BeadStudio Framework User Guide

A Modular Tool for Illumina Data Analysis

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Revision History

Revision Letter	Release Date
A	12/13/05

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Chapter 1

Overview

Topics

Introduction

Illumina's BeadStudio version 2.0 is a modular software application that allows you to analyze data from Illumina's Sentrix® array products.

BeadStudio's analysis modules allow you to interface with the data generated from Illumina's various assays.

After installing the BeadStudio 2.0 Framework, you separately install various BeadStudio analysis modules. BeadStudio automatically detects and loads any modules you have installed.

NOTE:

To use BeadStudio, you must have at least one licensed BeadStudio module installed.

The main BeadStudio graphical user interface (GUI) and certain features of the analysis modules look the same and have the same functionality across modules. These features are part of the BeadStudio Framework.

This manual describes Illumina's BeadStudio Framework.

Installing BeadStudio

To install the BeadStudio Framework application:

1. Double-click the **SetupBeadStudio.msi** icon

   in the BeadStudio directory of the CD you received and follow the on-screen instructions.

2. The **BeadStudio Setup Wizard** guides you through the installation process (Figure 1-1 - Figure 1-6).

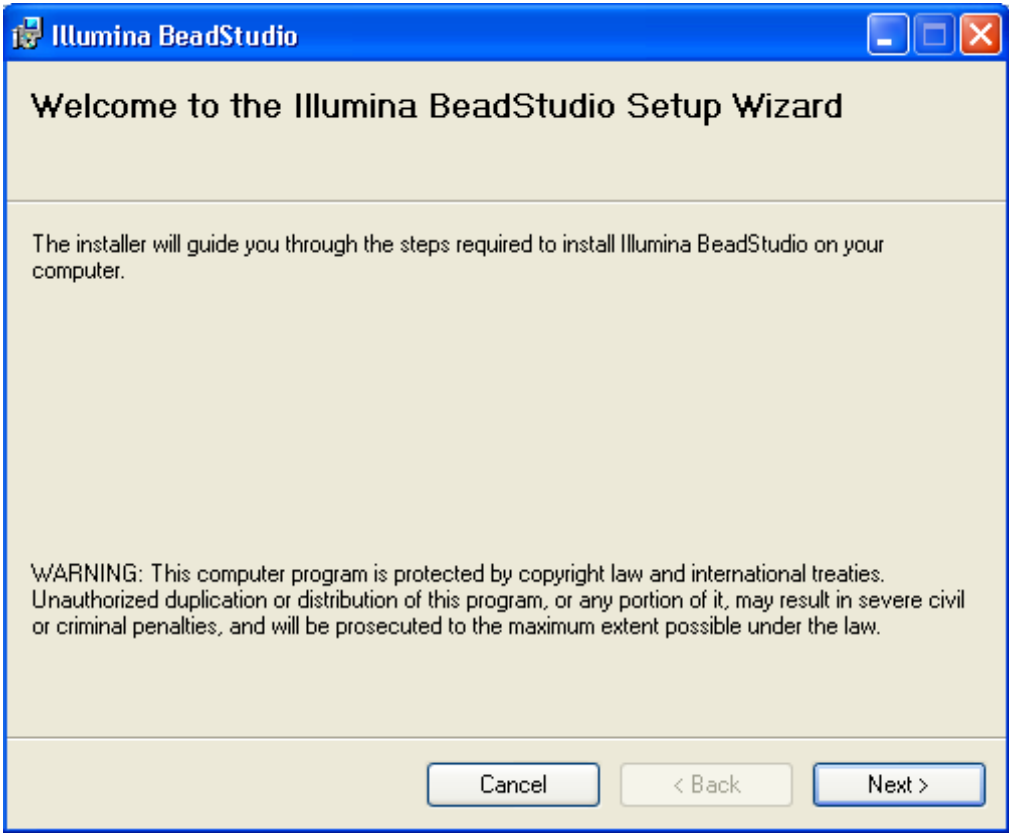

Figure 1-1 Welcome to the Illumina BeadStudio Setup Wizard

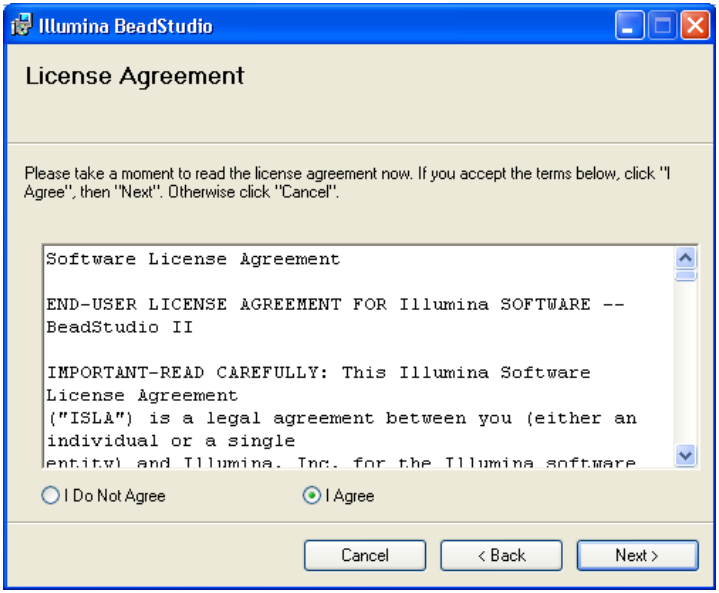

Figure 1-2 License Agreement

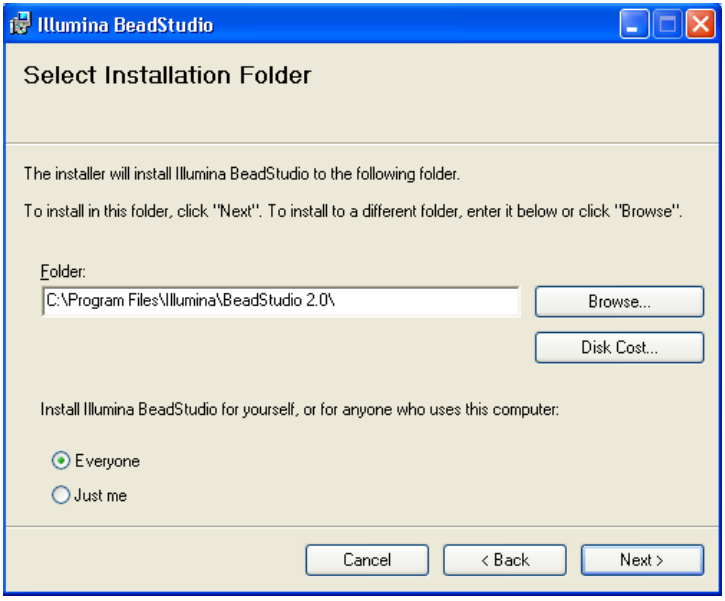

Figure 1-3 Select Installation Folder

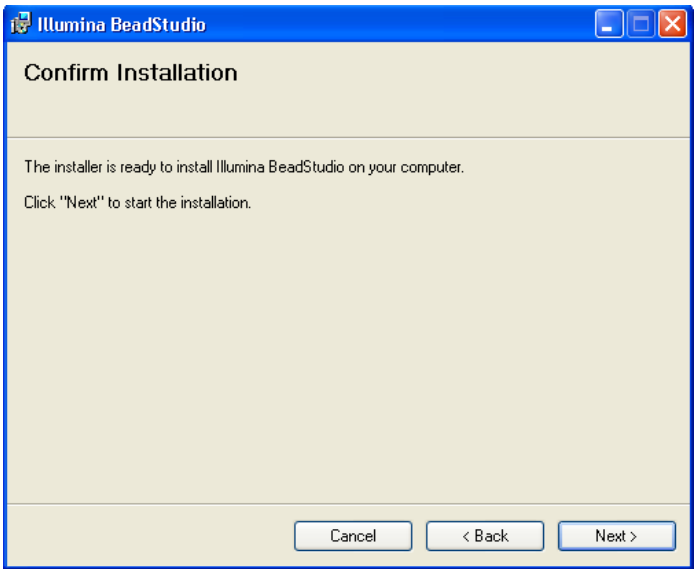

Figure 1-4 Confirm Installation

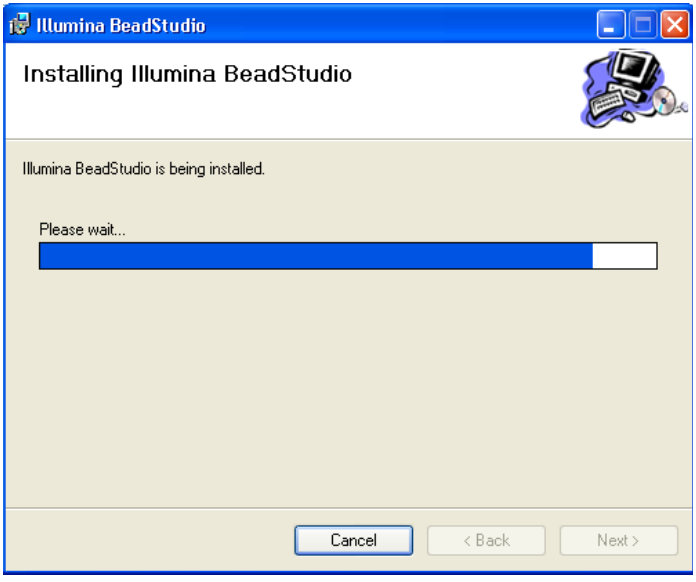

Figure 1-5 Installing Illumina BeadStudio

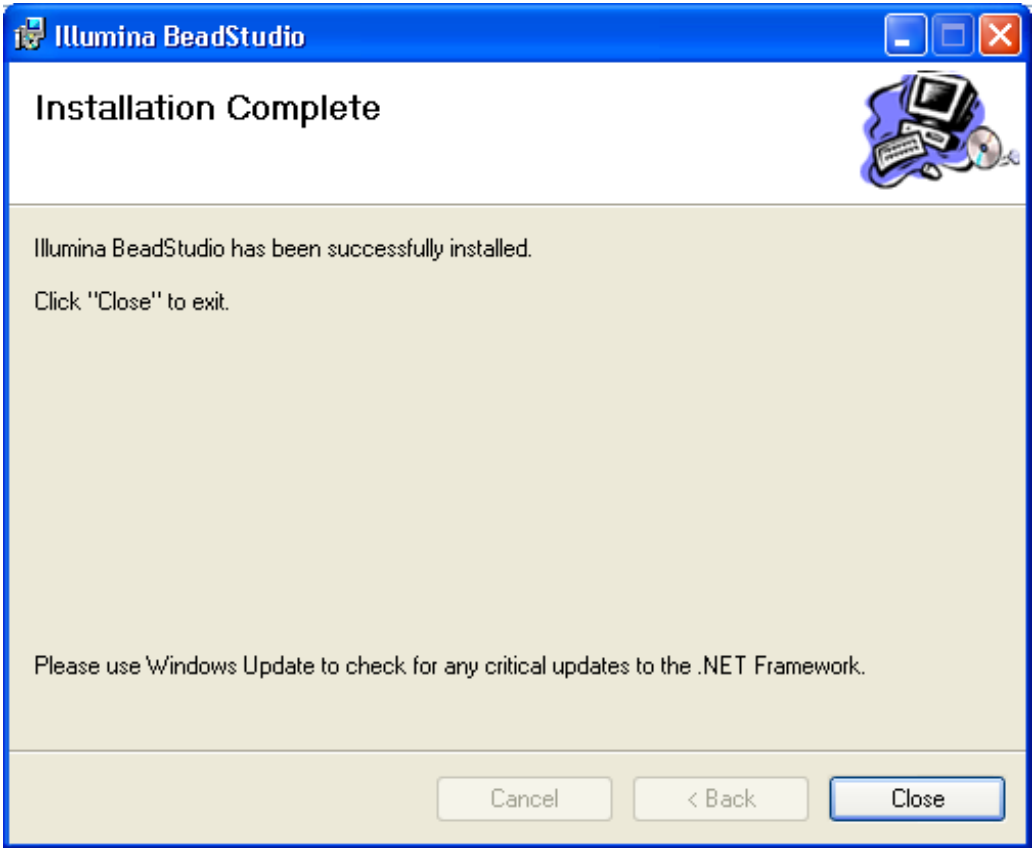

Figure 1-6 Installation Complete

Starting BeadStudio

To start the BeadStudio application 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 **Start Page** (Figure 1-7).

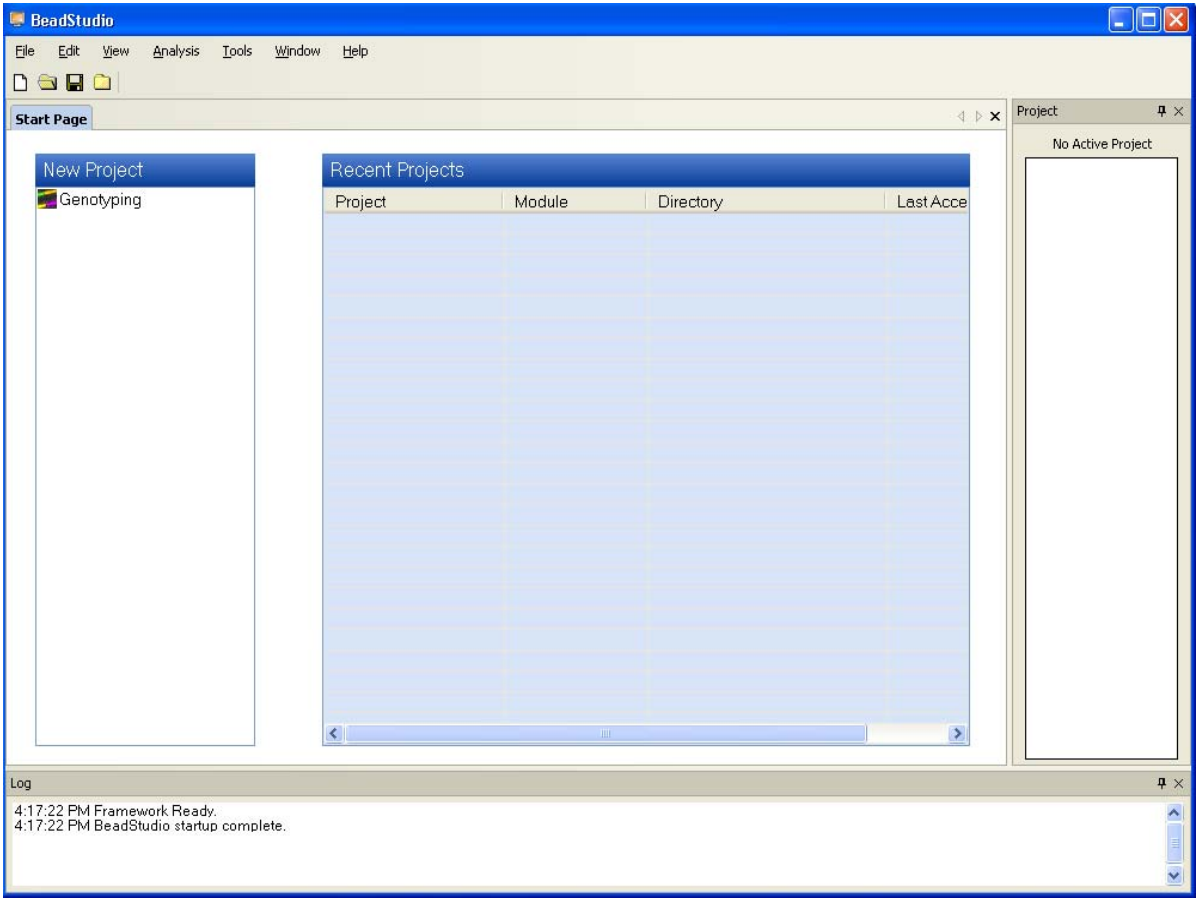

Figure 1-7 Start Page

Exiting BeadStudio

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

- Select **File | Exit**.
- Click the **Close** button in the upper-right corner of the main window.

  The BeadStudio application closes.

NOTES

Chapter 2

Detachable Docking Windows

Topics

Introduction

BeadStudio has a flexible interface that allows you to organize and view your data in a number of ways. The following sections describe the configurable parts of the BeadStudio interface which are common to all modules.

Working with Detachable Docking Windows

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

Use the table docking icons shown in Figure 2-1 through Figure 2-4 as guides when repositioning windows.

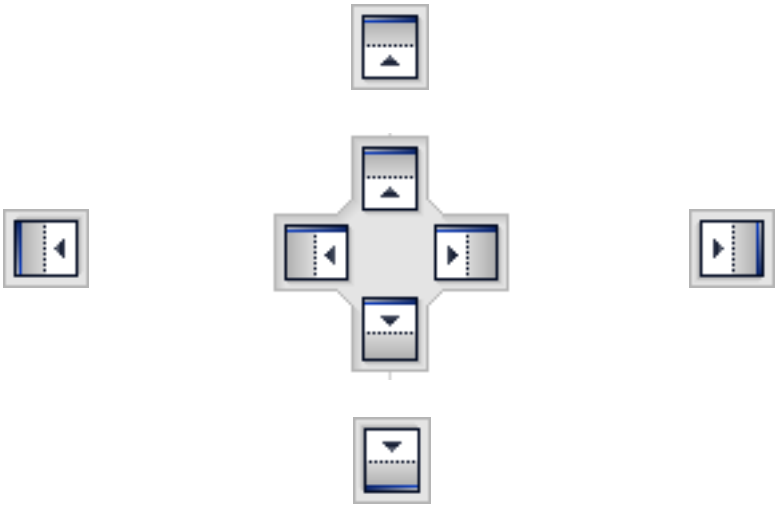

Figure 2-1 Docking Icons

Use the following techniques to detach and dock a window:

1. Click the window's title tab.
2. Drag the window's title tab over the main window.

As you drag the window over the Main Window, BeadStudio displays docking icons (Figure 2-1) that indicate potential docking locations. When you drag the window over a docking icon, BeadStudio draws an outline indicating where the window would be docked if you were to release the mouse button over that docking icon.

The windows docking function allows great flexibility; the best way to learn is to experiment, using the general guidelines below.

- To dock a window on top of an already-docked window (so that it becomes a member of the tab group), drag the mouse over the center docking icon in the destination window (Figure 2-2) and release the mouse button.

  The window is now docked on top of the already-docked window, and is a member of that window's tab group.

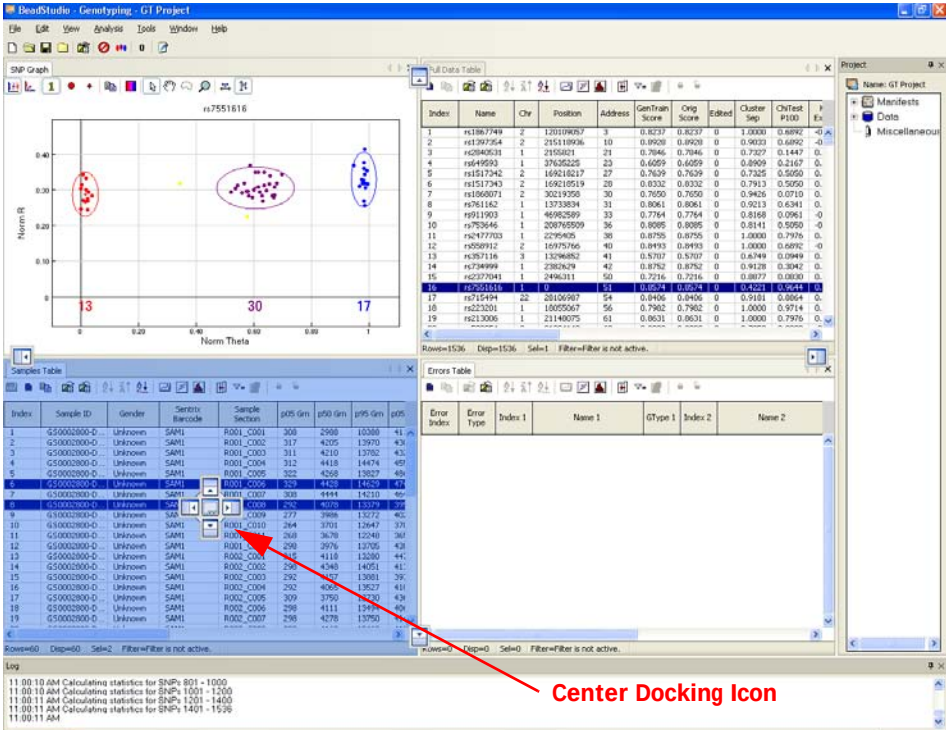

Figure 2-2 Docking a Window Over an Already-Docked Window

- To dock a window to the right of an already-docked window, drag the mouse over the right docking icon  , as shown in Figure 2-3, and release the mouse button.

  The window is now docked to the right of the already-docked window.

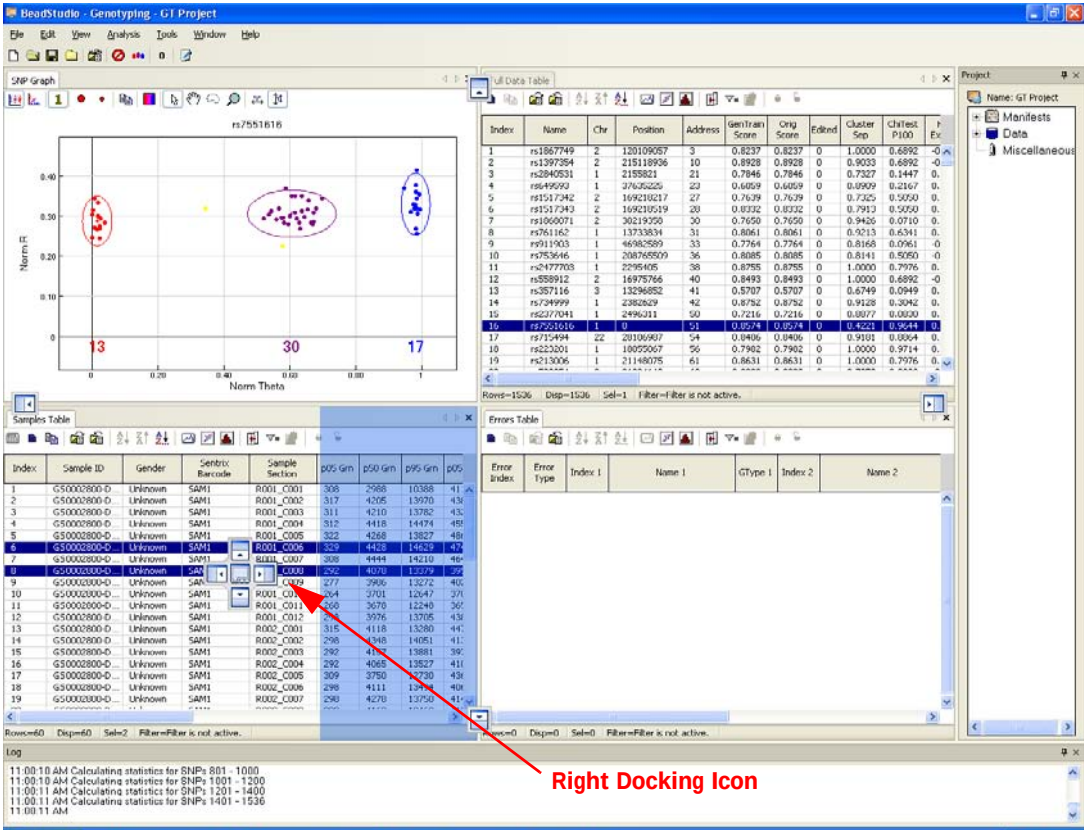

Figure 2-3 Docking a Window to the Right of an Already-Docked Window

- To dock a window to the left of an already-docked window, drag the mouse over the left docking icon  , as shown in Figure 2-3, and release the mouse button.

