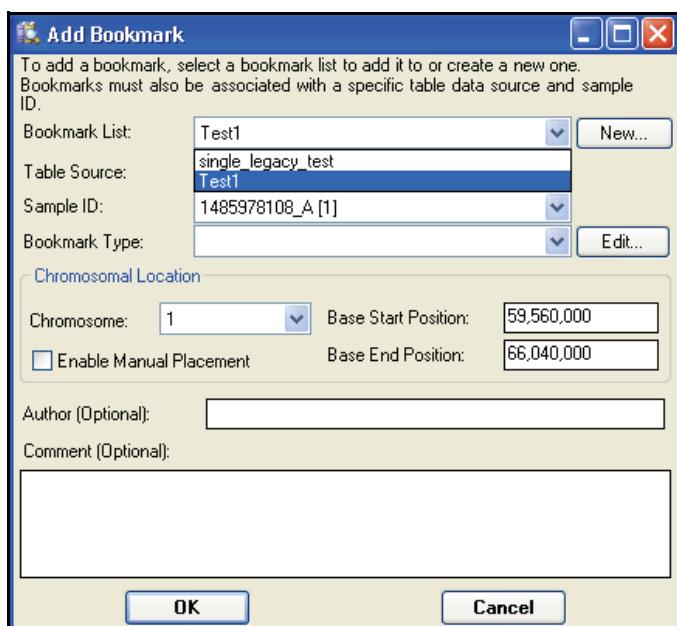


Figure 118 Add Bookmark Dialog Box, New Bookmark Visible

Editing Bookmark Characteristics

Once you have created a bookmark, you can edit its characteristics in the **Add Bookmark** dialog box.

Choosing & Editing Bookmark Types

To choose a bookmark type:

1. In the ICB, right click and select **Add Bookmark** from the context menu.
The **Add Bookmark** dialog box appears.
2. In the **Add Bookmark** dialog box, click **Edit**.
The **Edit Bookmark Types** dialog box appears (Figure 119).

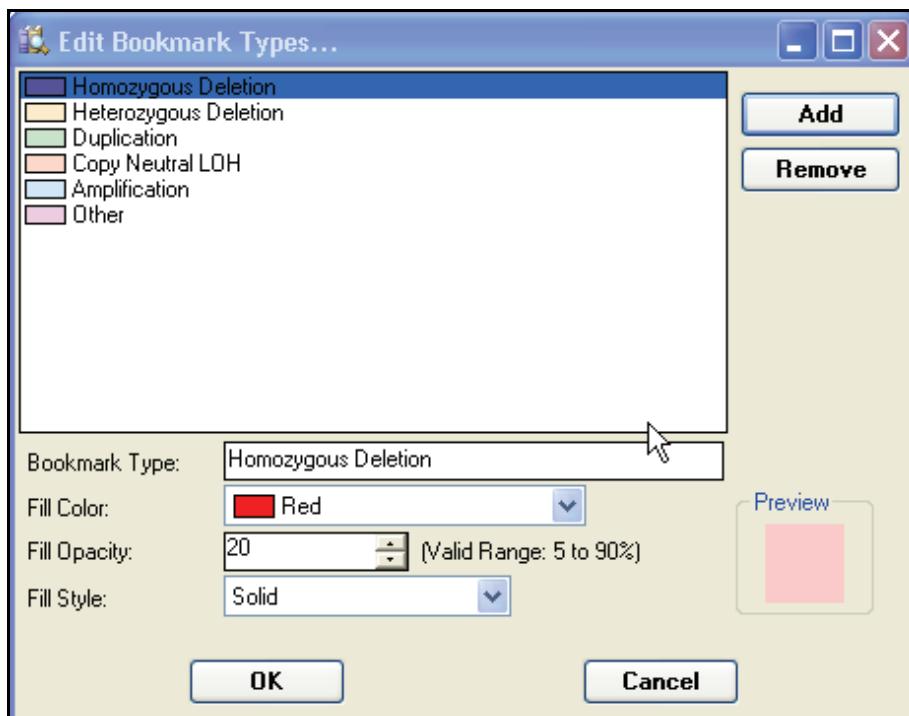


Figure 119 Edit Bookmark Types Dialog Box

3. Select a bookmark type for the region you want to bookmark.

Several bookmark types are set as default options:

- Homozygous deletion
- Heterozygous deletion
- Duplication
- Copy-Neutral LOH
- Amplification



NOTE

If this list does not include a bookmark type you want to use, choose **Other**. You can define and create additional bookmark types.