  The window is now docked to the left of the already-docked 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 2-4, and release the mouse button.

  The window is now docked in an entire side of the Main Window.

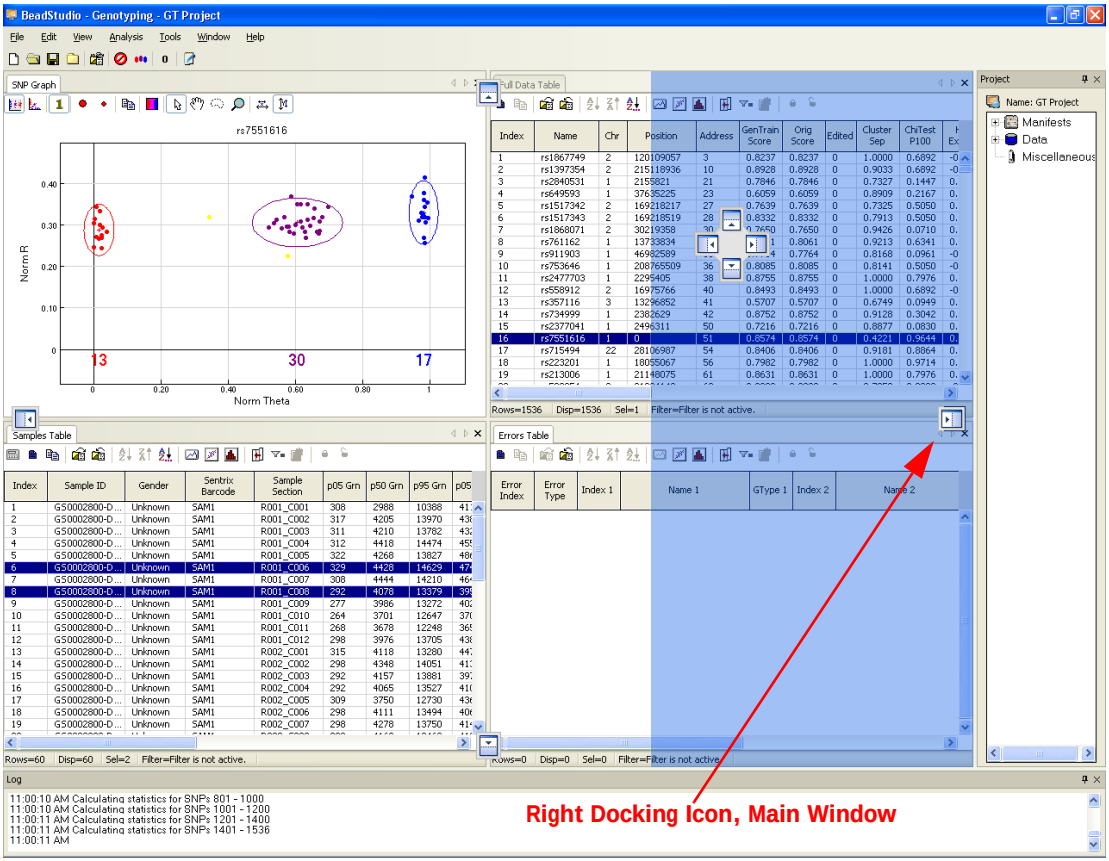

Figure 2-4 Docking a Window to Occupy an Entire Side of the Main Window

NOTE:

Experimentation is the best way to learn how docking works.

Saving Your Window Configuration

When you quit the application, BeadStudio saves your window configuration. The next time you start BeadStudio, the saved window configuration appears.

Reverting Back to the Default Window Configuration

To revert to the default window configuration at any time, select **View | Return to Default** (Figure 2-5).

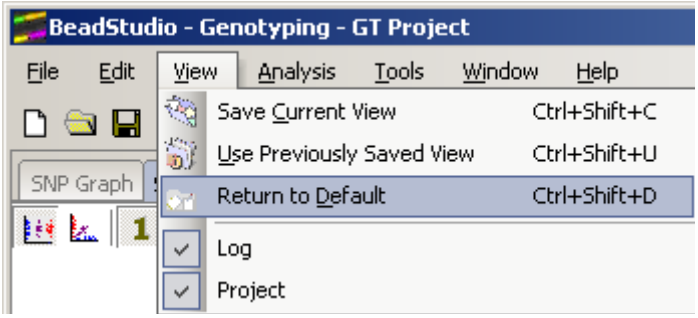

Figure 2-5 Revert to Default Window Configuration

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Chapter 3

Tables

Topics

Introduction

Tables are the centerpiece of BeadStudio's analysis modules. Tables behave the same way across all BeadStudio modules. This chapter describes how to work with tables in BeadStudio to manipulate and analyze data.

Working with Tables

There are two types of tables: those without subcolumns, and those with subcolumns. For example, in the Genotyping module, the **SNP Table** (Figure 3-1) does not have subcolumns. The **Full Data Table** (Figure 3-2) does have subcolumns. Tables with subcolumns have the same subcolumns for every column.

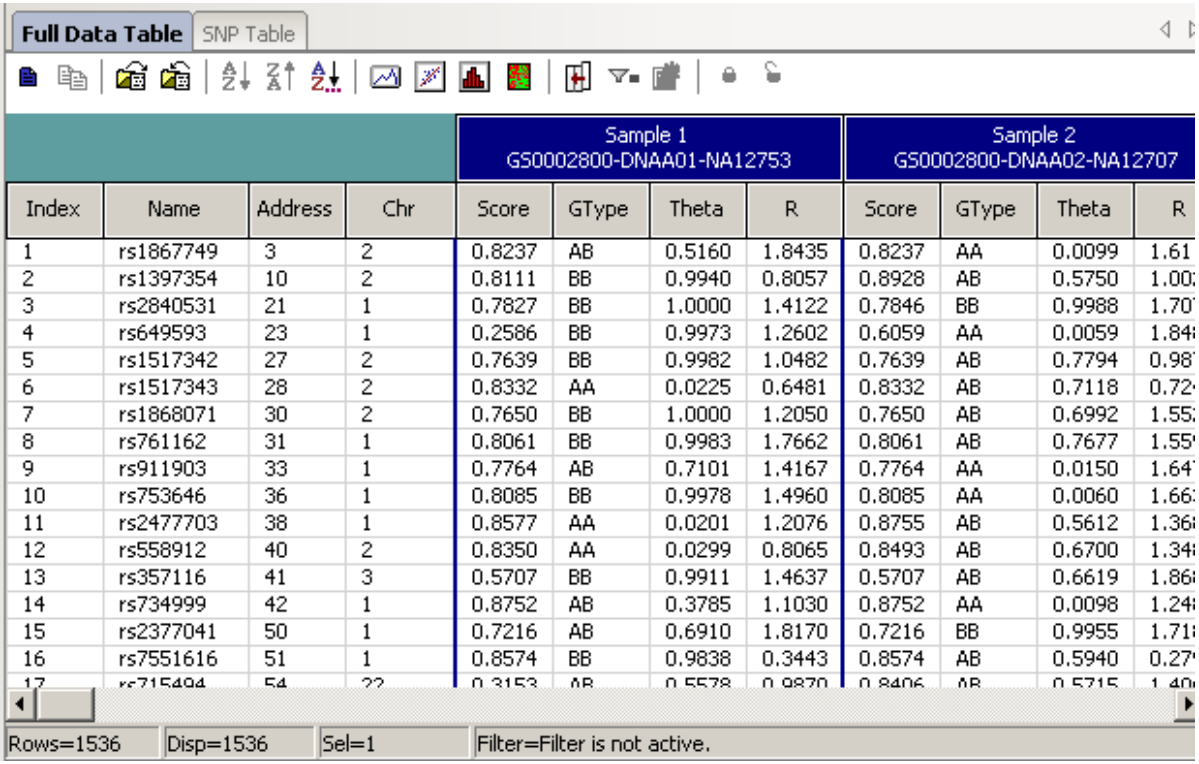
Full Data Table | SNP Table

Index	Name	Chr	Position	Address	GenTrain Score	Orig Score	Edited	Cluster Sep	ChiT P1(
1	rs1867749	2	120109057	3	0.8237	0.8237	0	1.0000	0.7(
2	rs1397354	2	215118936	10	0.8928	0.8928	0	0.9033	0.7
3	rs2840531	1	2155821	21	0.7846	0.7846	0	0.7327	0.0(
4	rs649593	1	37635225	23	0.6059	0.6059	0	0.8909	0.0
5	rs1517342	2	169218217	27	0.7639	0.7639	0	0.7325	0.5
6	rs1517343	2	169218519	28	0.8332	0.8332	0	0.7913	0.5
7	rs1868071	2	30219358	30	0.7650	0.7650	0	0.9426	0.0
8	rs761162	1	13733834	31	0.8061	0.8061	0	0.9213	0.7(
9	rs911903	1	46982589	33	0.7764	0.7764	0	0.8168	0.0(
10	rs753646	1	208765509	36	0.8085	0.8085	0	0.8141	0.8
11	rs2477703	1	2295405	38	0.8755	0.8755	0	1.0000	0.6(
12	rs558912	2	16975766	40	0.8493	0.8493	0	1.0000	0.6
13	rs357116	3	13296852	41	0.5707	0.5707	0	0.6749	0.1
14	rs734999	1	2382629	42	0.8752	0.8752	0	0.9128	0.2
15	rs2377041	1	2496311	50	0.7216	0.7216	0	0.8877	0.0
16	rs7551616	1	0	51	0.8574	0.8574	0	0.4221	0.8
17	rs715494	22	28106987	54	0.8406	0.8406	0	0.9181	0.7(
18	rs223201	1	18055067	56	0.7982	0.7982	0	1.0000	0.8
19	rs213006	1	21148075	61	0.8631	0.8631	0	1.0000	0.4

Rows=1536 | Disp=1536 | Sel=1 | Filter=Filter is not active.

Figure 3-1 Table without Subcolumns

In Figure 3-2, the subcolumns for each sample include **Score**, **GType** (genotype call), and intensity values (**Theta**, **R**).



Full Data Table | SNP Table

				Sample 1 GS0002800-DNAA01-NA12753				Sample 2 GS0002800-DNAA02-NA12707			
Index	Name	Address	Chr	Score	GType	Theta	R	Score	GType	Theta	R
1	rs1867749	3	2	0.8237	AB	0.5160	1.8435	0.8237	AA	0.0099	1.61
2	rs1397354	10	2	0.8111	BB	0.9940	0.8057	0.8928	AB	0.5750	1.00
3	rs2840531	21	1	0.7827	BB	1.0000	1.4122	0.7846	BB	0.9988	1.70
4	rs649593	23	1	0.2586	BB	0.9973	1.2602	0.6059	AA	0.0059	1.84
5	rs1517342	27	2	0.7639	BB	0.9982	1.0482	0.7639	AB	0.7794	0.98
6	rs1517343	28	2	0.8332	AA	0.0225	0.6481	0.8332	AB	0.7118	0.72
7	rs1868071	30	2	0.7650	BB	1.0000	1.2050	0.7650	AB	0.6992	1.55
8	rs761162	31	1	0.8061	BB	0.9983	1.7662	0.8061	AB	0.7677	1.55
9	rs911903	33	1	0.7764	AB	0.7101	1.4167	0.7764	AA	0.0150	1.64
10	rs753646	36	1	0.8085	BB	0.9978	1.4960	0.8085	AA	0.0060	1.66
11	rs2477703	38	1	0.8577	AA	0.0201	1.2076	0.8755	AB	0.5612	1.36
12	rs558912	40	2	0.8350	AA	0.0299	0.8065	0.8493	AB	0.6700	1.34
13	rs357116	41	3	0.5707	BB	0.9911	1.4637	0.5707	AB	0.6619	1.86
14	rs734999	42	1	0.8752	AB	0.3785	1.1030	0.8752	AA	0.0098	1.24
15	rs2377041	50	1	0.7216	AB	0.6910	1.8170	0.7216	BB	0.9955	1.71
16	rs7551616	51	1	0.8574	BB	0.9838	0.3443	0.8574	AB	0.5940	0.27
17	rs715494	54	22	0.3153	AB	0.5578	0.9870	0.8406	AB	0.5715	1.40

Rows=1536 | Disp=1536 | Sel=1 | Filter=Filter is not active.

Figure 3-2 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, or using the arrow keys or the mouse wheel.
- Select multiple contiguous table rows by holding down 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 holding down the **Ctrl** key and clicking the mouse button once on each row you want to select.
- Select all rows by clicking the **Select All Rows** button 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 3-3).

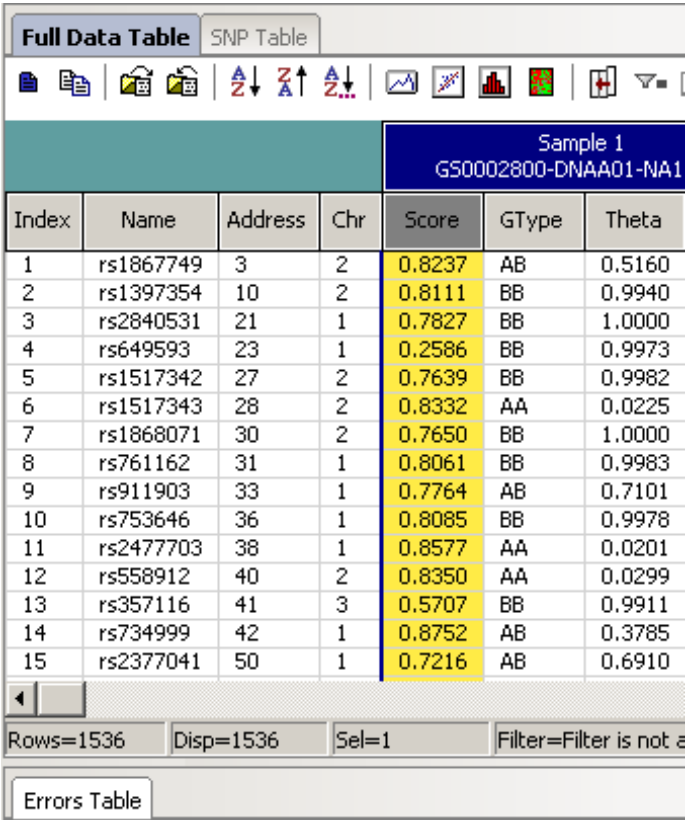


Figure 3-3 Table with a Column Selected

Showing/Hiding Columns

To show or hide columns:

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

   The **Column Chooser** dialog box appears (Figure 3-21).

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

   Alternatively, you can drag-and-drop the columns between the Displayed Columns box and the Hidden Columns box.

Marking Table Rows

To mark table rows:

1. Select the rows you want to mark by one of the following methods:

- For a single row:
  - *Click the row you want to mark.*
- For multiple contiguous rows:
  - *Click the first row in the range you want to mark.*
  - *Press and hold the Shift key.*
  - *Click the last row in the range you want to mark.*
- For multiple non-contiguous rows:
  - *Press and hold the Ctrl key.*
  - *Click each row you want to mark.*

2. Right-click once in the window to bring up the context menu.
3. Select **Configure Marks** (Figure 3-4).

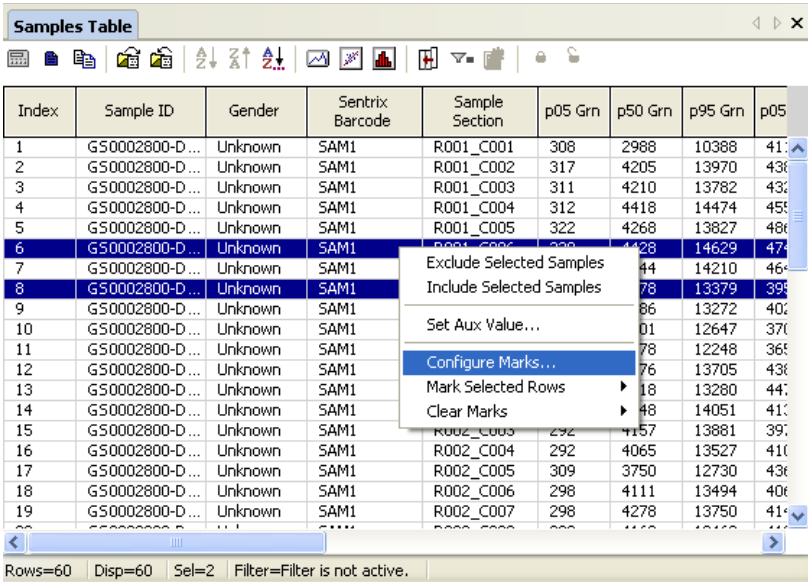


Figure 3-4 Configure Marks Selected

The **Configure Marks** dialog box appears (Figure 3-5).

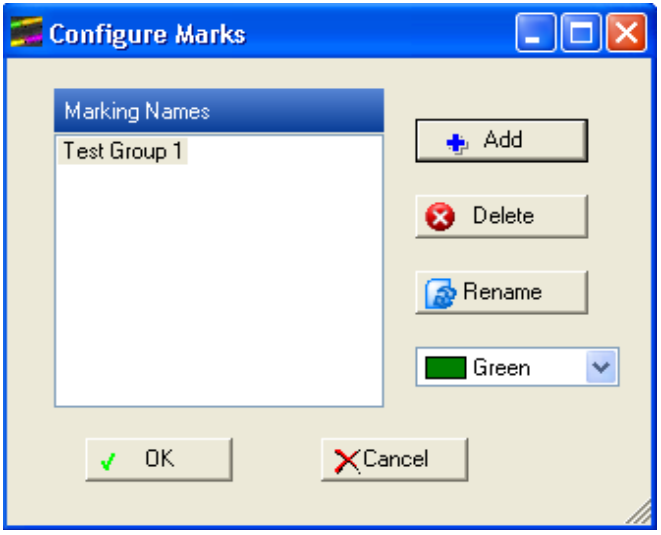


Figure 3-5 Configure Marks Dialog Box

4. Click **Add**.

   The **Select Mark Name** dialog box appears (Figure 3-6).

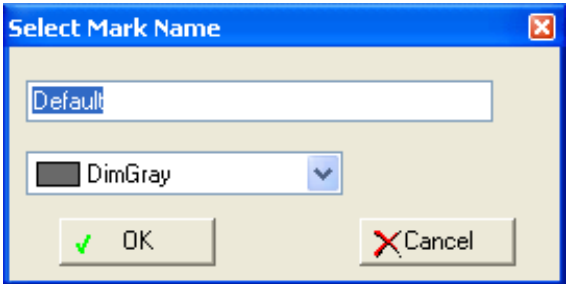


Figure 3-6 Select Mark Name Dialog Box

5. Type a label for your mark in the text box and click **OK**.

   The **Configure Marks** dialog box updates to include the selected mark name (Figure 3-7).

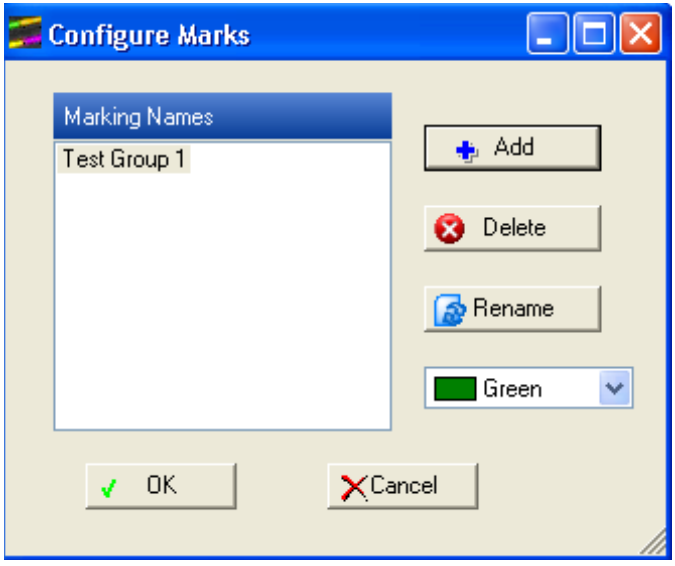
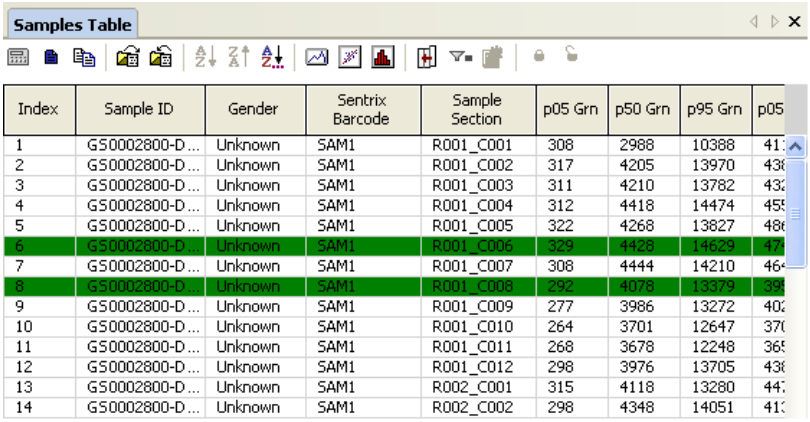


Figure 3-7 Selecting a Color for Your Mark

6. Click once on the label of the new mark you created to select it.
7. Click the color drop-down menu, choose a color for highlighting marked samples in the SNP graph, and release the mouse button.

The marked color you chose (shown in Figure 3-7) overrides the default BeadStudio colors (Figure 3-8).



Index	Sample ID	Gender	Sentrix Barcode	Sample Section	p05 Grn	p50 Grn	p95 Grn	p05
1	GS0002800-D...	Unknown	SAM1	R001_C001	308	2988	10388	41
2	GS0002800-D...	Unknown	SAM1	R001_C002	317	4205	13970	43
3	GS0002800-D...	Unknown	SAM1	R001_C003	311	4210	13782	43
4	GS0002800-D...	Unknown	SAM1	R001_C004	312	4418	14474	45
5	GS0002800-D...	Unknown	SAM1	R001_C005	322	4268	13827	48
6	GS0002800-D...	Unknown	SAM1	R001_C006	329	4428	14629	47
7	GS0002800-D...	Unknown	SAM1	R001_C007	308	4444	14210	46
8	GS0002800-D...	Unknown	SAM1	R001_C008	292	4078	13379	39
9	GS0002800-D...	Unknown	SAM1	R001_C009	277	3986	13272	40
10	GS0002800-D...	Unknown	SAM1	R001_C010	264	3701	12647	37
11	GS0002800-D...	Unknown	SAM1	R001_C011	268	3678	12248	36
12	GS0002800-D...	Unknown	SAM1	R001_C012	298	3976	13705	43
13	GS0002800-D...	Unknown	SAM1	R002_C001	315	4118	13280	44
14	GS0002800-D...	Unknown	SAM1	R002_C002	298	4348	14051	41

Figure 3-8 Selected Rows Marked

Clearing Marks

To clear marked colors:

1. Select **Clear Marks** from the **Samples Table** context menu (Figure 3-9).

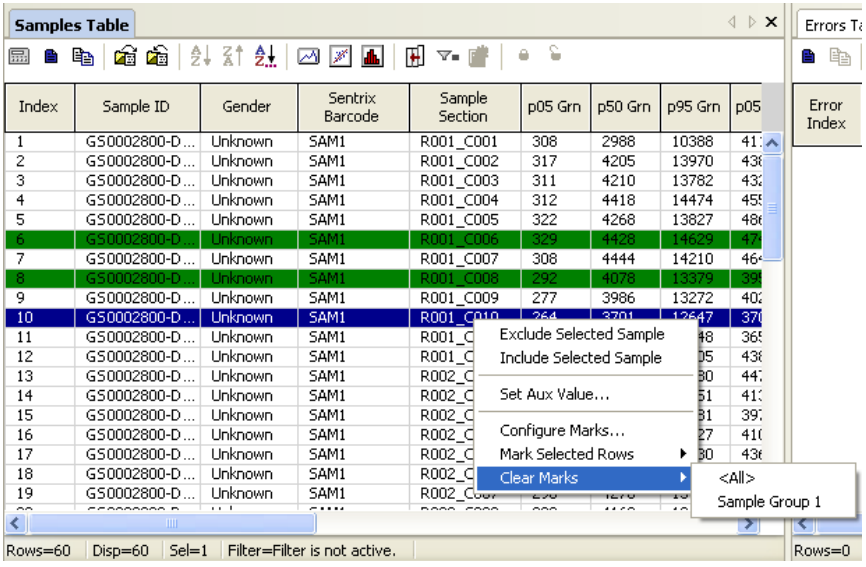


Figure 3-9 Clear Marks Selected

2. Choose whether you would like to clear all marks or a specific mark.

   The selected marks are removed and the rows display with BeadStudio default colors.

Exporting to a File

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

To export a column:

1. Click **Export to File**.

   The **Save As** dialog box appears.

2. In the **File Name** text box, enter a name for the file.

3. Click **Save**.

   The file is saved and the **Would you like to view this file?** dialog box appears.

4. Do one of the following:

- Click **Yes** if you would like to view the file now.
- Click **No** if you do not want to view the file.

Importing a Column

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

To import a column:

1. Click **Import Columns** .

   The **Import** dialog box appears.

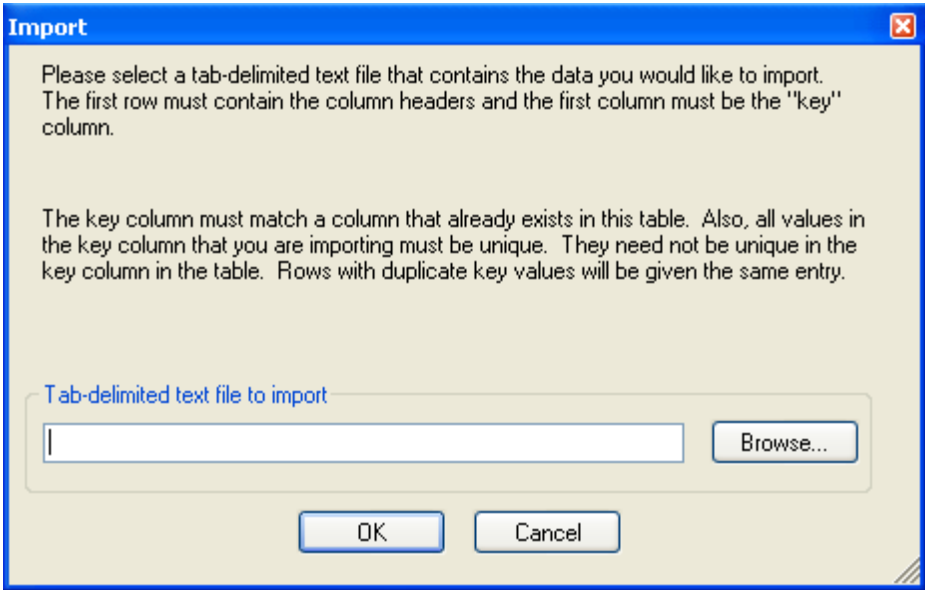


Figure 3-10 Import Dialog Box

2. Read the instructions in the Import dialog box.
3. Click **Browse**.
4. Browse to the tab-delimited file you want to import.

5. Click **Open**.
6. Click **OK**.

   The imported column displays in the left side of the table window.

NOTE:

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

Sorting by Column

You can sort a column by selecting the column and clicking the button to sort it in ascending order or the button to sort it in descending order. You can also click the button to bring up the **Sort** dialog box (Figure 3-11), which allows you to prioritize sorting by multiple columns.

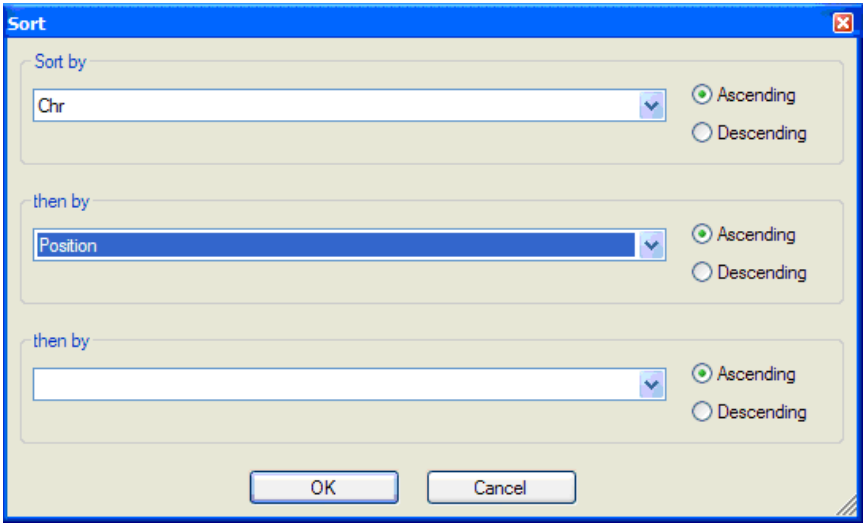


Figure 3-11 Multiple Column Sort Dialog Box

Creating a New Subcolumn

To create a new subcolumn:

1. Click the **New subcolumn** button f( ) .

   The **New Column** dialog box appears.

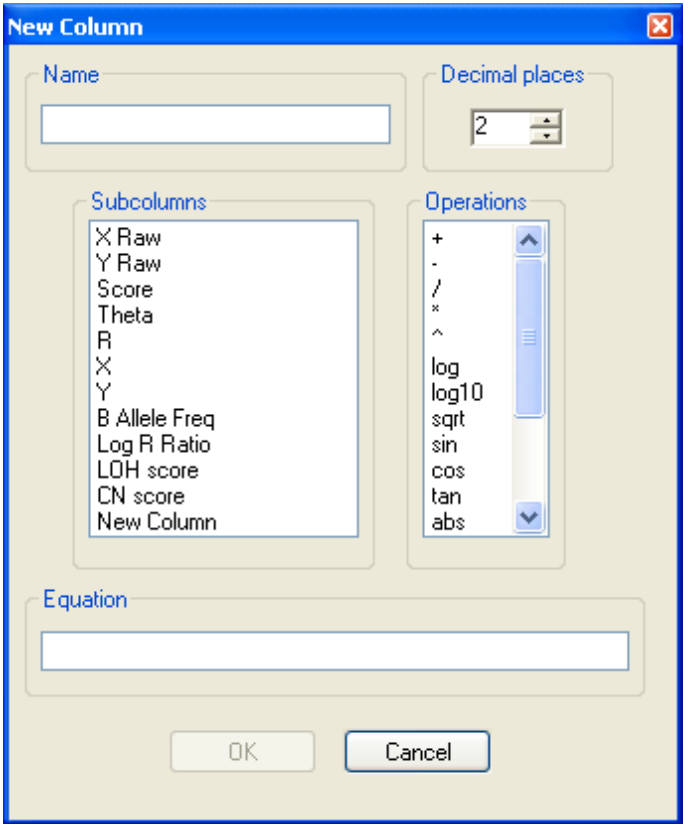


Figure 3-12 New Column Dialog Box

2. Type a name for your new subcolumn in the **Name** text box.
3. Select or type the number of decimal places you want to display in the new subcolumn in the **Decimal places** combo box.
4. Build your equation by clicking the subcolumns and operations you want (Table 3-1).

Table 3-1 New Sub-Column, Operations and Descriptions

Operations	Descriptions
+	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
ceiling	Smallest integer that is greater than or equal to x
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

Your equation appears in the **Equation** text box. (Figure 3-13).

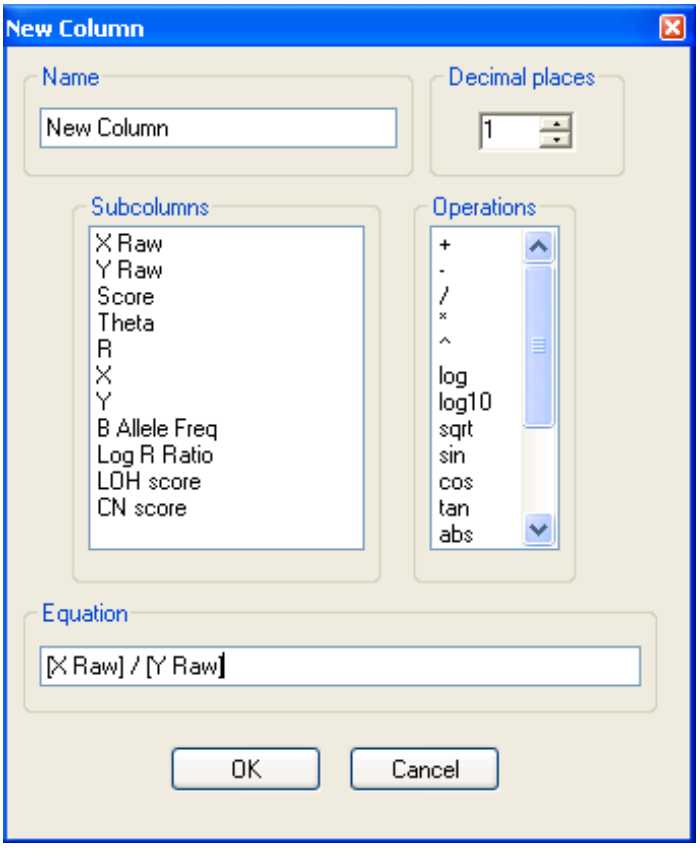


Figure 3-13 New Column Dialog Box with Selections

5. Click **OK**.

   The new subcolumn is added to the table for every sample.

Edit View Analysis Tools Window Help

Data Table | SNP Table

				Sample 1 GS0002800-DNAA01-NA12753					Sample 2 GS0002800-DNAA02-NA12707				
lex	Name	Address	Chr	Score	GType	Theta	R	New Column	Score	GType	Theta	R	New Column
	rs1867749	3	2	0.8237	AB	0.5160	1.8435	0.8	0.8237	AA	0.0099	1.6118	20.7
	rs1397354	10	2	0.8111	BB	0.9940	0.8057	0.0	0.8928	AB	0.5750	1.0027	0.7
	rs2840531	21	1	0.7827	BB	1.0000	1.4122	0.0	0.7846	BB	0.9988	1.7071	0.0
	rs649593	23	1	0.2586	BB	0.9973	1.2602	0.0	0.6059	AA	0.0059	1.8484	26.0
	rs1517342	27	2	0.7639	BB	0.9982	1.0482	0.0	0.7639	AB	0.7794	0.9870	0.3
	rs1517343	28	2	0.8332	AA	0.0225	0.6481	9.3	0.8332	AB	0.7118	0.7242	0.4
	rs1868071	30	2	0.7650	BB	1.0000	1.2050	0.0	0.7650	AB	0.6992	1.5524	0.4
	rs761162	31	1	0.8061	BB	0.9983	1.7662	0.0	0.8061	AB	0.7677	1.5597	0.3
	rs911903	33	1	0.7764	AB	0.7101	1.4167	0.4	0.7764	AA	0.0150	1.6473	17.4
	rs753646	36	1	0.8085	BB	0.9978	1.4960	0.0	0.8085	AA	0.0060	1.6636	24.8
	rs2477703	38	1	0.8577	AA	0.0201	1.2076	12.1	0.8755	AB	0.5612	1.3688	0.7
	rs558912	40	2	0.8350	AA	0.0299	0.8065	8.8	0.8493	AB	0.6700	1.3485	0.5
	rs357116	41	3	0.5707	BB	0.9911	1.4637	0.0	0.5707	AB	0.6619	1.8687	0.5
	rs734999	42	1	0.8752	AB	0.3785	1.1030	1.2	0.8752	AA	0.0098	1.2481	18.8
	rs2377041	50	1	0.7216	AB	0.6910	1.8170	0.4	0.7216	BB	0.9955	1.7182	0.0
	rs7551616	51	1	0.8574	BB	0.9838	0.3443	0.1	0.8574	AB	0.5940	0.2793	0.7

Figure 3-14 New Subcolumn Added to the Table for Every Sample

Filtering Rows

You can filter your table to display only rows that meet certain criteria.

To filter your table:

1. Click the **Filter Rows** button .

   The **Filter Table Rows** dialog box appears (Figure 3-15).

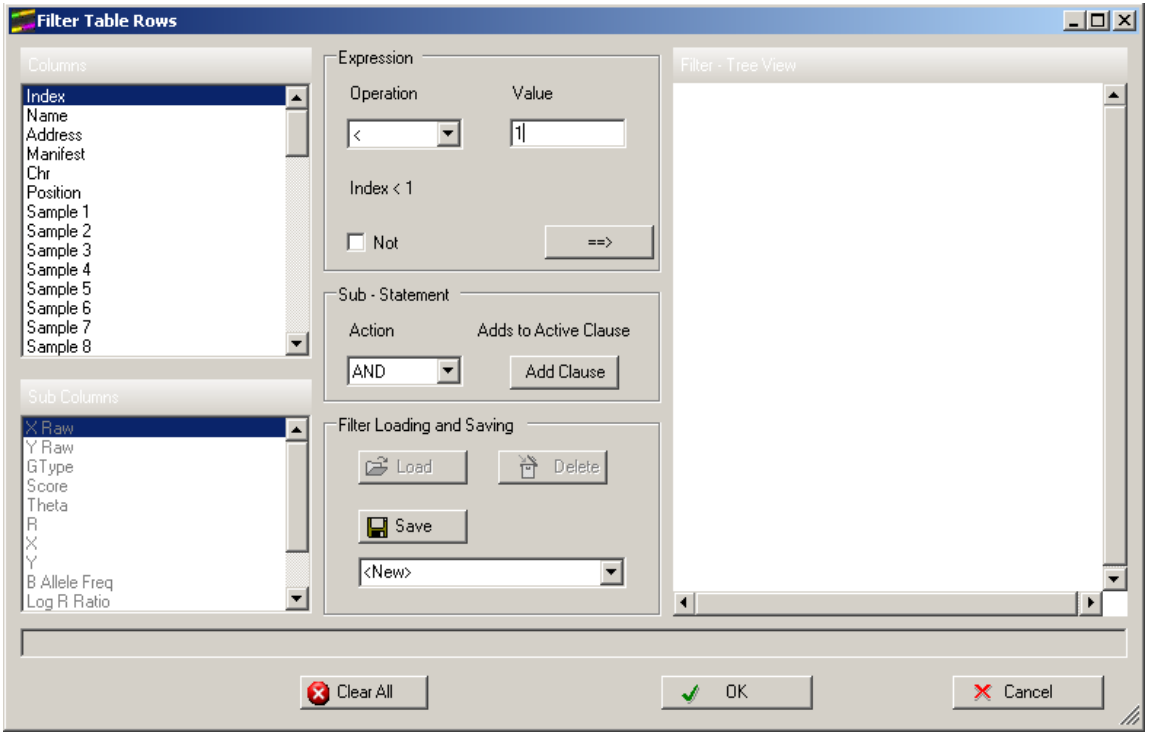


Figure 3-15 Filter Table Rows Dialog Box

2. Select a column. If the column you have selected has subcolumns, also select a subcolumn.
3. Select an operation from the **Operation** drop-down menu.
4. Enter a value in the **Value** text box.
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**.

NOTE:

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

8. Click **OK**.

NOTE:

An alternate way of filtering table rows in some tables is to select one or more rows and right-click. Choose **Show Only Selected Rows** from the popup 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 box.
4. Click ==> .

The dialog box should look similar to Figure 3-16.

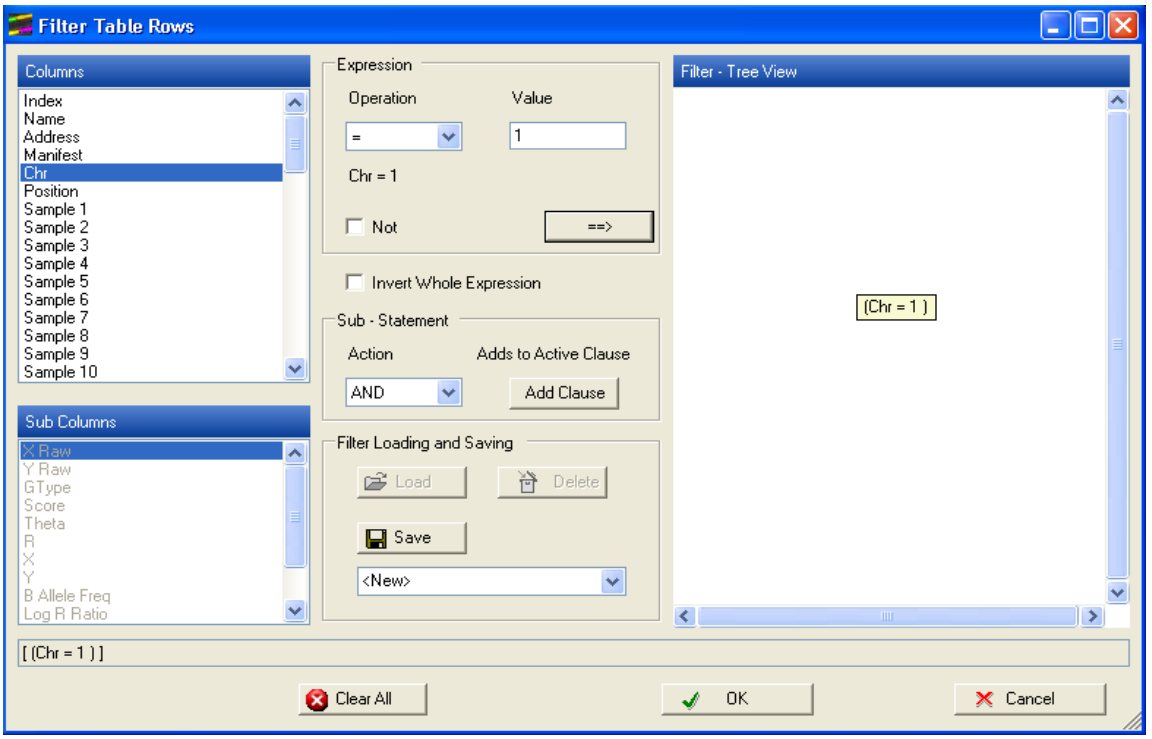


Figure 3-16 Creating a Simple Filter

Creating a More Complex Filter

You can also create more complex boolean expressions.

For example, you can select for 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** pull-down 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 3-17).

7. Select **Sample 1** in the **Columns** list box.
8. Select **GType** in the **Sub Columns** list box.
9. Type **AA** in the **Value** text box.
10. Click ==> .
11. Select **Sample 2** in the **Columns** list box.
12. Select **Score** in the **Sub Columns** list box.
13. Select **>** in the **Operation** pull down menu.
14. Type **0.5** in the **Value** text box.
15. Click ==> .

The dialog box should look similar to Figure 3-17.

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)]]**.

NOTE:

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.

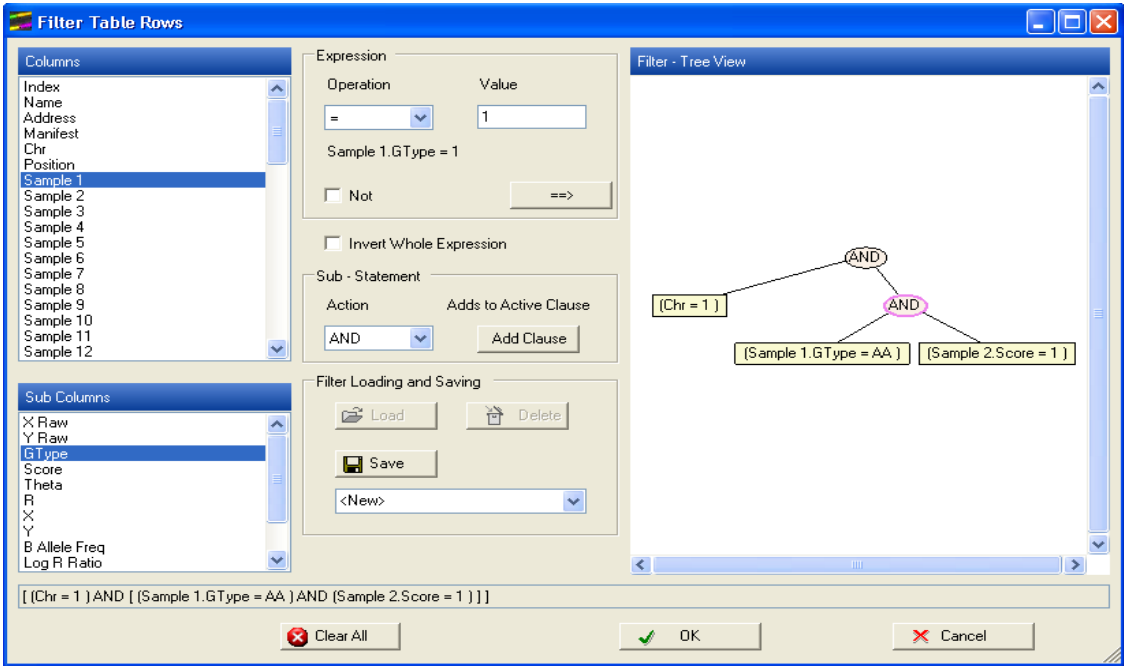


Figure 3-17 Creating a More Complex Filter

Saving a Filter

Filters can be saved and used later in different projects.

To save a filter to use later:

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

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

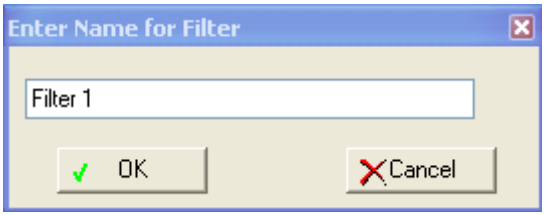


Figure 3-18 Enter Name for Filter Dialog Box

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

NOTE:

To assure 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>

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** dialog box, select the filter you want to use in the dropdown menu.