- 4.** **[Optional]** If you would like to customize the appearance of your bookmarks, select options for the following characteristics:

- **Fill opacity**—the opacity percentage of the bookmark.
A lower opacity percentage allows you to see the data through the bookmark, while a higher opacity percentage obscures the data behind the bookmark.
- **Color**—the color of the bookmark.
- **Style**—the pattern of the bookmark.

- 5.** Click **OK**.

Your bookmark is displayed in the ICB.

Adding Comments to Bookmarks

To add comments to a bookmark:

1. In the **Add Bookmark** window, enter your comments in the **Comment (Optional)** text field (Figure 120).

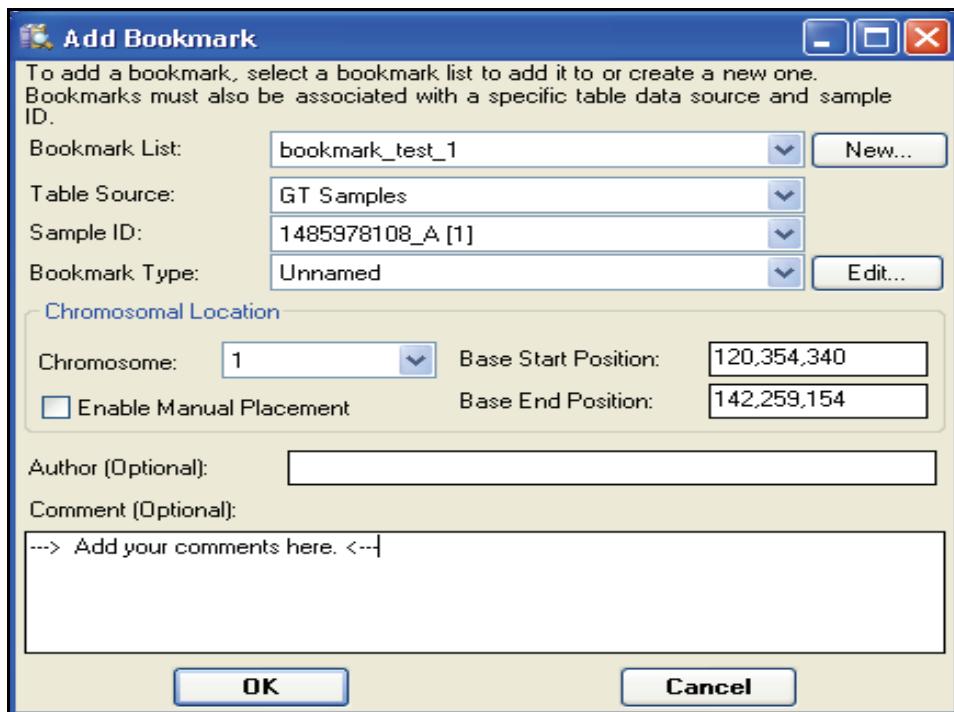


Figure 120 Adding Comments to a Bookmark

2. Click **OK**.

Your comments are saved with the bookmark.



NOTE

You can view your comments with corresponding bookmarks in the Bookmark Viewer and in LOH Reports.

Displaying & Hiding Bookmarks

Bookmarks for various samples can be displayed simultaneously, or you can choose to display only user-defined bookmarks in the ICB.

Displaying Bookmarks

Use one of the following procedures to display bookmarks.

Displaying Selected Bookmarks

To display selected bookmarks:

1. In the ICB, select **View | Bookmark Viewer**.
The **Bookmark Analyses** window appears (Figure 121).
2. Select the individual bookmarks you want to display in the ICB. If a bookmark is not selected, it will not be displayed.

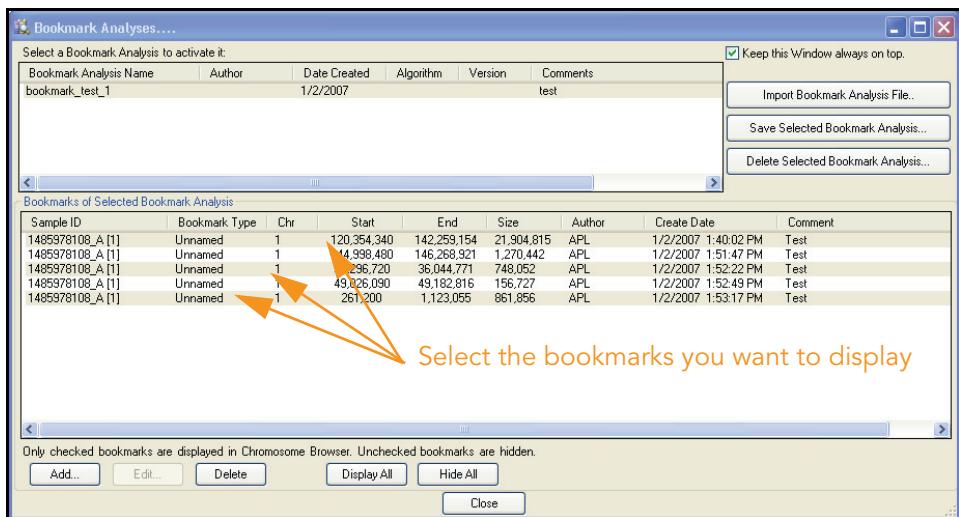


Figure 121 Selecting Bookmarks to Display in the ICB

3. Click **OK**.

The selected bookmarks appear in the ICB.

Displaying All Bookmarks

To display all bookmarks:

1. In the ICB, select **View | Bookmark Viewer**.
The **Sample Bookmarks** window appears (Figure 122).
2. Click **Display All Bookmarks**.

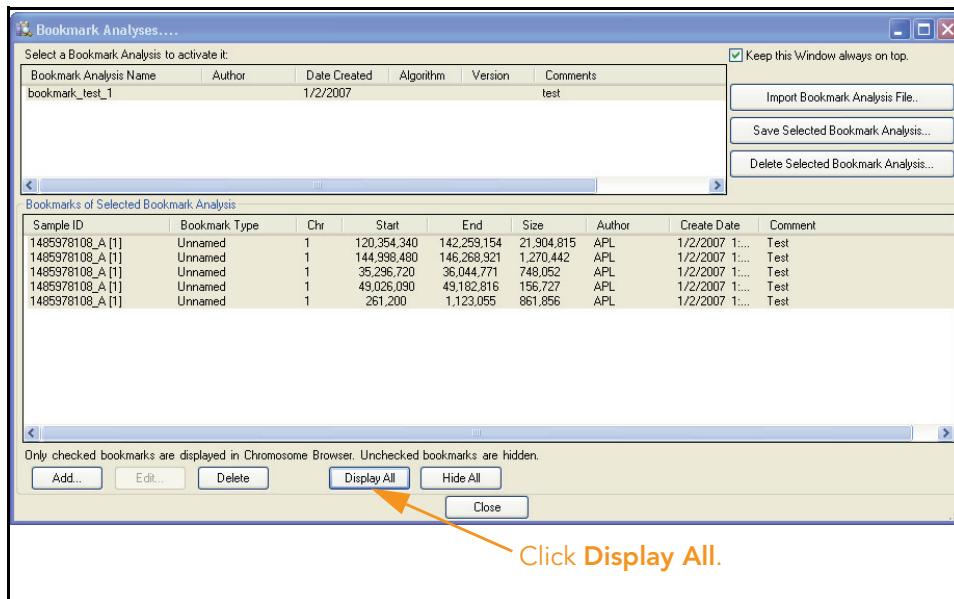


Figure 122 Displaying All Bookmarks

3. Click **Close**.

The checkboxes for all bookmarks are selected, and all bookmarks appear in the ICB.

Hiding Bookmarks

You can choose to hide only bookmarks you select, or you can hide all bookmarks.

Hiding Selected Bookmarks

To hide selected bookmarks:

1. In the ICB, select **View | Bookmark Viewer**.

The **Bookmark Analyses** window appears (Figure 123).