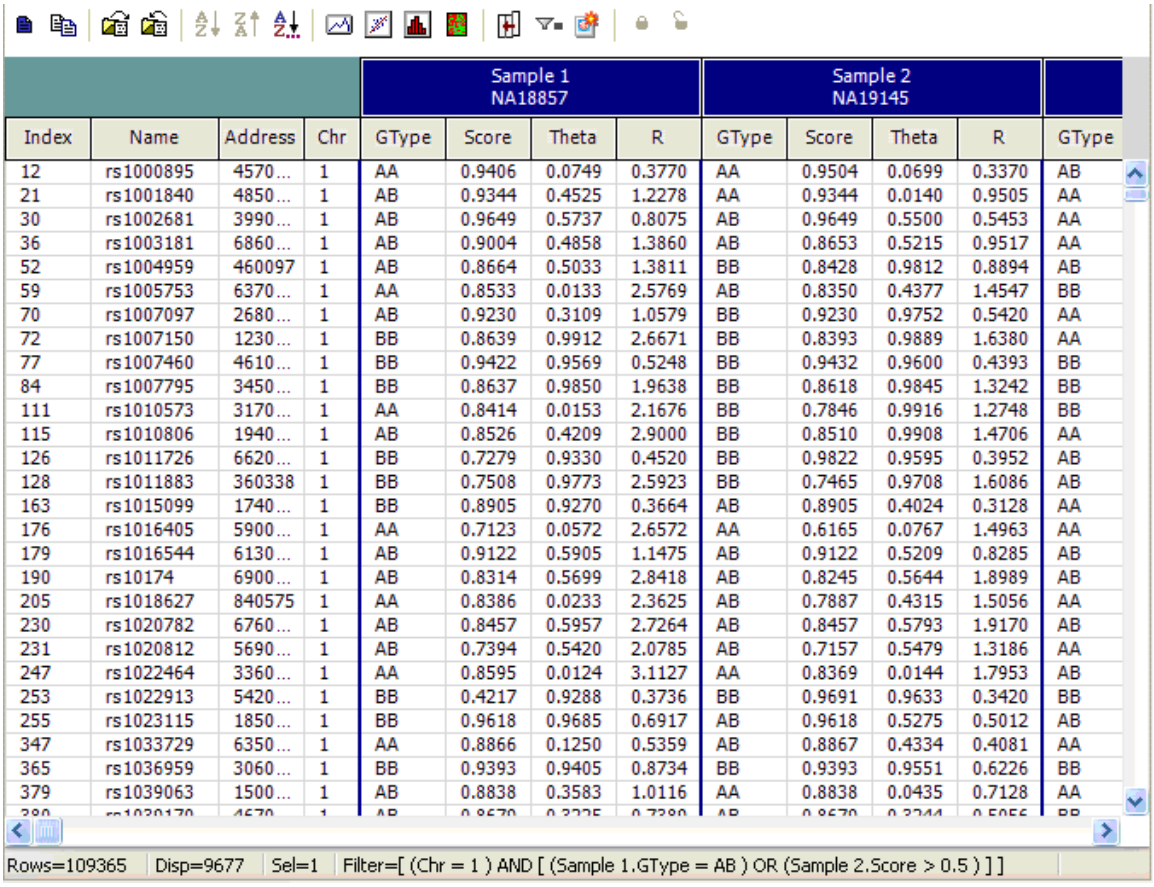
   The **Load** button becomes active.

2. Click **Load**.

   Your filter is loaded and ready to use.

Results of Filtering

Figure 3-19 shows the results of the example filter shown in Figure 3-17. The filter formula is displayed in the status bar of the table, and the number of rows that passed the filter is displayed.



				Sample 1 NA18857				Sample 2 NA19145				
Index	Name	Address	Chr	GType	Score	Theta	R	GType	Score	Theta	R	GType
12	rs1000895	4570...	1	AA	0.9406	0.0749	0.3770	AA	0.9504	0.0699	0.3370	AB
21	rs1001840	4850...	1	AB	0.9344	0.4525	1.2278	AA	0.9344	0.0140	0.9505	AA
30	rs1002681	3990...	1	AB	0.9649	0.5737	0.8075	AB	0.9649	0.5500	0.5453	AA
36	rs1003181	6860...	1	AB	0.9004	0.4858	1.3860	AB	0.8653	0.5215	0.9517	AA
52	rs1004959	460097	1	AB	0.8664	0.5033	1.3811	BB	0.8428	0.9812	0.8894	AB
59	rs1005753	6370...	1	AA	0.8533	0.0133	2.5769	AB	0.8350	0.4377	1.4547	BB
70	rs1007097	2680...	1	AB	0.9230	0.3109	1.0579	BB	0.9230	0.9752	0.5420	AA
72	rs1007150	1230...	1	BB	0.8639	0.9912	2.6671	BB	0.8393	0.9889	1.6380	AA
77	rs1007460	4610...	1	BB	0.9422	0.9569	0.5248	BB	0.9432	0.9600	0.4393	BB
84	rs1007795	3450...	1	BB	0.8637	0.9850	1.9638	BB	0.8618	0.9845	1.3242	BB
111	rs1010573	3170...	1	AA	0.8414	0.0153	2.1676	BB	0.7846	0.9916	1.2748	BB
115	rs1010806	1940...	1	AB	0.8526	0.4209	2.9000	BB	0.8510	0.9908	1.4706	AA
126	rs1011726	6620...	1	BB	0.7279	0.9330	0.4520	BB	0.9822	0.9595	0.3952	AB
128	rs1011883	360338	1	BB	0.7508	0.9773	2.5923	BB	0.7465	0.9708	1.6086	AB
163	rs1015099	1740...	1	BB	0.8905	0.9270	0.3664	AB	0.8905	0.4024	0.3128	AA
176	rs1016405	5900...	1	AA	0.7123	0.0572	2.6572	AA	0.6165	0.0767	1.4963	AA
179	rs1016544	6130...	1	AB	0.9122	0.5905	1.1475	AB	0.9122	0.5209	0.8285	AB
190	rs10174	6900...	1	AB	0.8314	0.5699	2.8418	AB	0.8245	0.5644	1.8989	AB
205	rs1018627	840575	1	AA	0.8386	0.0233	2.3625	AB	0.7887	0.4315	1.5056	AA
230	rs1020782	6760...	1	AB	0.8457	0.5957	2.7264	AB	0.8457	0.5793	1.9170	AB
231	rs1020812	5690...	1	AB	0.7394	0.5420	2.0785	AB	0.7157	0.5479	1.3186	AA
247	rs1022464	3360...	1	AA	0.8595	0.0124	3.1127	AA	0.8369	0.0144	1.7953	AB
253	rs1022913	5420...	1	BB	0.4217	0.9288	0.3736	BB	0.9691	0.9633	0.3420	BB
255	rs1023115	1850...	1	BB	0.9618	0.9685	0.6917	AB	0.9618	0.5275	0.5012	AB
347	rs1033729	6350...	1	AA	0.8866	0.1250	0.5359	AB	0.8867	0.4334	0.4081	AA
365	rs1036959	3060...	1	BB	0.9393	0.9405	0.8734	BB	0.9393	0.9551	0.6226	BB
379	rs1039063	1500...	1	AB	0.8838	0.3583	1.0116	AA	0.8838	0.0435	0.7128	AA

Figure 3-19 Results of Filtering

Clearing a Filter

To clear the filter:

- Click the **Clear Filter** button .

  The filter is cleared.

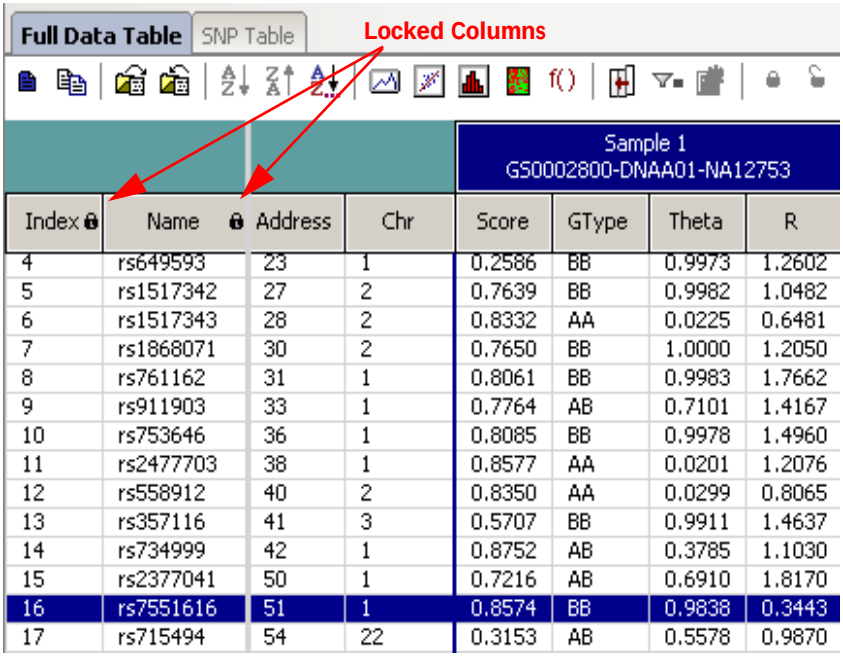
Filtering Notes

- Leaves are added to the currently selected node (highlighted in pink).
- To change the active node, click another node.
- You can change the action for a node by right-clicking the node and selecting the action from the pop-up 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** dialog box.
- You can delete a node or a leaf by right-clicking it and selecting **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 3-20).



Index	Name	Address	Chr	Score	GType	Theta	R
4	rs649593	23	1	0.2586	BB	0.9973	1.2602
5	rs1517342	27	2	0.7639	BB	0.9982	1.0482
6	rs1517343	28	2	0.8332	AA	0.0225	0.6481
7	rs1868071	30	2	0.7650	BB	1.0000	1.2050
8	rs761162	31	1	0.8061	BB	0.9983	1.7662
9	rs911903	33	1	0.7764	AB	0.7101	1.4167
10	rs753646	36	1	0.8085	BB	0.9978	1.4960
11	rs2477703	38	1	0.8577	AA	0.0201	1.2076
12	rs558912	40	2	0.8350	AA	0.0299	0.8065
13	rs357116	41	3	0.5707	BB	0.9911	1.4637
14	rs734999	42	1	0.8752	AB	0.3785	1.1030
15	rs2377041	50	1	0.7216	AB	0.6910	1.8170
16	rs7551616	51	1	0.8574	BB	0.9838	0.3443
17	rs715494	54	22	0.3153	AB	0.5578	0.9870

Figure 3-20 Table with Two Locked 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 3-21).

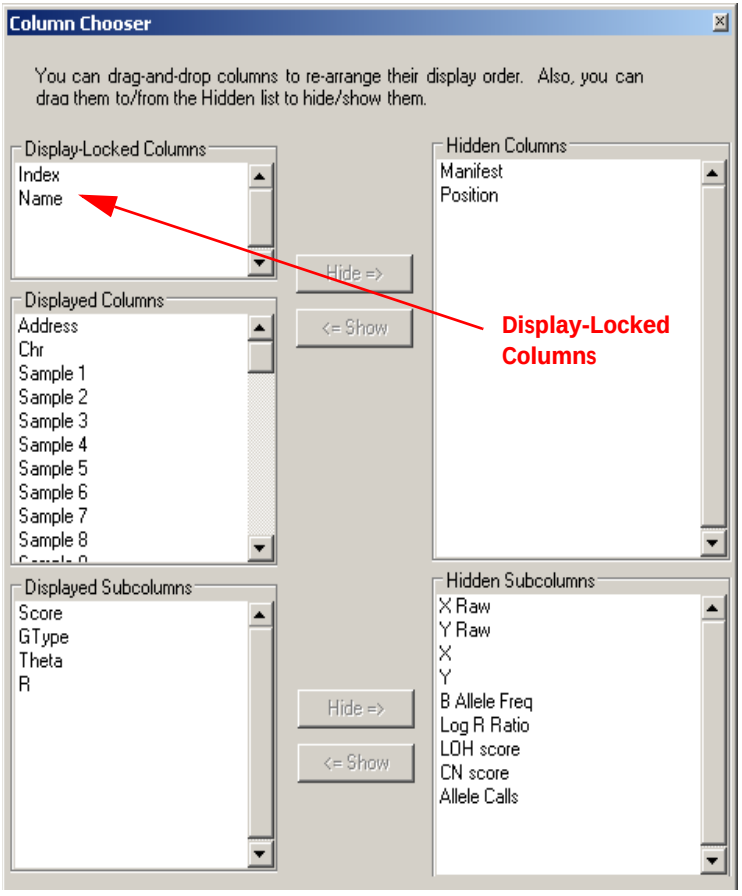


Figure 3-21 Column Chooser Window, Display-Locked Columns

NOTES

Chapter 4

Graphs

Topics

Introduction

BeadStudio's graphing functions allow you to plot data displayed in the analysis module tables in a number of ways.

Histograms

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

1. Click the **Histogram** button  in the table toolbar.

   The **Plot Columns** dialog box opens with **Histogram** selected (Figure 4-1).

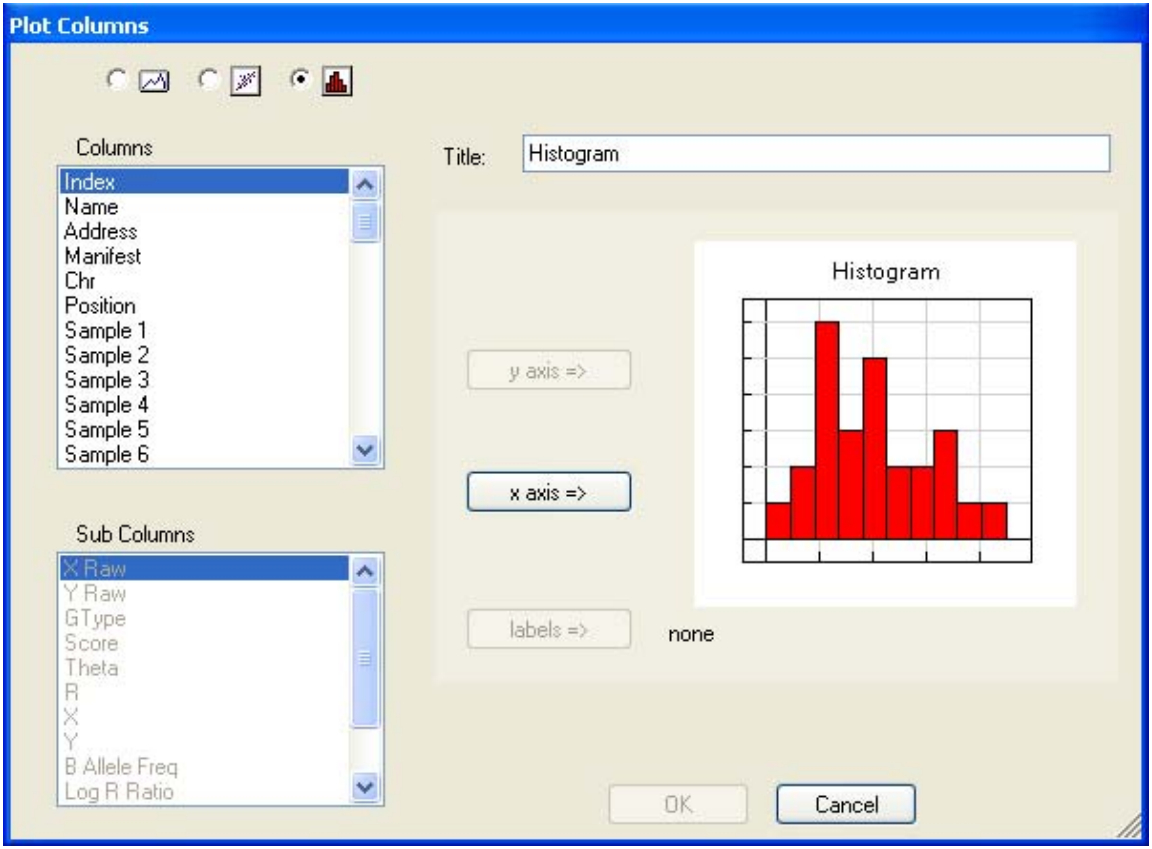


Figure 4-1 Plot Columns Dialog Box with Histogram Selected

2. Choose graphing options from the **Columns** and **Sub Columns** list boxes.
3. Click **OK**.

   The histogram appears with the parameters you selected (3.).

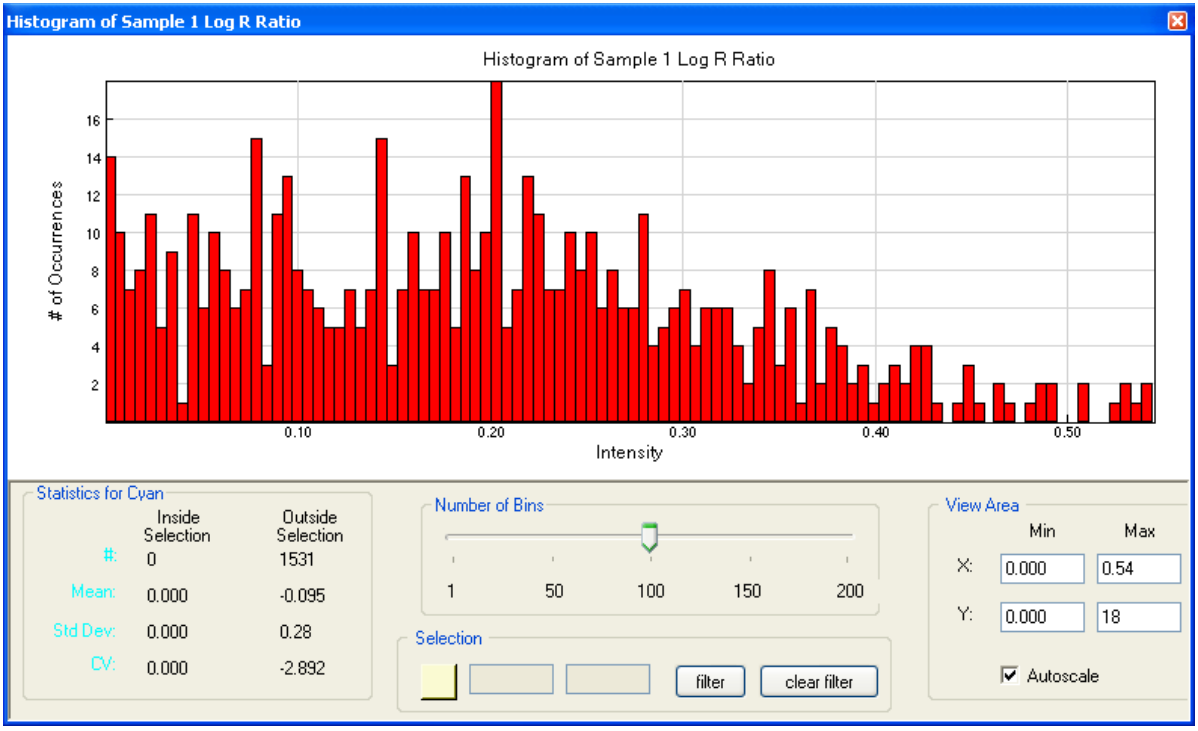


Figure 4-2 Histogram

Line Plots

Use the **Line Plot** option to plot data from a single column.

1. Click the **Line Plot** button in the table to open the **Plot Columns** dialog box with **Line Plot** selected (Figure 4-3).

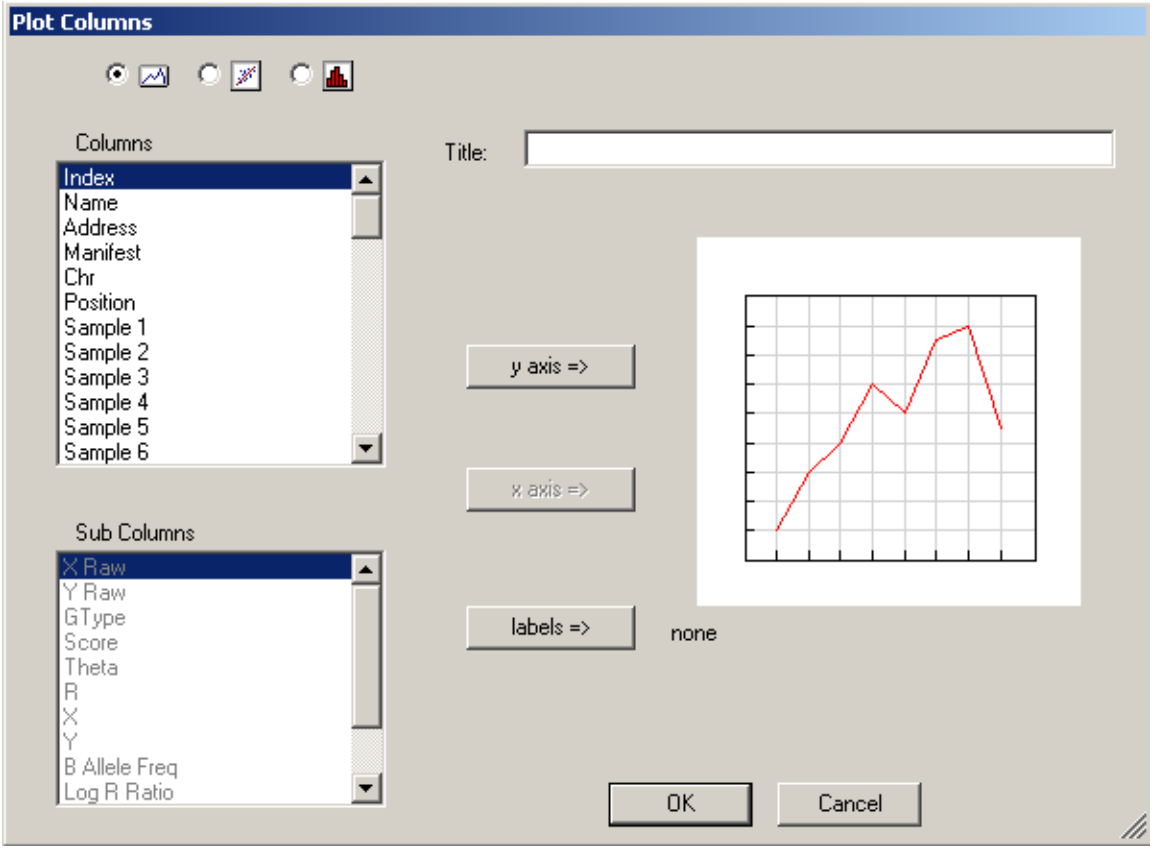


Figure 4-3 Plot Columns Dialog Box with Line Plot Selected

2. Choose graphing options from the **Columns** and **Sub Columns** list boxes.
3. Click **OK**.

   The line plot appears with the parameters you selected (Figure 4-6).

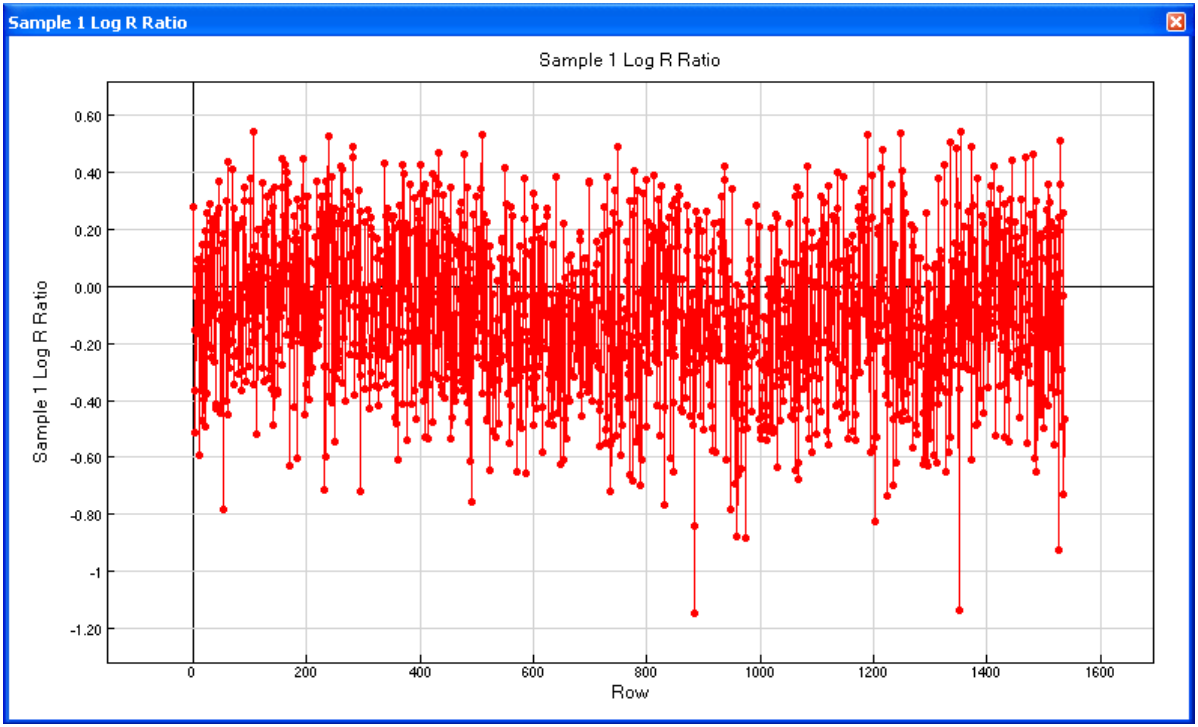


Figure 4-4 Line Plot

Scatter Plots

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

1. Click the **Scatter Plots** button in the table to open the **Plot Columns** dialog box with **Scatter Plots** selected (Figure 4-5).

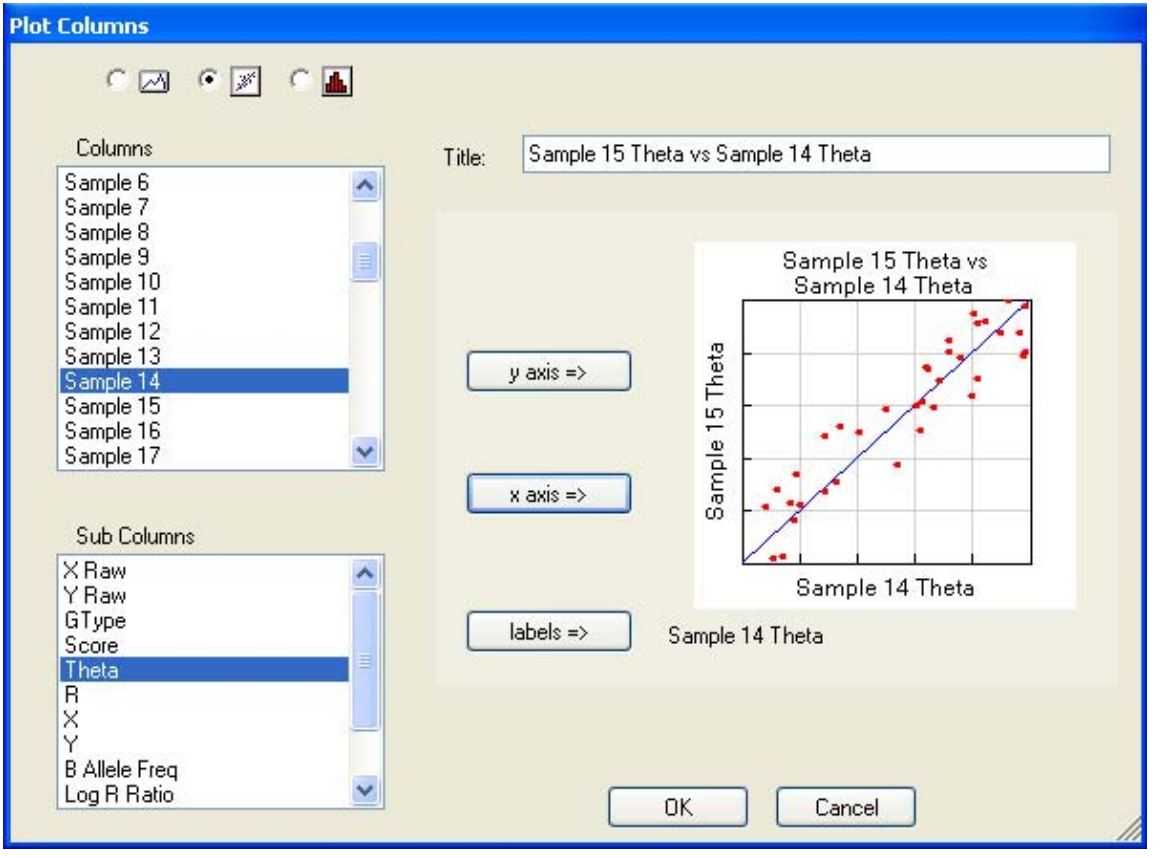


Figure 4-5 Plot Columns Dialog Box with Scatter Plot Selected

2. Choose graphing options from the **Columns** and **Sub Columns** list boxes and click **OK**.

   The scatter plot appears with the parameters you selected (Figure 4-6).

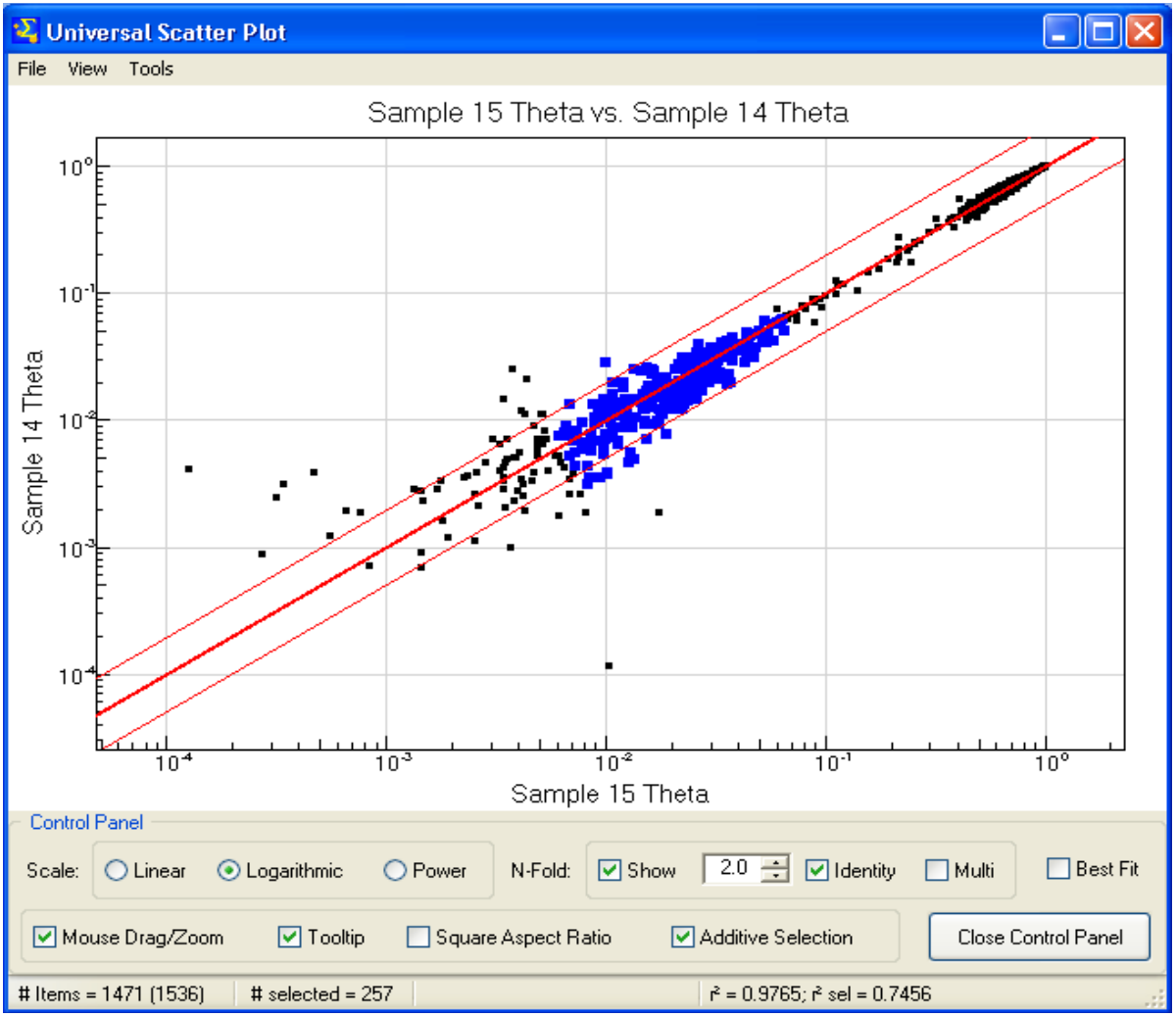


Figure 4-6 Universal Scatter Plot

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 the **Heat Map** button  in the toolbar of a table with subcolumns to open the **Plot Sample Sub-Columns 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 the **Heat Map** button  .

   The **Plot Sample Sub-Columns in a Heat Map** dialog box appears (Figure 4-7).

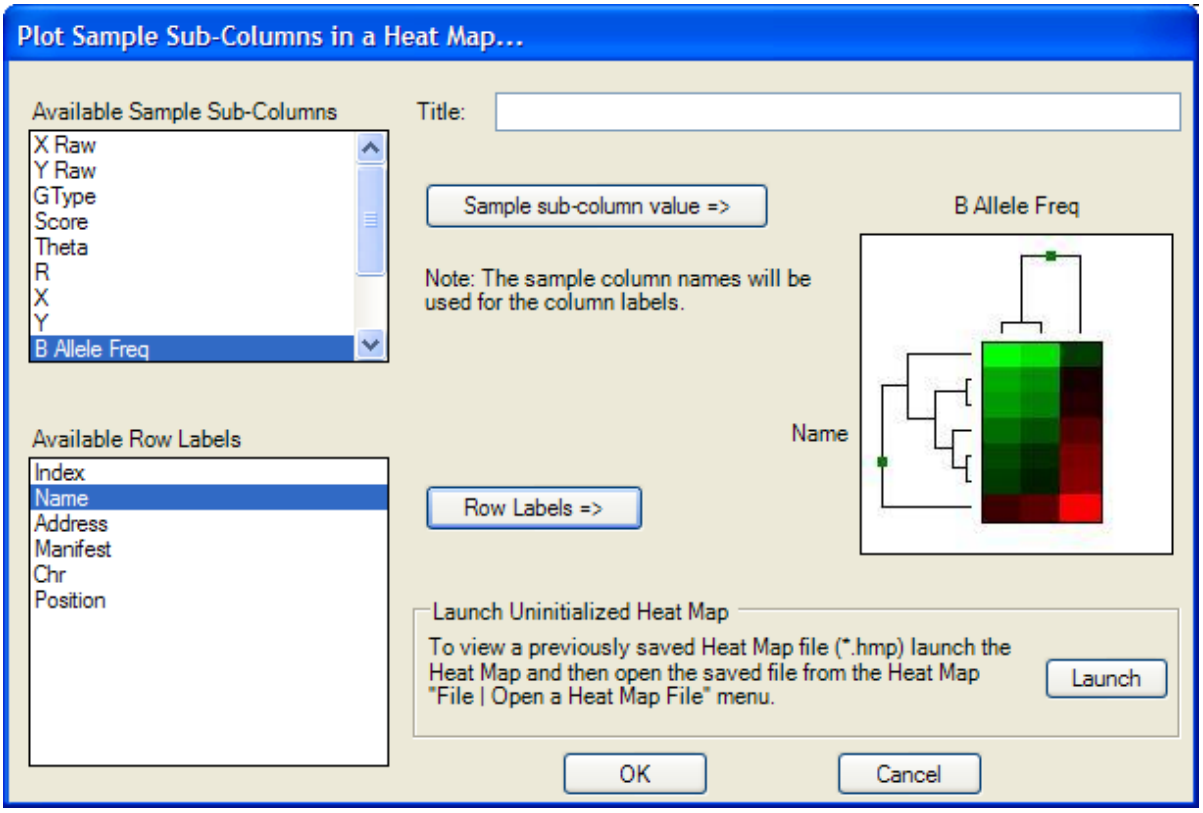


Figure 4-7 Plot Sample Sub-Columns in a Heat Map Dialog Box

2. In the **Title** text box, enter a title for your heat map.
3. In the **Available Sample Sub-Columns** listbox, select a data series you wish to map in a heat map.
4. Click **Sample sub-column 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 become the row labels of the heat map.
6. Click **Row Labels**.

7. Click **OK**.

   The heat map displays with the row labels and data series you selected (Figure 4-8).

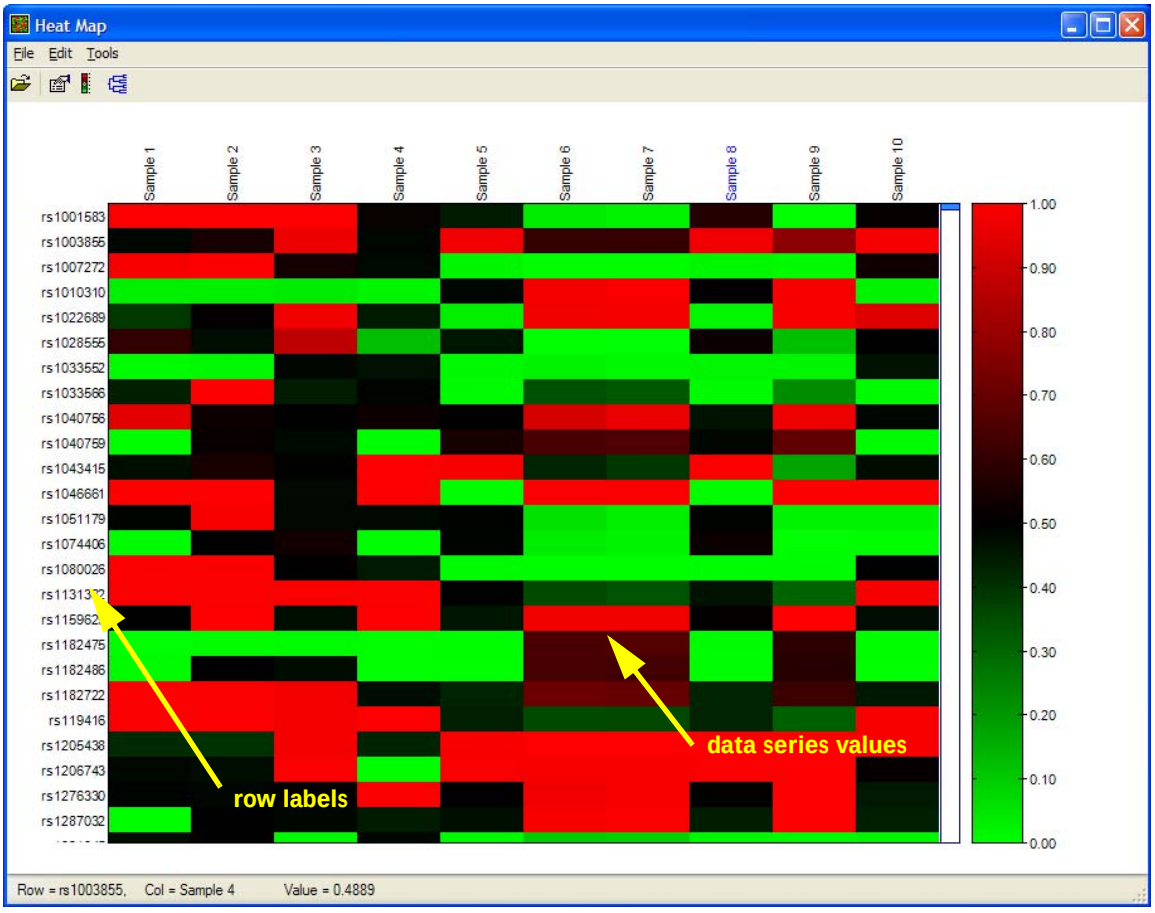


Figure 4-8 Heat Map with Row Labels and Data Series

Clustering Heat Map Data

To cluster heat map data:

1. In the **Cluster Options** window (Figure 4-9), select one or both of the following:

   - **Cluster Rows**
   - **Cluster Columns (usually samples)**

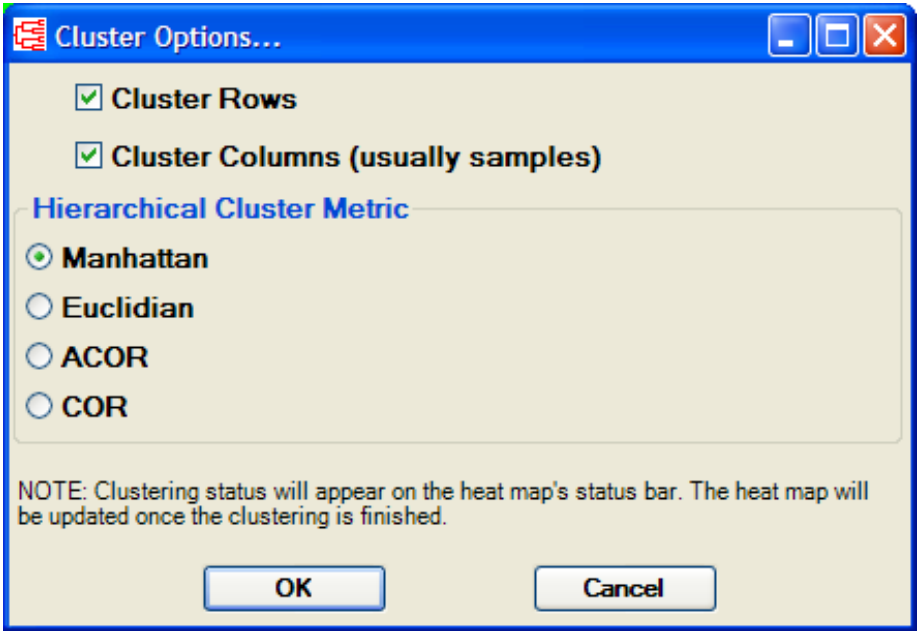


Figure 4-9 Cluster Options Dialog Box

2. 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.

3. 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 4-12).

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 $X = \{X_1, X_2, \ldots X_N\}$ and $Y = \{Y_1, Y_2, \ldots Y_N\}$ 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 4-10).

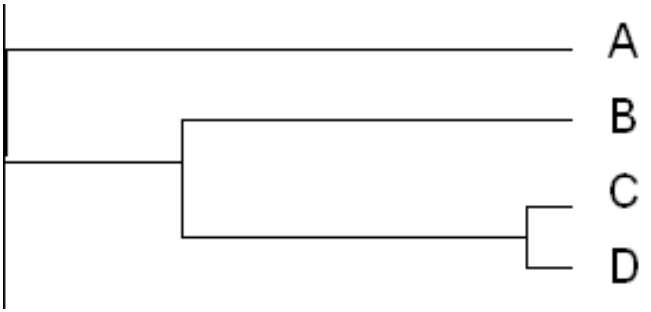


Figure 4-10 Dendrogram, Similarity Example

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 4-11).

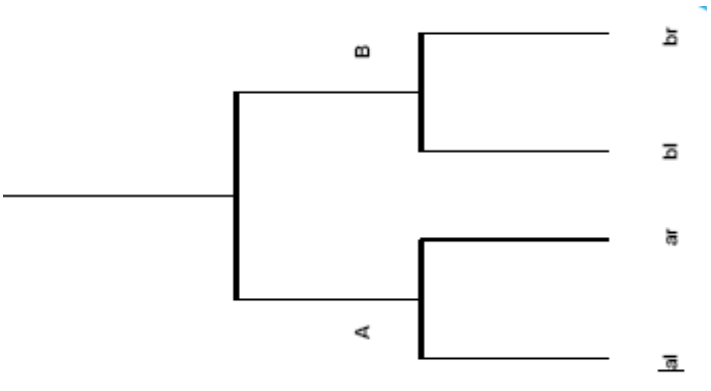


Figure 4-11 Dendrogram, Showing Nodes

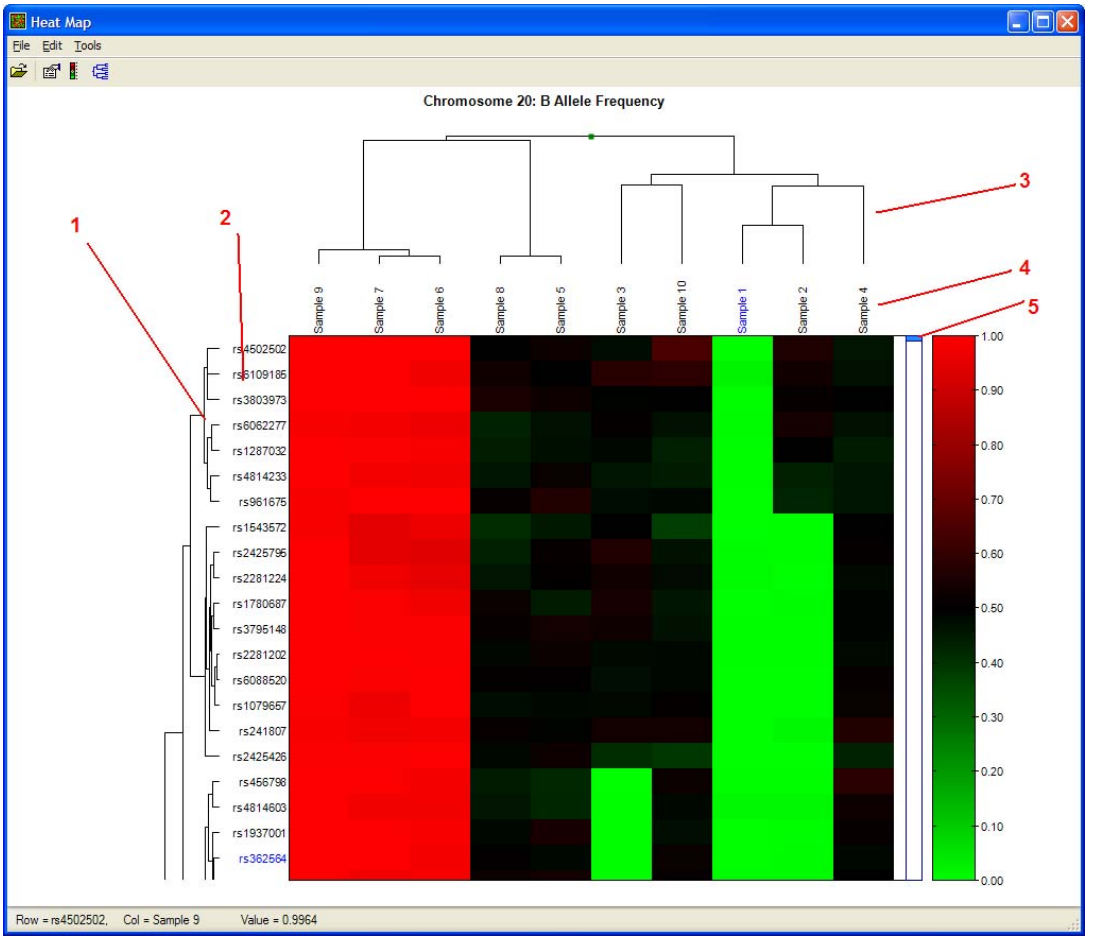


Figure 4-12 Heat Map, Display and Scrolling Properties

Resizing the Row Tree Cluster

The row tree cluster is shown in Figure 4-12, callout 1.

To resize the row tree cluster:

1. Click and hold the **Shift** button.
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 4-12, callout 2.

To change the width of the row label area:

1. Click and hold the **Shift** button.
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.

To change the height of the row label area, do one of the following:

- Using the mouse wheel:

1. Position the cursor over the row label area.
2. Move the mouse wheel up or down.

- Using the mouse button:

1. Position the cursor over the row label area.
2. 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 4-12, callout 3.