2. Deselect each bookmark you want to hide.

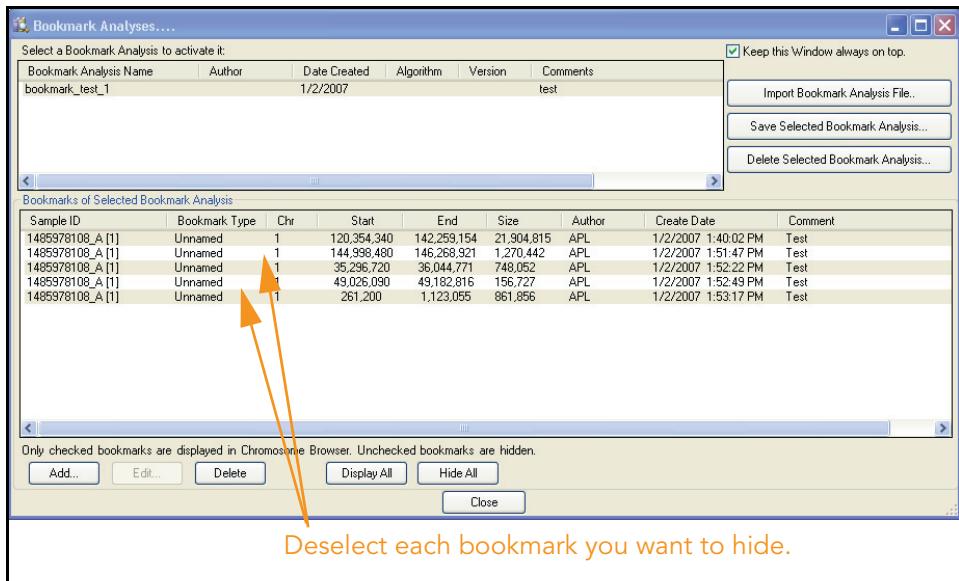


Figure 123 Hiding Selected Bookmarks

3. Click **Close**.

The bookmarks you deselected are hidden.

Hiding All Bookmarks

To hide all bookmarks:

1. In the ICB, select **View | Bookmark Viewer**.

The **Bookmark Analyses** window appears (Figure 124).

2. Click **Hide All**.

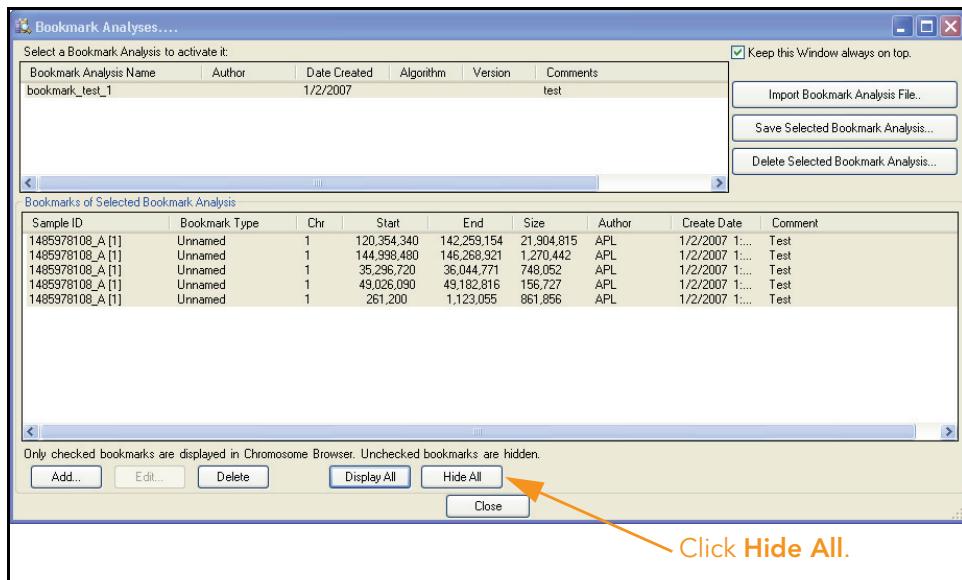


Figure 124 Hiding All Bookmarks

3. Click **Close**.

No bookmarks are visible in the ICB.

Viewing Bookmarks in the ICB

Viewing bookmarks in the ICB is a good way to visualize regions of interest across multiple samples.

Displaying Bookmark Information

There are several ways to display bookmark information:

1. Right-click on a bookmark of interest.
2. Choose **Bookmark Viewer**.

The information for each bookmark is displayed in the Bookmark Viewer window.

Alternatively, do one of the following:

- Go to **View | Bookmark Viewer**.
- To display information pertaining to each individual bookmark, place the mouse cursor over a bookmark in the ICB.

A popup window that contains the bookmark information you entered appears (Figure 125).