To resize the column tree cluster:

1. Click and hold the **Shift** button.
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 4-12, callout 4.

To change the height of column labels:

1. Click and hold the **Shift** button.
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, and 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 4-12, callout 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 on the scroll bar and hold, and 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 displays.

Saving a Heat Map

To save a heat map to a file:

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

   The **Save Heat Map to File** dialog box appears.
2. Browse to the location where you want to save your heat map.
3. Type a name for your heat map in the **File Name** text box.
4. Click **Save**.

   Your heat map is saved in the location you specified.

Exiting the Heat Map Window

To exit the heat map window, select **File | Exit**.

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

When you select **File | Exit**, the Unsaved Data dialog box appears.

Click **Yes** to save the clustered heat map.

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 the **Edit Heat Map Properties** button  .

  The **Heat Map Properties** dialog box appears (Figure 4-13).

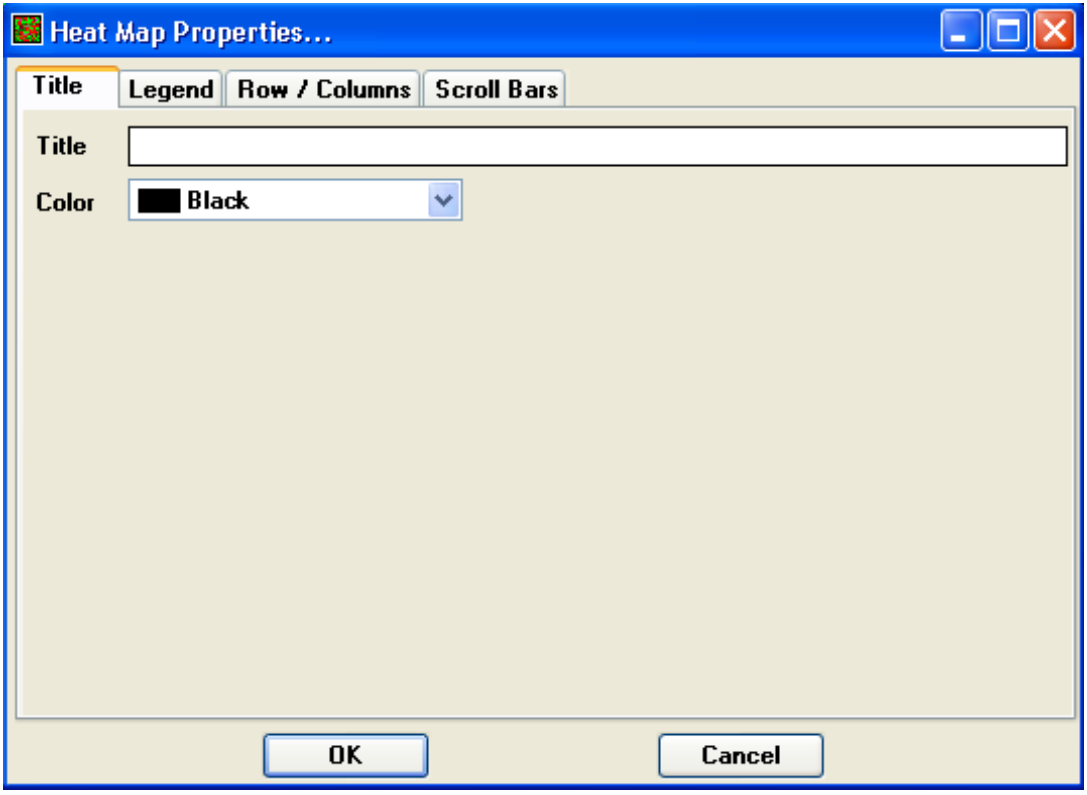


Figure 4-13 Heat Map Properties, Title Tab

2. In the **Title** tab, enter a title for your heat map in the **Title** text box.
3. Optionally, choose a color for the heat map title by selecting a color from the **Color** pull-down menu.
4. Click **OK**.

   The heat map displays 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 4-14).

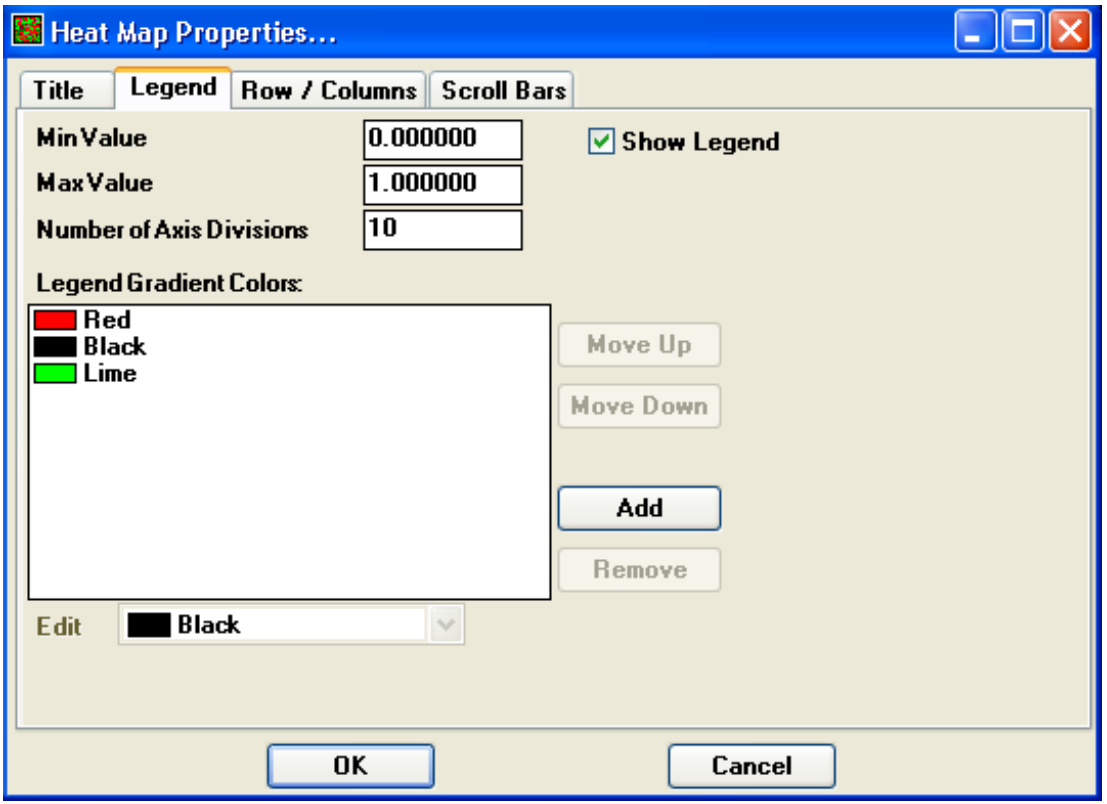


Figure 4-14 Heat Map Properties, Legend Tab

2. Enter a **Min Val**, **Max Val**, and **Number of Axis Divisions**.

Showing/Hiding the Legend

To show the legend with the heat map, check the **Show Legend** checkbox (Figure 4-14). To hide the legend, clear the **Show Legend** checkbox.

Changing the Legend Gradient Colors

The default Legend Gradient Colors are **Red**, **Black**, and **Lime**.

1. To change the legend gradient colors, select different colors from the **Edit** pull-down menu (Figure 4-14).
2. To change the assignment of the colors, Click a color in the **Legend Gradient Colors** area. Click **Move Up** or **Move Down** to reassign the color.
3. To add a color to the heat map, click **Add**.

   The color dialog box appears.
4. Choose a color from the drop-down menu.
5. Click **OK**.

   The color you chose is added to the heat map.
6. To remove a color from the heat map, click **Remove**.

   The color you chose is removed from the heat map.
7. Click **OK** to apply your changes.

Row/Column Properties

You can change heat map row and column properties (Figure 4-15) to suit your preferences.

Showing/Hiding Labels

- To show row labels, check the **Show labels** box in the **Rows** area.
- To hide row labels, clear the **Show labels** box in the **Rows** area.
- To show column labels, check the **Show labels** box in the **Columns** area.
- To hide column labels, clear the **Show labels** box in the **Columns** area.

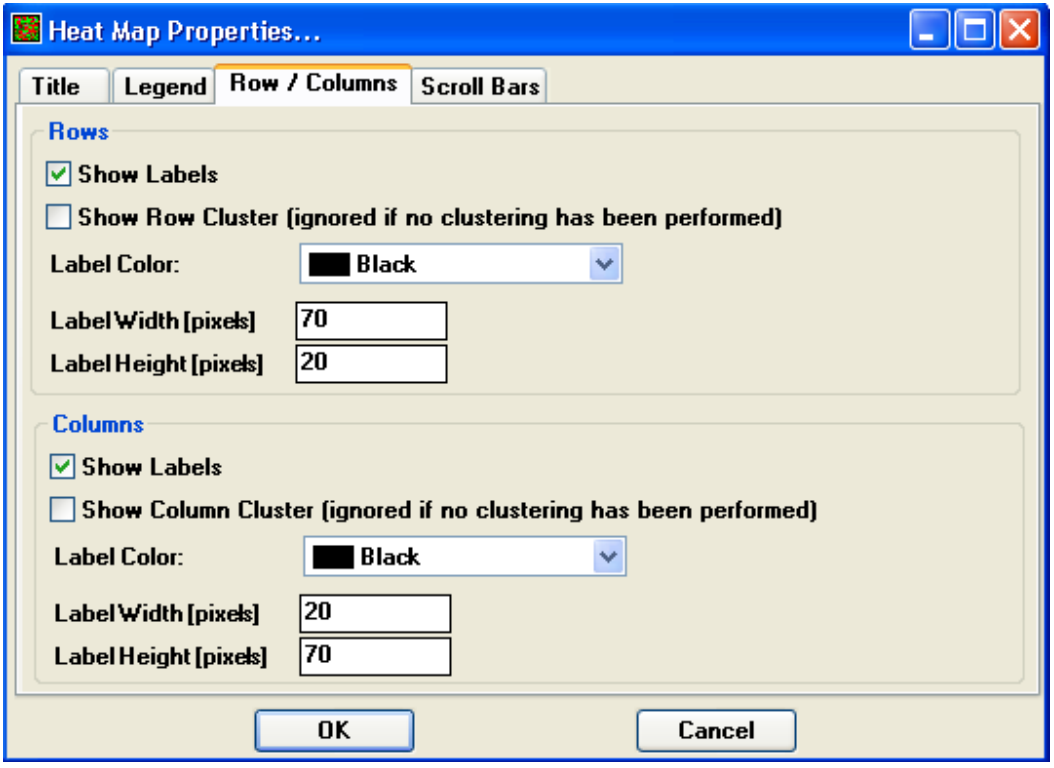


Figure 4-15 Heat Map Properties, Row / Columns Tab

Showing/Hiding Clusters

- To show the row cluster, check the **Show Row Cluster** box.
- To hide the row cluster, clear the **Show Row Cluster** box.
- To show the column cluster, check the **Show Column Cluster** box.
- To hide the column cluster, clear the **Show Column Cluster** box.

Changing the Label Color

To change the label color for rows or columns:

- Select a new color from the corresponding **Label Color** drop-down 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 box.
- Enter a new width in the corresponding **Label Width** text box.

Scroll Bar Properties

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

To change scroll bar properties (Figure 4-16), select new colors using the **Scroll Area Color**, **Scroll Bar Color**, and **Scroll Border Color** drop-down menus.

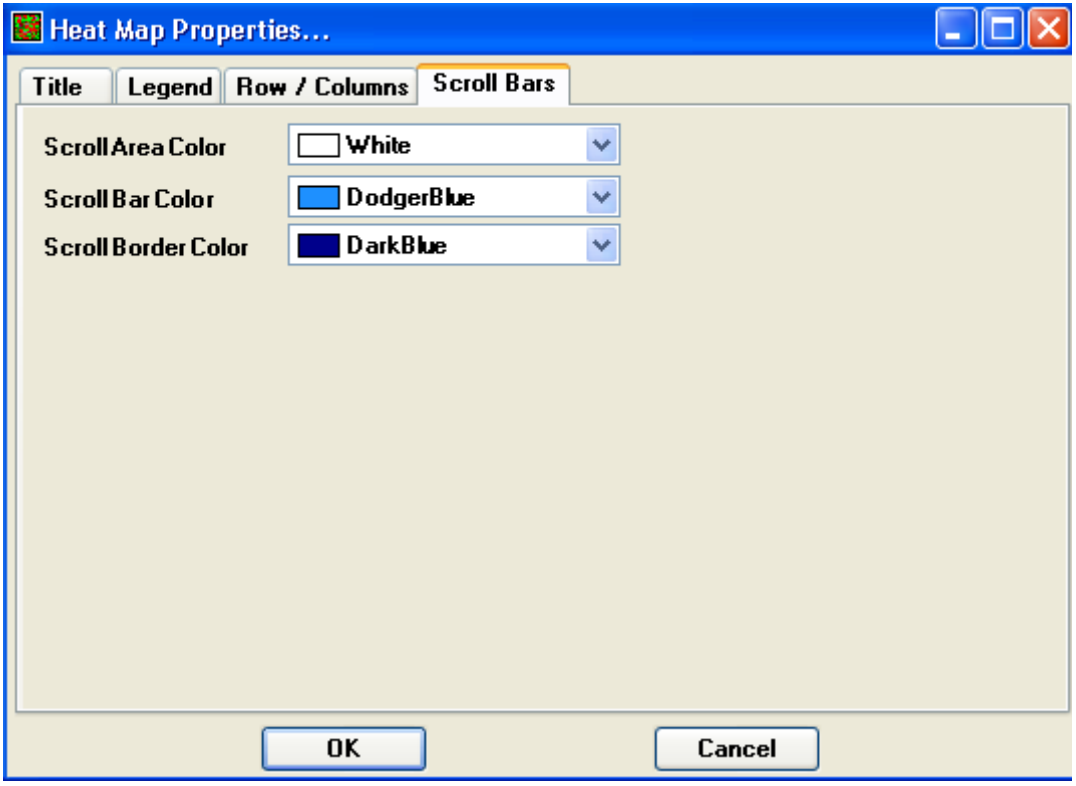


Figure 4-16 Heat Map Properties, Scroll Bars Tab

Creating a Presentation Image

To create a presentation image:

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

   The **Presentation Image Setup** dialog box appears (Figure 4-17).

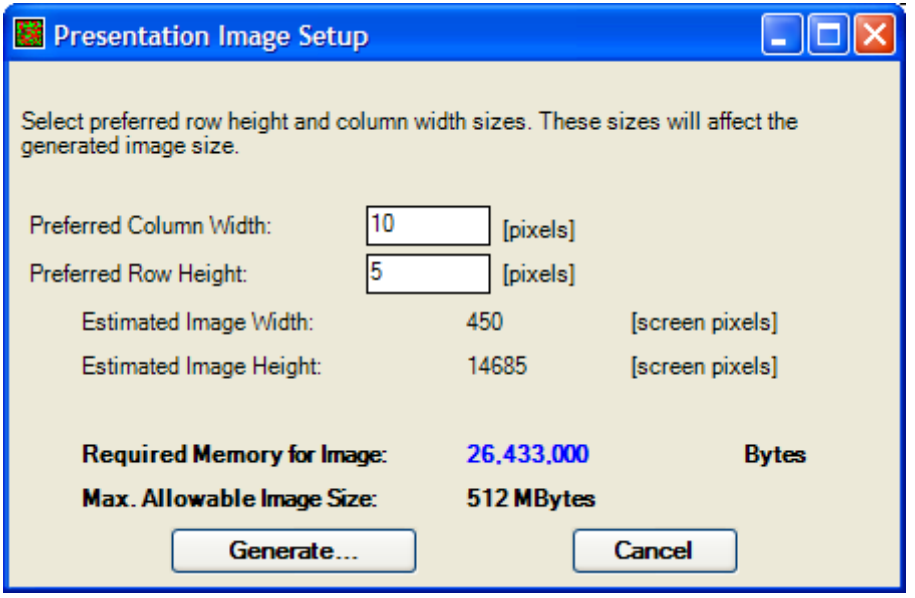


Figure 4-17 Presentation Image Setup Dialog Box

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 dialog 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 4-18).

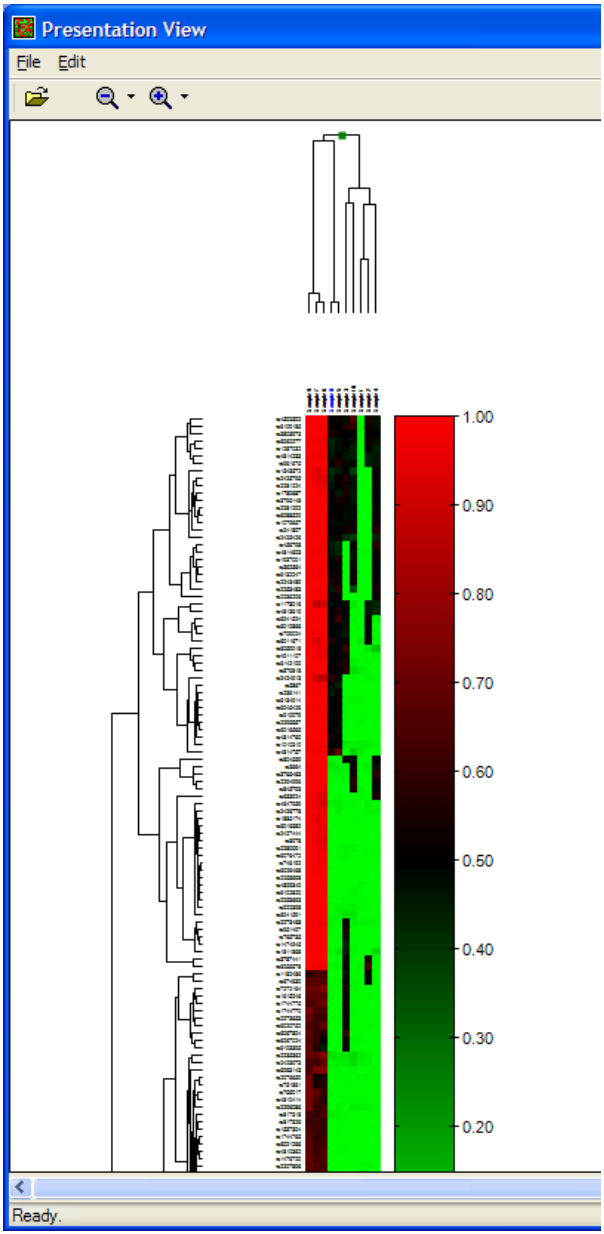


Figure 4-18 Heat Map, Presentation View

NOTES

Chapter 5

Visualization Tools

Topics

Introduction

This chapter describes how to use BeadStudio visualization tools such as the Illumina Genome Viewer (IGV) and the Illumina Chromosome Browser (ICB).

Using the Illumina Genome Viewer

The IGV allows you to visualize your data on a genome-wide scale.

Launch the IGV by selecting **Analysis | Show columns in Genome Viewer**.

Figure 5-1 below shows the IGV main window. IGV can display up to four plots at a time (a single data series each) over any chosen chromosome.

In addition to using the IGV, you can browse your data at the chromosomal level using **Illumina Chromosome Browser** (ICB) and/or the **Zoom Plot**.

Launch the ICB by double-clicking on a chromosome in the Genome Viewer (Figure 5-1, callout 1). For more information about the ICB, see *Using the Illumina Chromosome Browser* on page 5-11.

Launch the Zoom Plot by double-clicking on any plot in the Genome Viewer (Figure 5-1, callout 2). For more information about the Zoom Plot, see *Zoom Plot* on page 5-10.

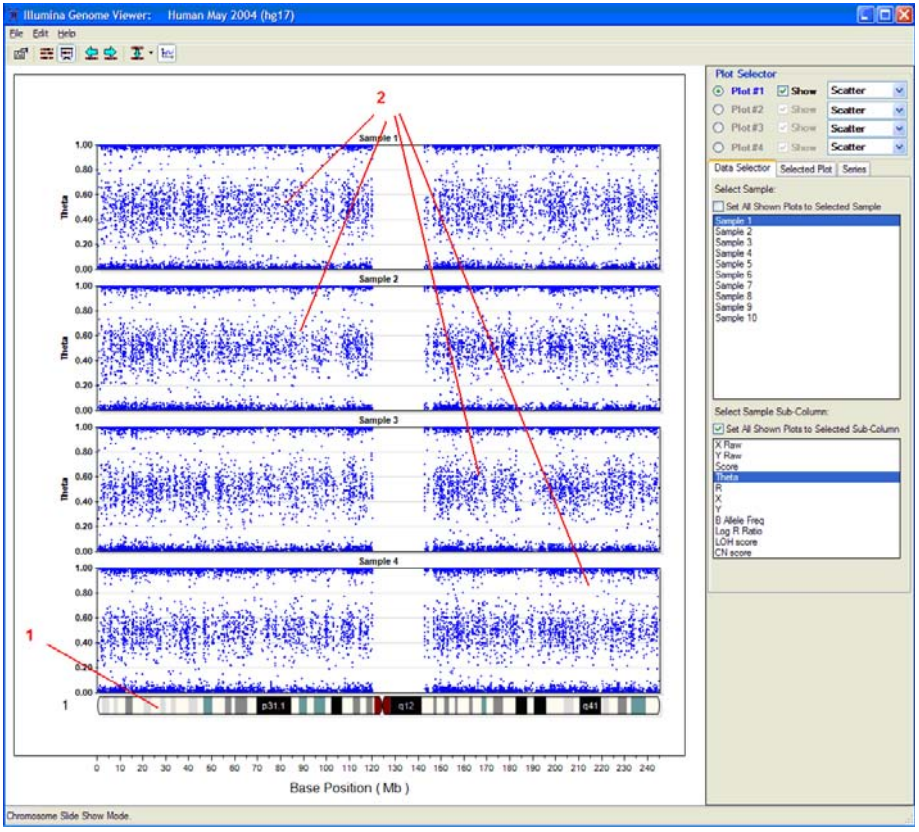


Figure 5-1 Data Visualization at the Chromosomal Level

Getting Data Files

The IGV is installed with a human and mouse genome from the UCSC GoldenPath database.

If you want genomes or builds other than those installed with the IGV, download additional genome annotation files using the following procedure:

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 5-2).

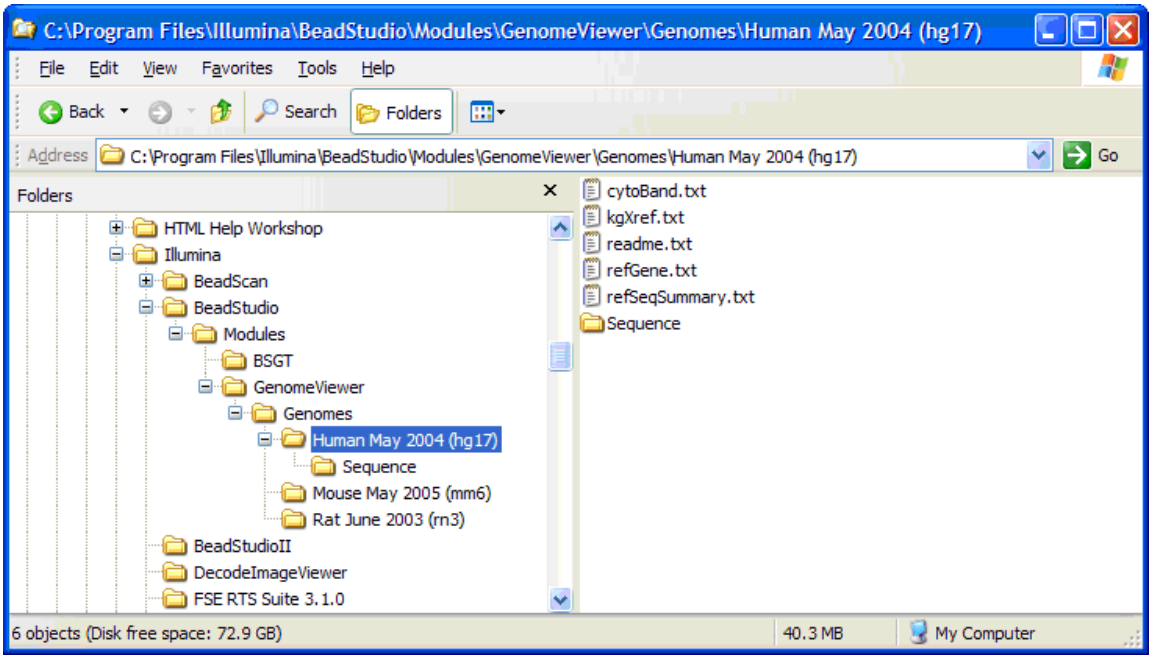


Figure 5-2 IGV, Genome Directory Structure

5. In your internet browser, click the annotation database.
6. Scroll down to the download list.
7. Download and 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*

Download the following optional files if you would like:

- *refSeqSummary.txt*
- *FASTA formatted sequence files in Sequence sub-folder (see Figure 5-3)*

  If you want a FASTA formatted sequence file (e.g., chr1.fa.zip), go to the Data set by chromosome page. Download and unzip the file to a subfolder named "Sequence" (Figure 5-3).

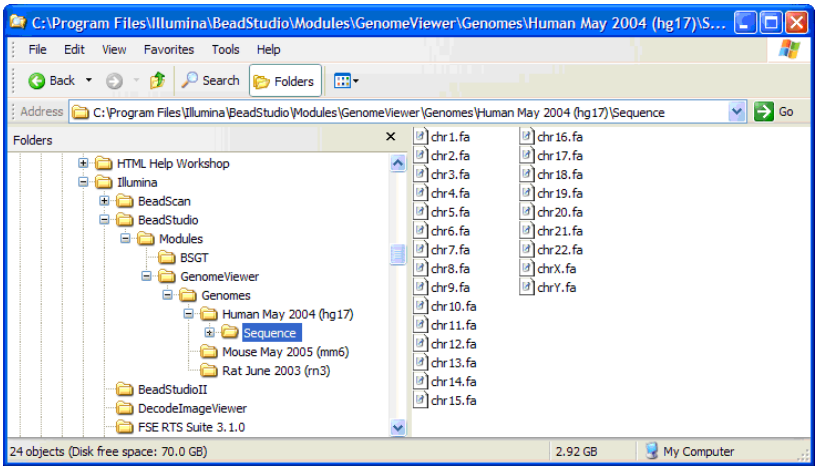


Figure 5-3 IGV, Optional Genome Sequence File Sub-Folder

8. Unzip the files you downloaded in Step 7.
9. To make the newly downloaded genome visible to the IGV, exit the BeadStudio application and restart it.
10. To view a newly added genome, go to **Edit | User Preferences**.

    The **Preferences** dialog box appears.
11. Select the genome you want to view in the **ChangeActiveGenome** list-box.
12. Click **OK**.

    The genome you selected displays in the IGV main window.

Working with the IGV Toolbar

This section contains a brief description of each IGV toolbar function. Figure 5-4 shows the IGV toolbar and callouts for the toolbar buttons.

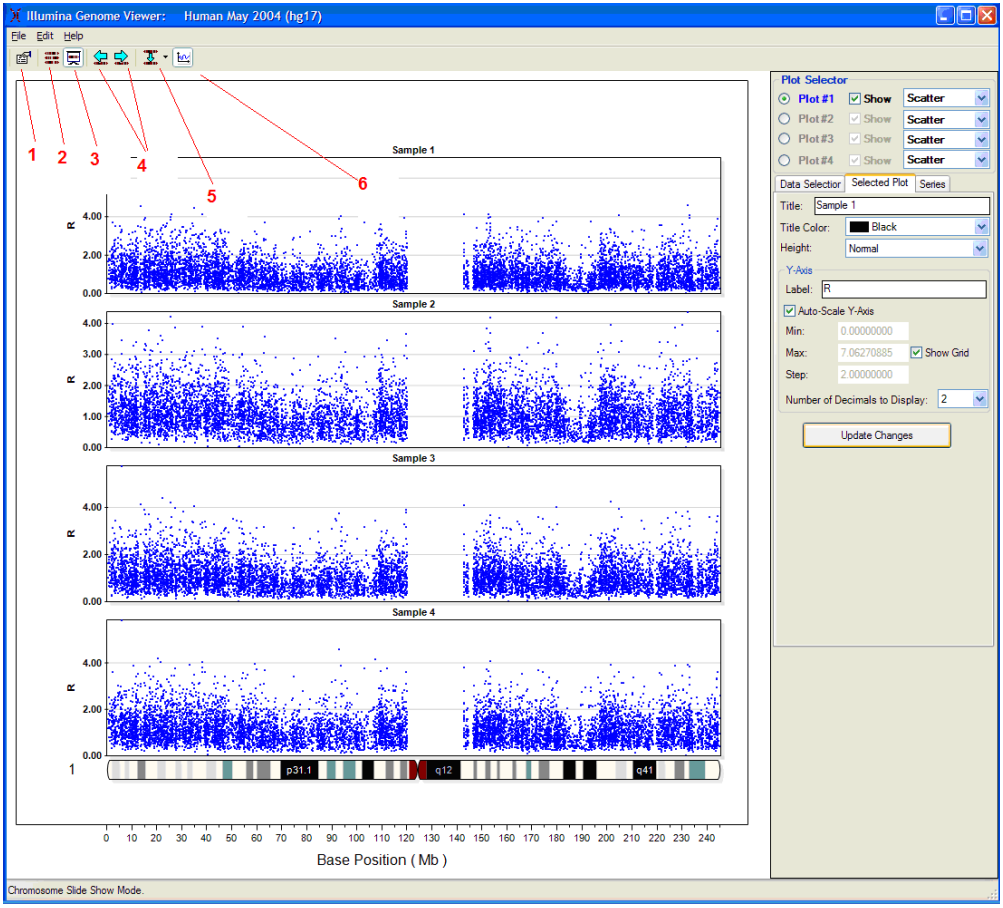


Figure 5-4 Illumina Genome Viewer Toolbar

1.  The **Edit User Preferences** button

Clicking the **Edit User Preferences** toolbar button displays the **Preferences** dialog box shown in Figure 5-5.

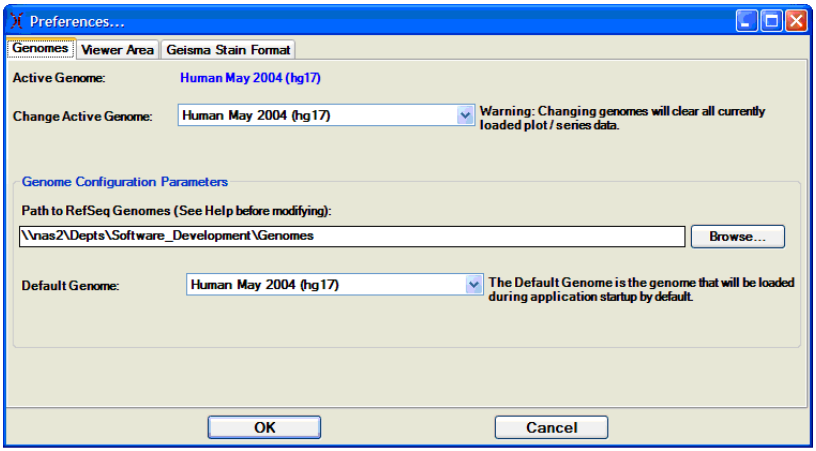


Figure 5-5 Preferences Dialog Box

2.  The **Whole Genome View Mode** button

   Clicking the **Whole Genome View Mode** toggle button shows all genome chromosomes with all enabled plots shown above each chromosome. **Whole Genome View Mode** is the default IGV mode.

3.  The **Chromosome Slide Show Mode** button

   Clicking the **Chromosome Slide Show Mode** toggle button enables the slide show mode which allows the user to view from 1 to 4 chromosomes at the same time.

4.  The **View Previous Slide** button

   Clicking the **View Previous Slide** button displays the previous chromosome slide. This button is only enabled if the **Chromosome Slide Show Mode** toolbar toggle button is active.

    The **View Next Slide** button

   Clicking the **View Next Slide** button displays the next chromosome slide. This button is only enabled if the **Chromosome Slide Show Mode** toolbar toggle button is active.

5.  The **Jump to a Specific Chromosome** button

   Clicking the **Jump to a Specific Chromosome** toolbar button displays a menu that allows you to jump to a particular chromosome slide.

6.  The **Show Plot Selector Panel** button

   Clicking the **Show Plot Selector Panel** toolbar button toggles between showing and hiding the **Plot Selector Panel**. This button is only enabled if the **Chromosome Slide Show Mode** toggle button is active.

Working with Menus

Table 5-1 describes the IGV menus and their functions.

Table 5-1 Illumina Genome Viewer Menus

Element	Description
File Menu	
Exit	Closes the Illumina Genome Viewer window.
Edit Menu	
User Preferences	Displays a dialog box from which you can edit genome, viewing area, and geisma stain properties.
Edit Plots and Series	Displays a dialog box from which you can edit plot and series properties.
Viewed Chromosomes	Displays a dialog box that allows you to specify which chromosomes to display in Whole Genome View mode. This feature is only enabled in Whole Genome View mode.

Plotting Sample Columns

When the Illumina Genome Viewer is launched from the Analysis menu, it has access to the 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 5-6 shows a close-up of the **Plot Selector** Panel.

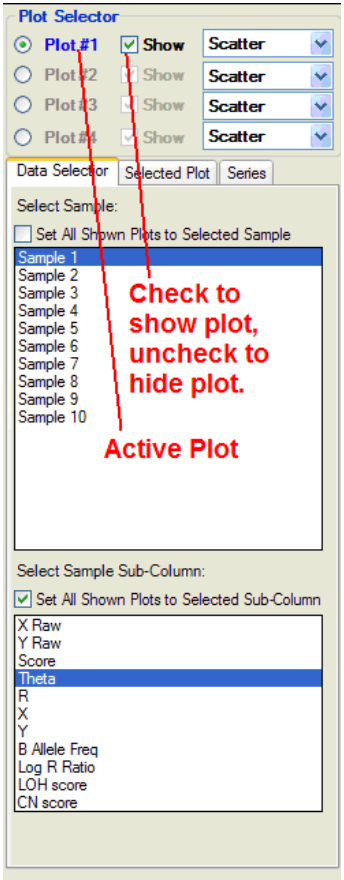


Figure 5-6 Plot Selector Panel

The radio buttons labeled **Plot 1** to **Plot 4** allow you to select which plot to edit. The checkboxes labeled **Show** toggle the particular plot's visibility (leaving this checkbox unchecked hides the plot).

In Figure 5-6, Plot 1 is selected and the data series that is plotted in plot #1 will be the Theta values of Sample 1 as shown by the current selected items in the **Select Sample Sub-Column** and **Select Sample** list boxes, respectively.

To select the sample as the data source for all shown plots, check the **Set All Shown Plots to Selected Sample** checkbox in the **Select Sample** listbox. 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 list box.

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

Zoom Plot

The **Zoom Plot** allows you to zoom into a particular chromosomal region to view data in finer detail relative to known chromosomal annotation. Launch the Zoom Plot by double-clicking on any plot in the IGV (see Figure 5-1). A sample Zoom Plot is shown in Figure 5-7.

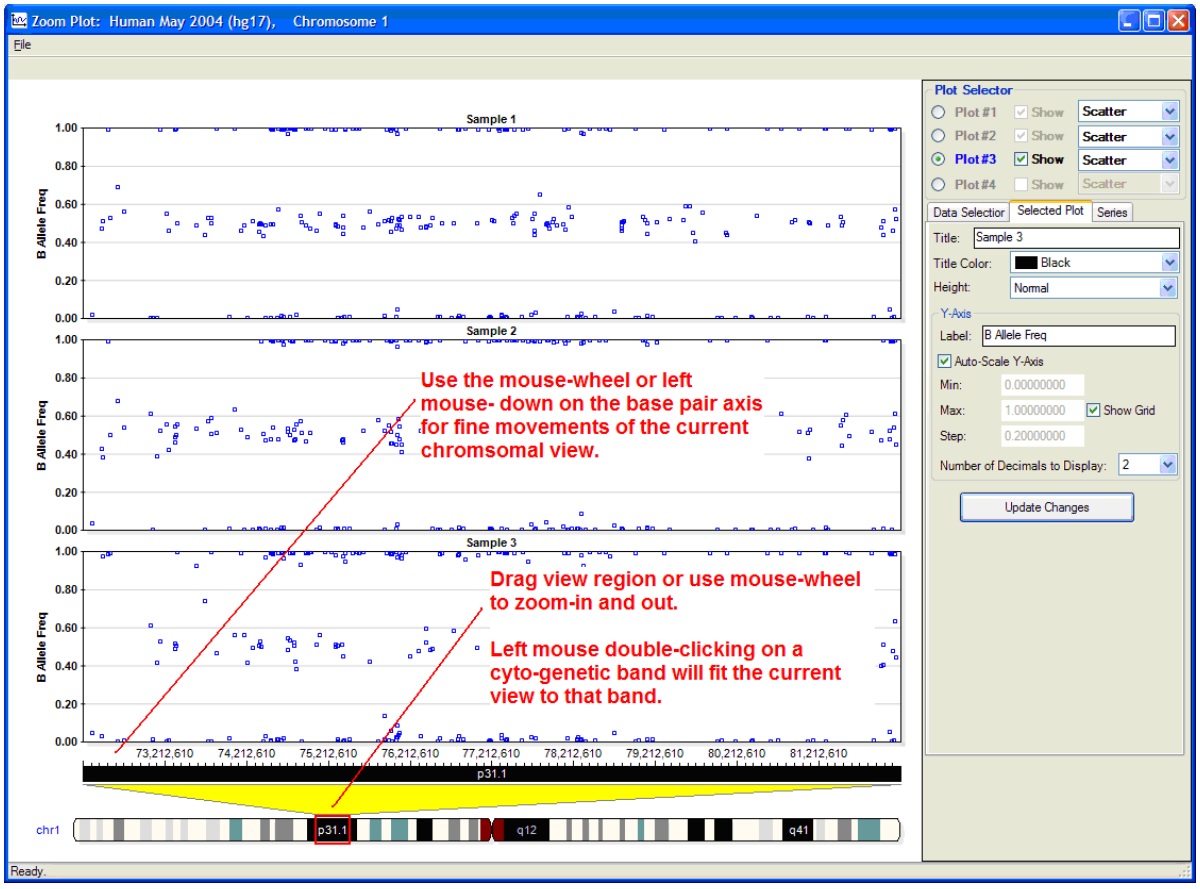


Figure 5-7 Zoom Plot

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 Illumina Chromosome Browser

Launch the ICB by double-clicking on a chromosome in the Illumina Genome Viewer (see callout 1 in Figure 5-8).

Navigating the Illumina Chromosome Browser

Figure 5-8 shows the ICB main window.

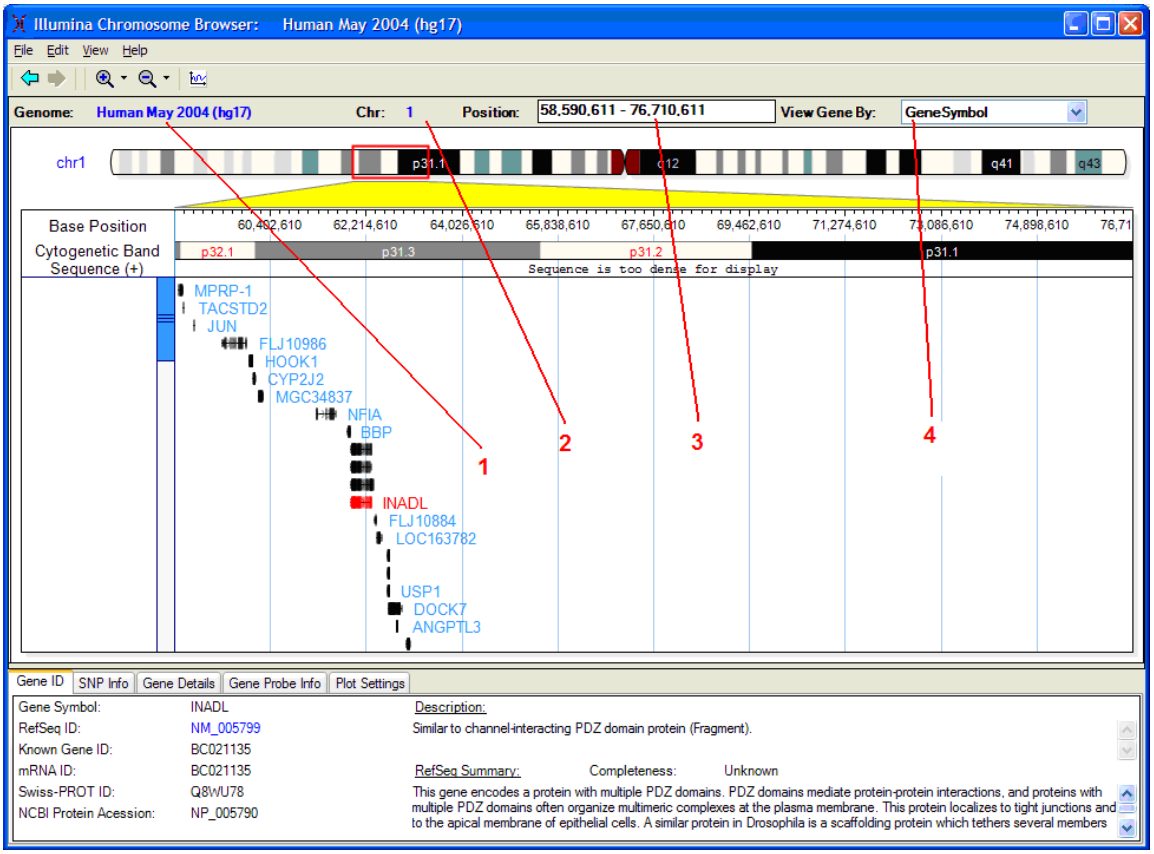


Figure 5-8 Illumina Chromosome Browser (ICB)

1. The **Genome** area -- shows the name of the current genome.
2. The **Chr** area -- shows the current chromosome displayed for the current genome.
3. The **Position** text box -- 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 the **Enter** key to accept the new entered base pair range.

4. The **View Gene By** listbox -- toggles how the gene IDs are shown in the gene region. Genes can be viewed by RefSeq ID, gene symbol, SWISS-PROT ID, NCBI Accession Number, etc.

Figure 5-9 shows additional ICB navigation features. The callouts in the figure are listed and described below.

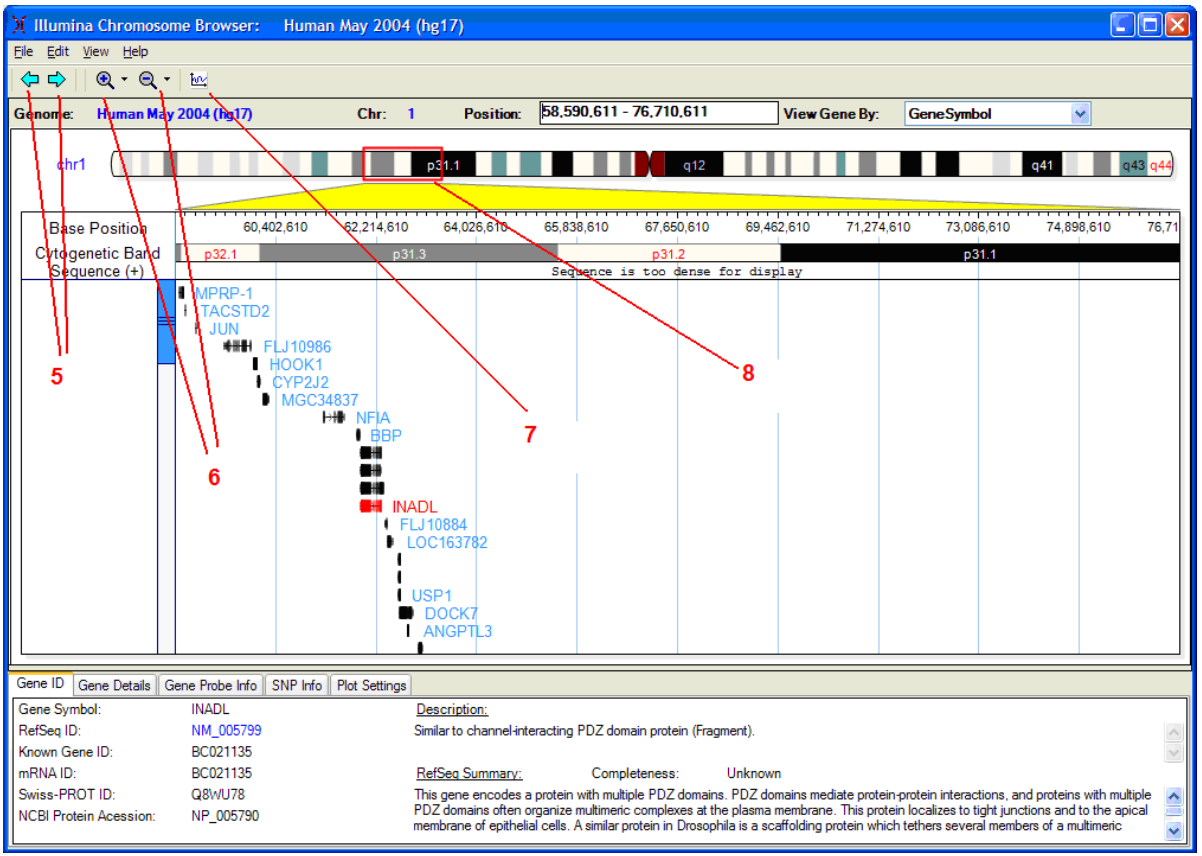


Figure 5-9 Additional Navigation Features of the ICB

5.  **Back**/**Forward** buttons -- behave much like the Back/Forward buttons in an internet browser; they take you backward to and forward from previous views.

6.  **Zoom-In**/**Zoom-Out** buttons -- allow you to magnify or reduce the size of the current view.

7.  **Toggle Plot Display** button -- displays a plot of data above the chromosome. The plot series data can be selected from samples in the **Plot Settings** tabbed window at the bottom of the form.

8. To move the **View Region** (defined by the red rectangle), do one of the following:

   a. Position the cursor inside the red rectangle.

   b. Click and hold the left mouse button and drag the red rectangle to the area you want to view.

   or:

   a. Position the cursor inside the Base Position axis row.

   b. 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:

- 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.

Viewing Gene Information

The gene you select displays in red (Figure 5-9). Information about the selected gene is shown in the **Gene ID** and **Gene Details** tabbed windows at the bottom of the form.

To zoom the current view to fit the whole gene, double-click a gene.

The **Gene ID** tabbed window shows all available gene cross reference names from various databases. A brief description and RefSeq Summary (if available) is 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 5-9).

The **Gene Details** tabbed window 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 list box.

Figure 5-10 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.