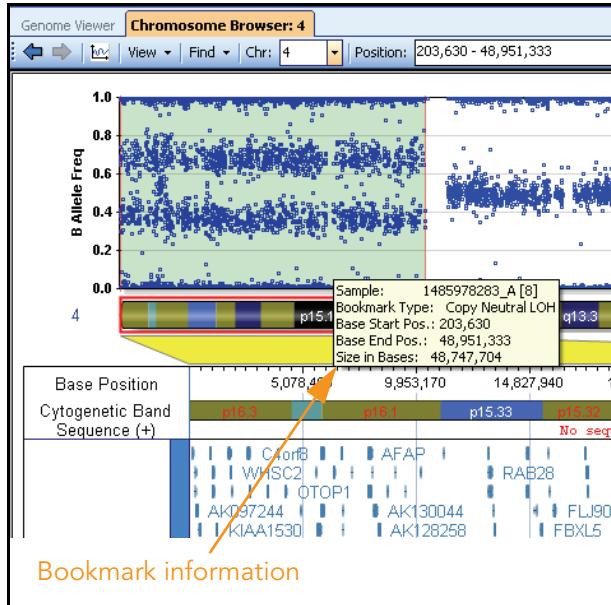


Figure 125 Bookmark Information Popup Window

Zooming Into a Selected Region

At times it may be useful to view only certain portions of the data shown in the ICB.

To zoom into a selected region:

1. In the ICB, select **Display Plot**.
2. Using the View Region, choose which region of data you would like to display in the ICB.
3. Select **Zoom Plot to Current View**.

The selected region is shown in the ICB (Figure 126).

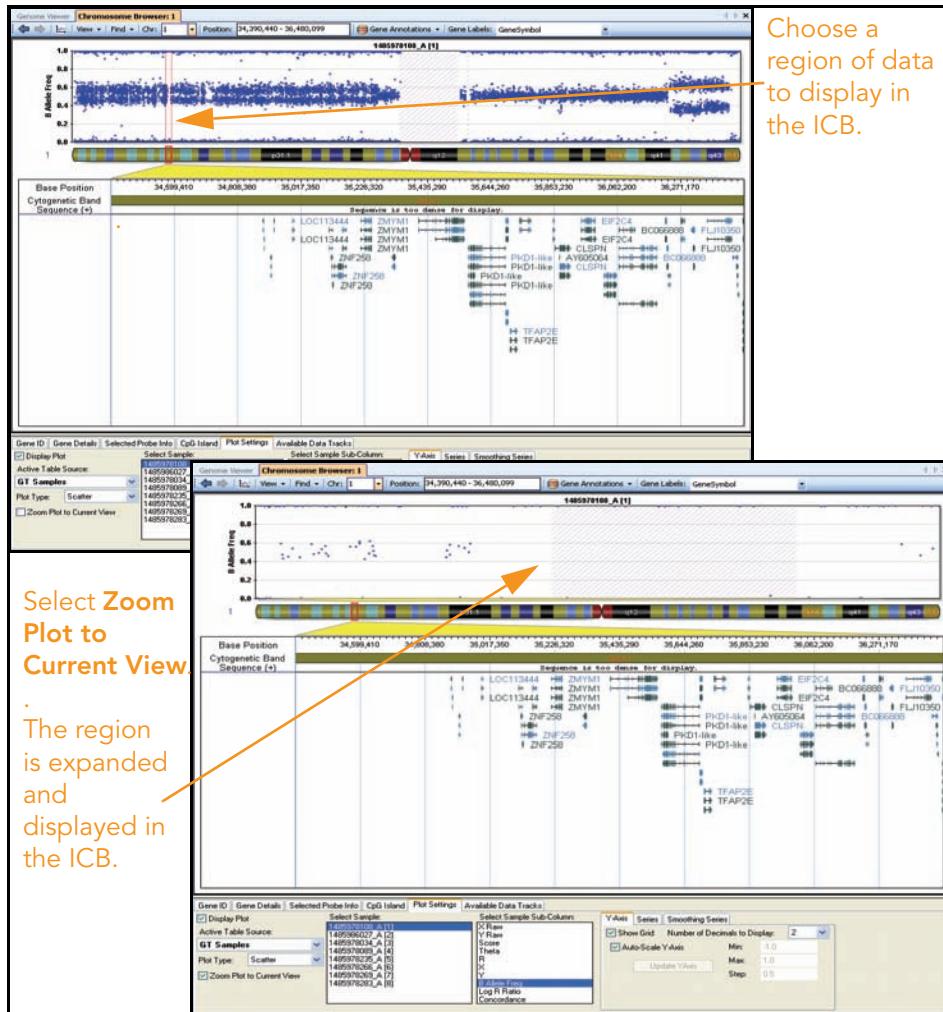


Figure 126 Zoom Plot to Current View

Locking Bookmarks

You may want to lock your bookmarks to prevent them from being moved inadvertently.

To lock bookmarks:

1. Open the **Bookmark** window and select the bookmark you want to edit.

2. Click **Edit**.

The **Edit Bookmark** window appears (Figure 127).

3. Clear the **Enable Manual Placement** checkbox.

4. This bookmark is now fixed in place and cannot be moved in the ICB.

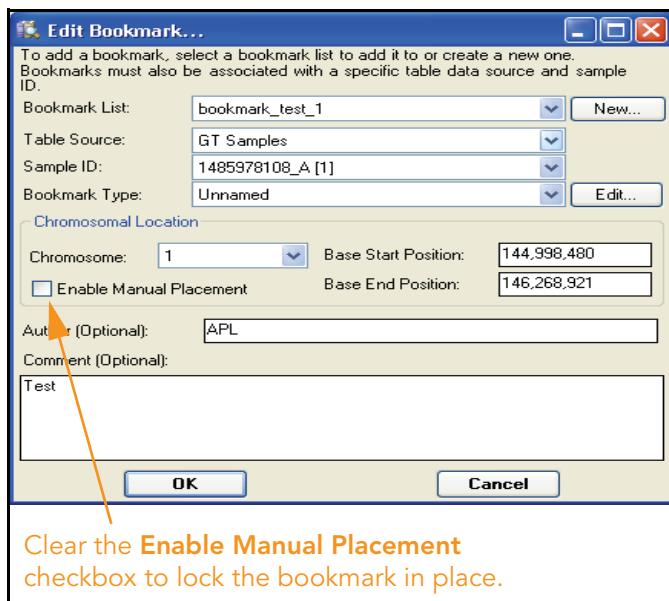


Figure 127 Enable Manual Placement Checkbox Cleared

Adjusting Bookmark Size & Position

The size and position of each bookmark can be adjusted to cover more (or fewer) data points, or a different region, in the ICB.

To adjust bookmark size:

- ▶ Resize the bookmarked region by selecting and dragging the left or right edge of the bookmark.

To adjust bookmark position:

- ▶ If the **Enable manual placement** checkbox is selected, a bookmark can be manually repositioned by choosing it with the mouse and dragging to a new position.
- ▶ If the **Enable manual placement checkbox** is cleared, a bookmark cannot be moved to a new position.

Deleting Bookmarks

If you no longer need one or more bookmarks, you can delete them from the ICB individually or in a batch.

Deleting Individual Bookmarks

To delete an individual bookmark:

1. In the ICB, select **View | Bookmark Viewer**.
 - In the **Bookmark Viewer** window, click once on the descriptive information for the bookmark you want to delete.
2. Click **Delete**.

The **Delete Selected Bookmarks** dialog box appears (Figure 128).

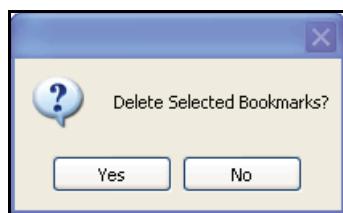


Figure 128 Delete Selected Bookmarks Dialog Box

3. Click **Yes**.

The bookmark is removed from the **Bookmark Viewer** window and the ICB.

Deleting Multiple Bookmarks

To delete multiple bookmarks:

1. In the ICB, select **View | Bookmark Viewer**.
 2. In the **Bookmark Viewer** window, click once on the first bookmark you want to delete.
 3. Press and hold the **Ctrl** key on your keyboard.
 4. Select additional bookmarks you want to delete.
- The bookmarks you have chosen to delete are highlighted (Figure 129).

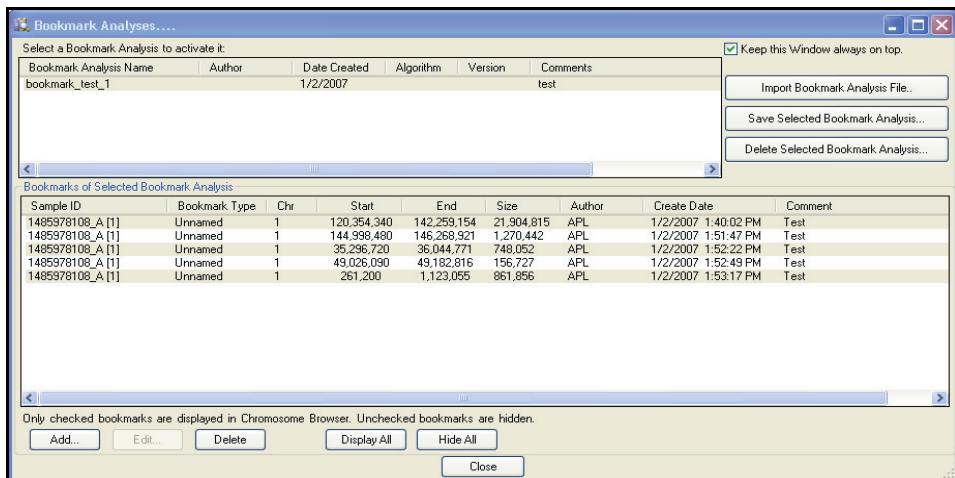


Figure 129 Deleting Multiple Bookmarks

5. Click **Delete**.

The **Delete Selected Bookmarks** dialog box appears.

6. Click **Yes**.

The selected bookmarks are deleted from the **Bookmark Viewer** window and the ICB.

Exporting Bookmarks

Exporting bookmarks is an easy way to share results with other BeadStudio users.

To export bookmarks to an *.xml file:

1. Go to **File | Save Bookmarks to File**.
 2. Type a name for your bookmark file.
 3. Browse to the location where you want to save your bookmark file.
- The bookmark file is saved as an *.xml file.
4. Click **Save**.

Your bookmark file is saved in the location you specified.

You can share a bookmark file with colleagues by telling them the location of the bookmark file and having them import it into their BeadStudio project(s).

Importing Bookmarks

Bookmarks from one BeadStudio project can be imported into any other BeadStudio project as long as the sample names are the same and you are using the same module (GT, LOH+, etc.). By importing and exporting bookmark files, you can share bookmark files with other BeadStudio users, and save bookmark files to be used again later.

To import a bookmark file:

1. Go to **Analysis | Show Columns in Genome Viewer**.
 2. In the Illumina Genome Viewer (IGV), click on the chromosome to open the Illumina Chromosome Browser (ICB).
 3. Click  **Toggle Plot Display** to activate the **Plot Settings** tab. The **Display Plot** checkbox is automatically selected.
Alternatively, go to the **Plot Settings** tab and manually select the **Display Plot** checkbox.
 4. Position the cursor somewhere in the main window, right-click, and select **Bookmark Viewer** from the context menu.
Alternatively, go to **View | Bookmark Viewer**.
- The **Sample Bookmarks** window opens.

5. Go to **File | Import Bookmark File**.
6. Browse to the location of the bookmark file you want to import.
7. Click **Open**.

The bookmarks are loaded in the **Sample Bookmarks** window and overlayed on the data displayed in the ICB.

**NOTE**

To view or hide specific bookmarks, choose various options in the lower part of the **Sample Bookmarks** window.

Appendix A

Troubleshooting Guide

Topics

- 146 Introduction
- 146 Frequently Asked Questions

Introduction

Use this troubleshooting guide to assist you with any questions you may have about the BeadStudio Framework.

Frequently Asked Questions

Table 10 lists frequently asked questions and associated responses.

Table 10 Frequently Asked Questions

| # | Question | Response |
|---|---|--|
| 1 | What is a data repository? | A data repository is a parent directory that contains subdirectories with *.idat files generated from the Sentrix Array products you have scanned in your experiments. BeadScan, the scanning software for the BeadArray Reader, automatically creates a subdirectory in the data repository you defined for each Sentrix array product it scans. You specify a data repository when you create a new project. |
| 2 | What is a project repository? | The project repository is a parent directory that contains BeadStudio project files. You specify a project repository when you create a new project. |
| 3 | What is an *.idat file? | An *.idat file is an intensity data file. It contains statistics for every bead type on your Sentrix Array product. The statistics in an *.idat file include the number of beads, the mean, and the standard deviation for each color sample. There is one *.idat file per sample per channel. |
| 4 | How can I share a project with a colleague? | The project folder from your repository can be copied and shared, and your colleague can open it within BeadStudio. |