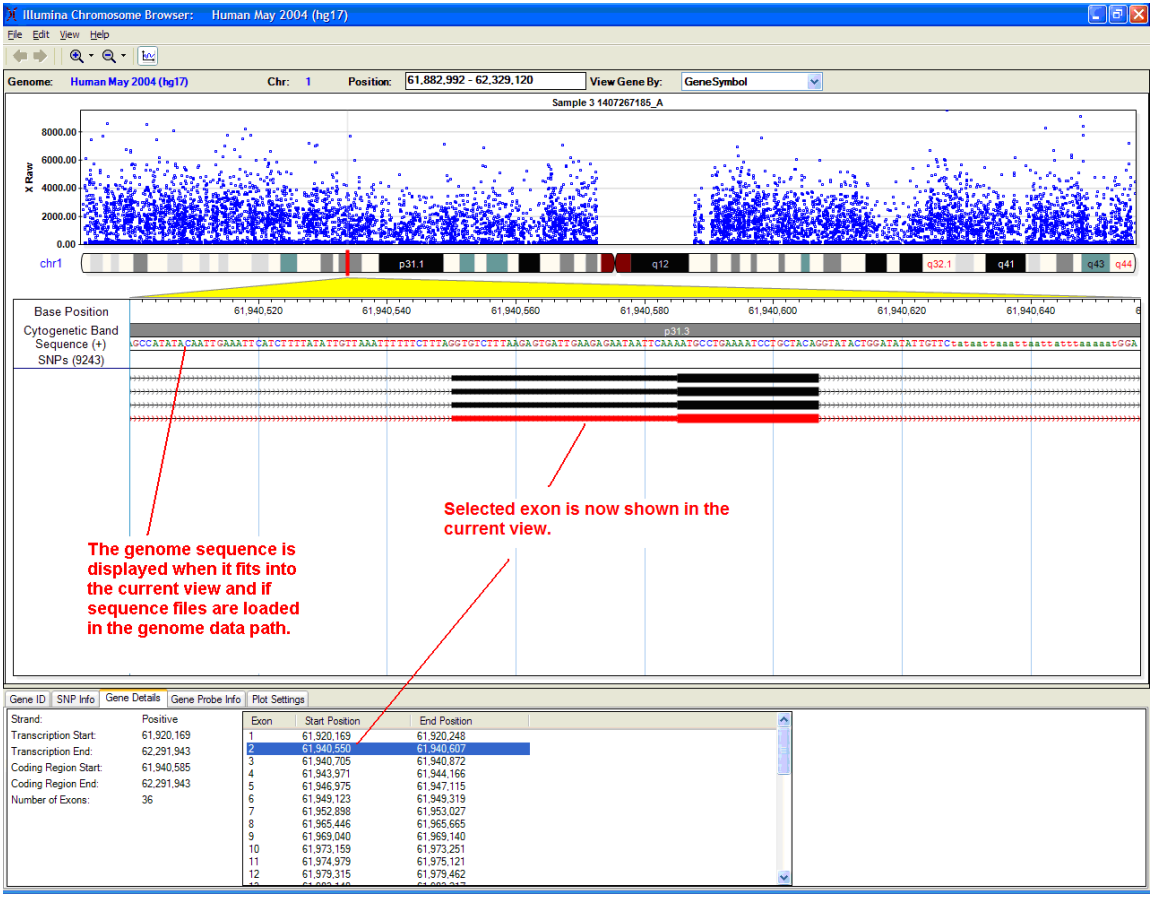


Figure 5-10 Viewing Gene Exons in Detail

Plotting Sample Columns

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

- Click the **Toggle Plot Display** button  .

  The plot toggles to display mode and the **Plot Settings** tab at the bottom of the screen becomes active (Figure 5-11).

To hide the plot:

- Click the **Toggle Plot Display** button  . 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. The plot type, y-axis properties, and series properties can be set in the **Plot Settings** tab.

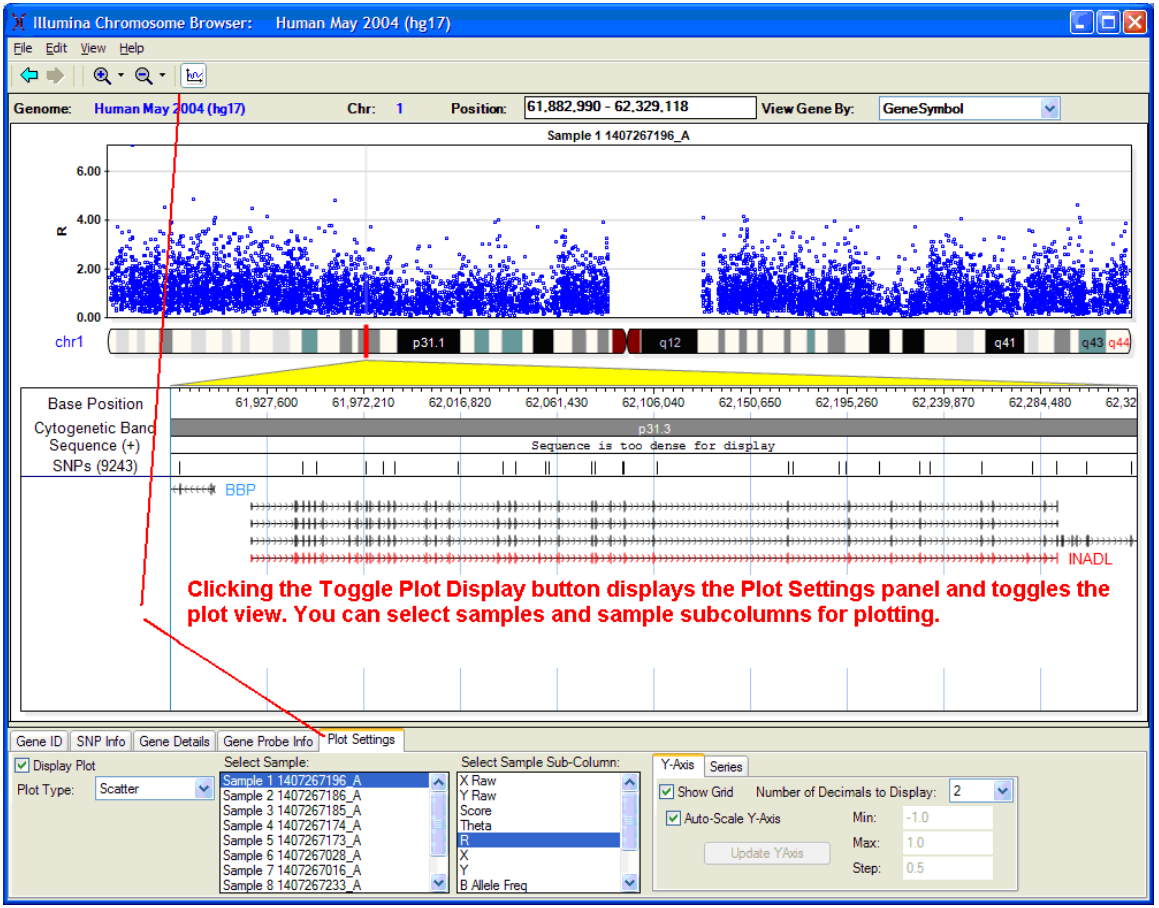


Figure 5-11 Plotting Sample Data in the ICB

Viewing Project Manifest SNPs

To display SNPs in the current project manifest:

Select **File | Load Manifest | SNPs**.

The SNPs in the current manifest are displayed (Figure 5-12).

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.

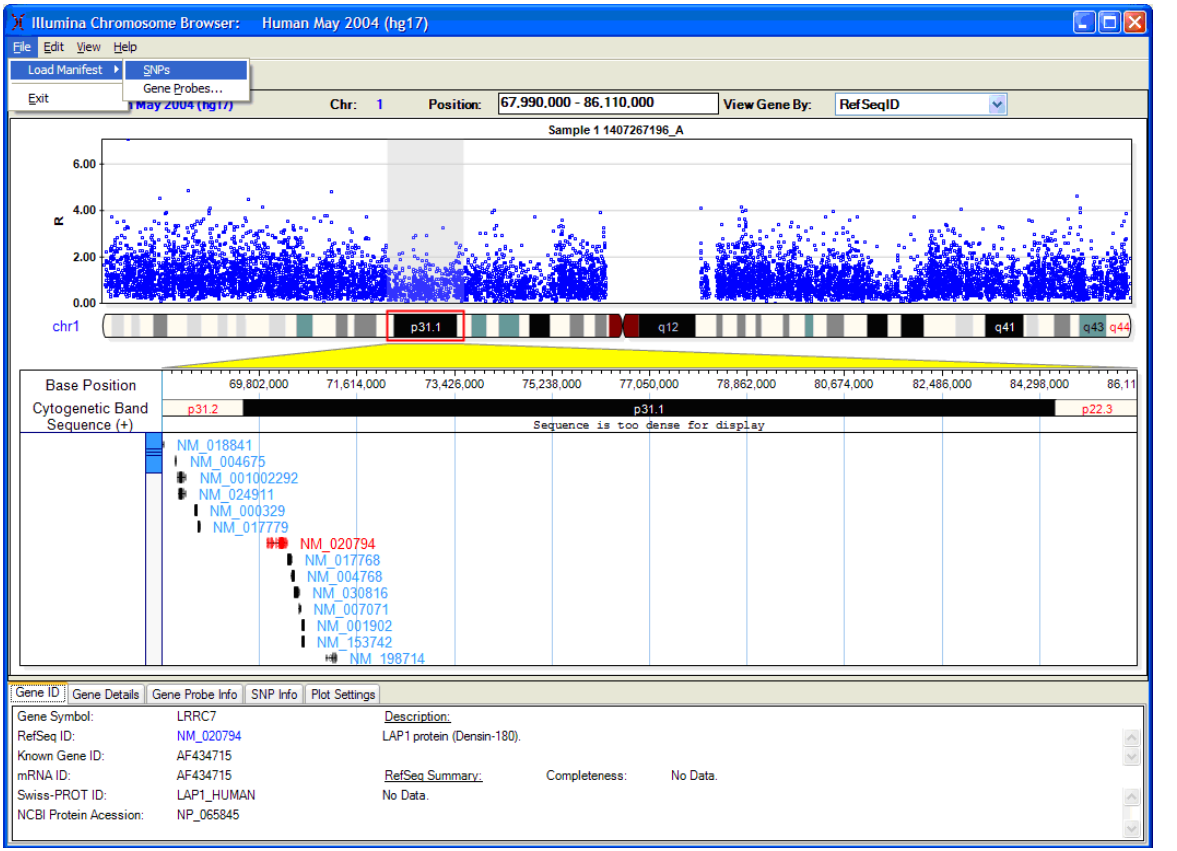


Figure 5-12 Loading Project Manifest SNPs into the ICB

Figure 5-13 shows manifest SNPs loaded after selecting **File | Load Manifest | SNPs**.

SNPs are represented by vertical bars. To activate the **SNP Info** tabbed window, click a SNP. To cause the current view to zoom into the selected SNP, double-click a SNP.

The **SNP Info** tabbed window shows the RefSNP ID, allele, chromosome position, and top genomic sequence for the SNP.

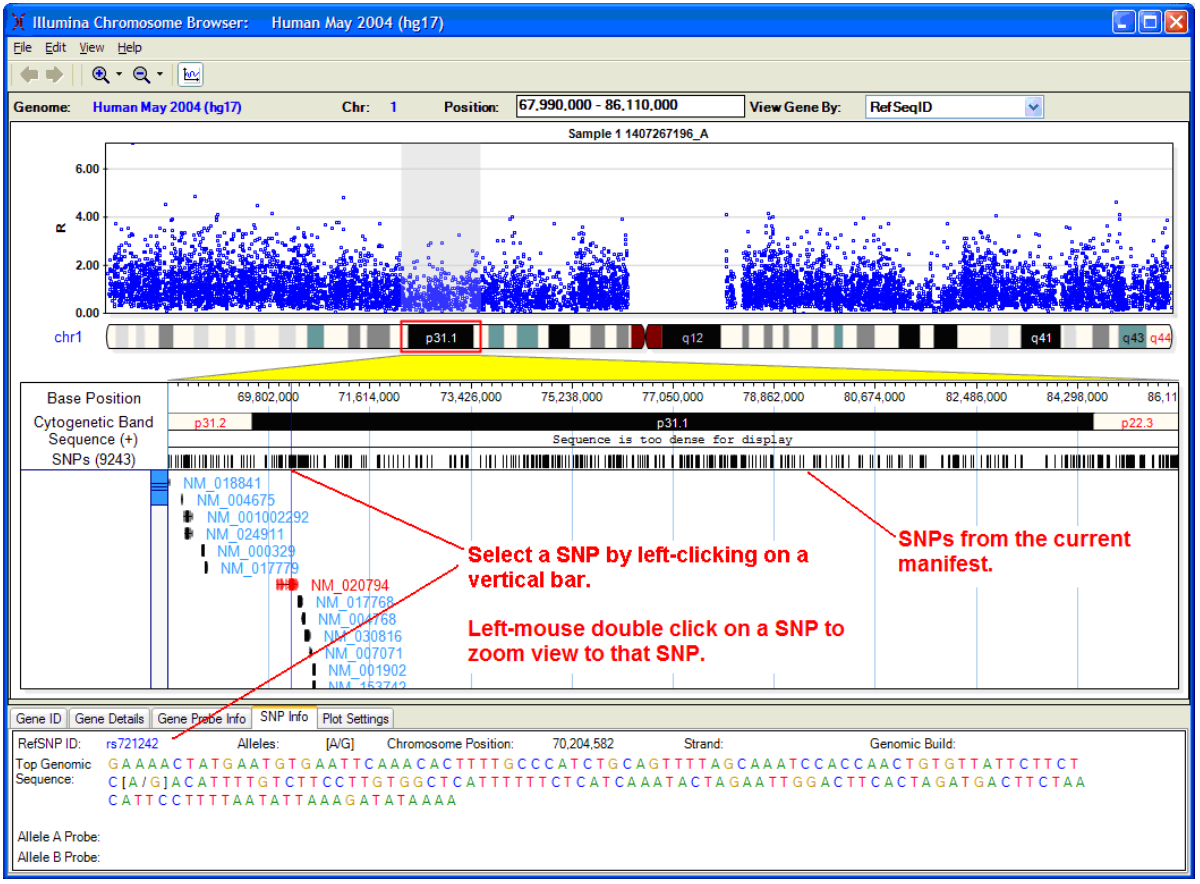


Figure 5-13 Viewing GT Manifest SNPs in the ICB

A vertical blue line overlays the SNP, showing any genes with which the SNP intersects (Figure 5-14).

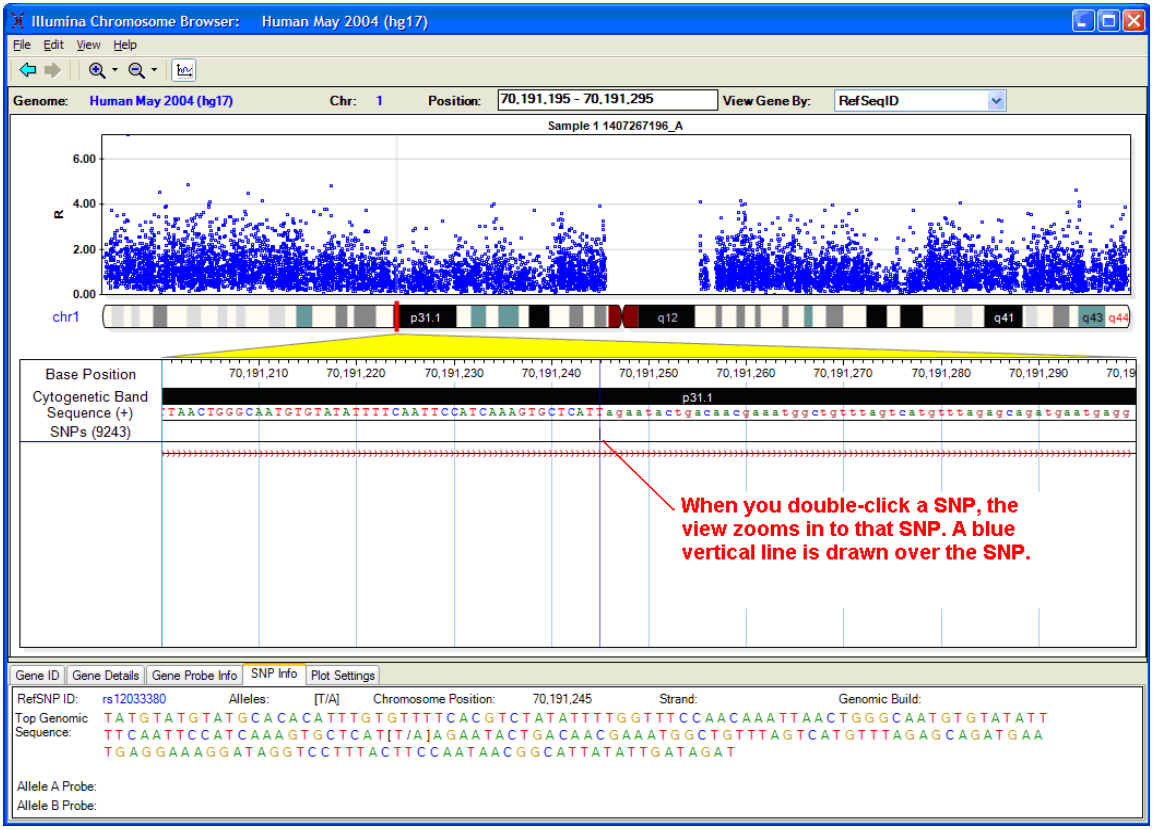


Figure 5-14 Zoomed View (Double-clicking a SNP)

Using the Illumina Sequence Viewer

To use the Illumina Sequence Viewer (ISV):

1. In the IGV, go to **Analysis | Show columns**.
2. Click on a region of the chromosome.

   The **Illumina Chromosome Browser** appears (Figure 5-15).

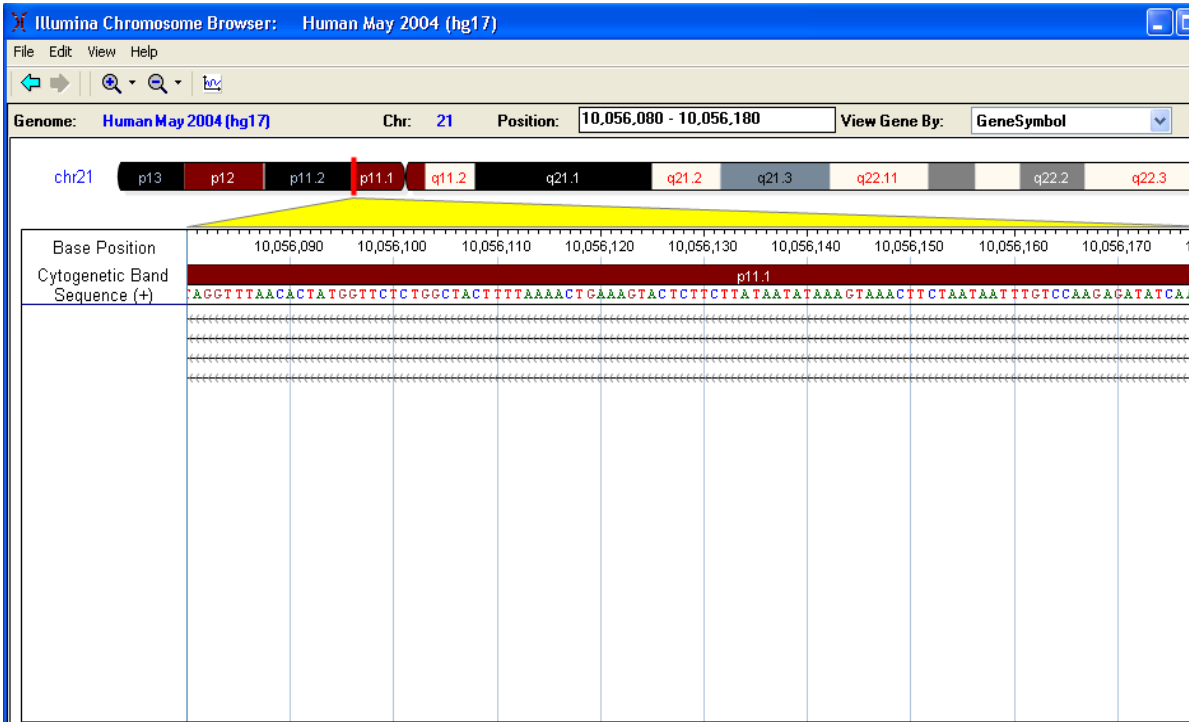


Figure 5-15 Illumina Chromosome Browser

3. Go to **View | Sequence Viewer**.

   The **Illumina Sequence Viewer** displays (Figure 5-16).

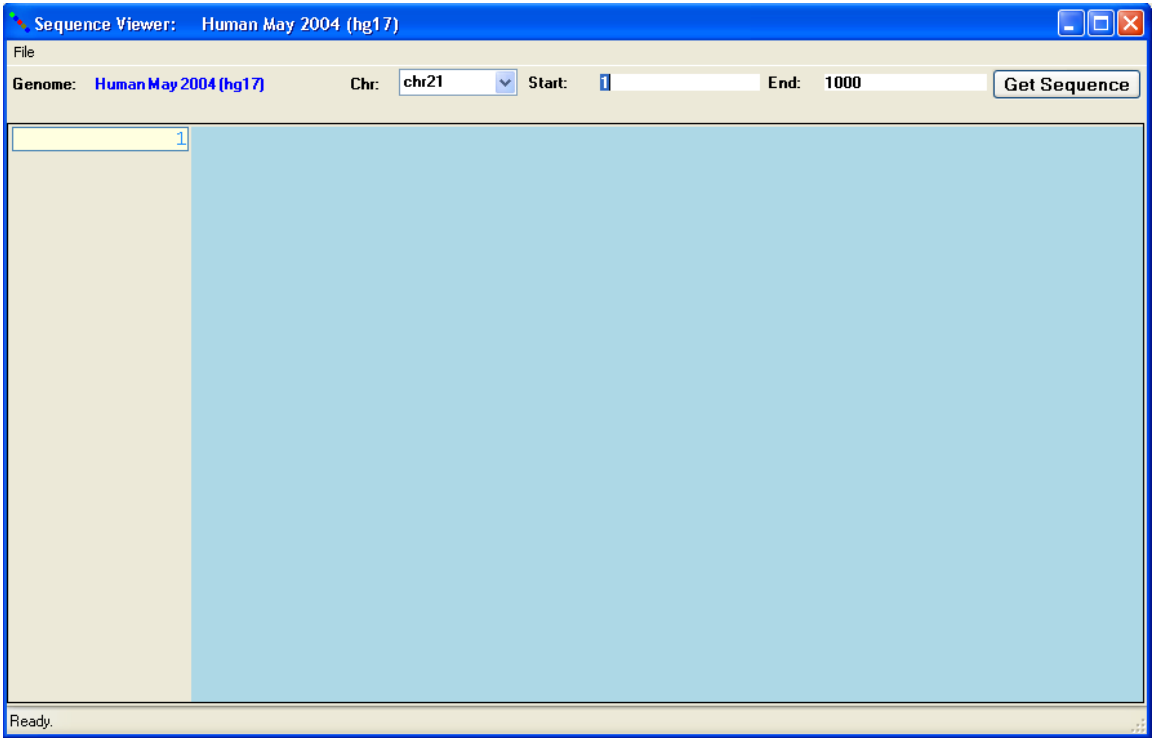


Figure 5-16 Illumina Sequence Viewer

To view a specific sequence in the ISV:

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

   The sequence you selected appears in the ISV.

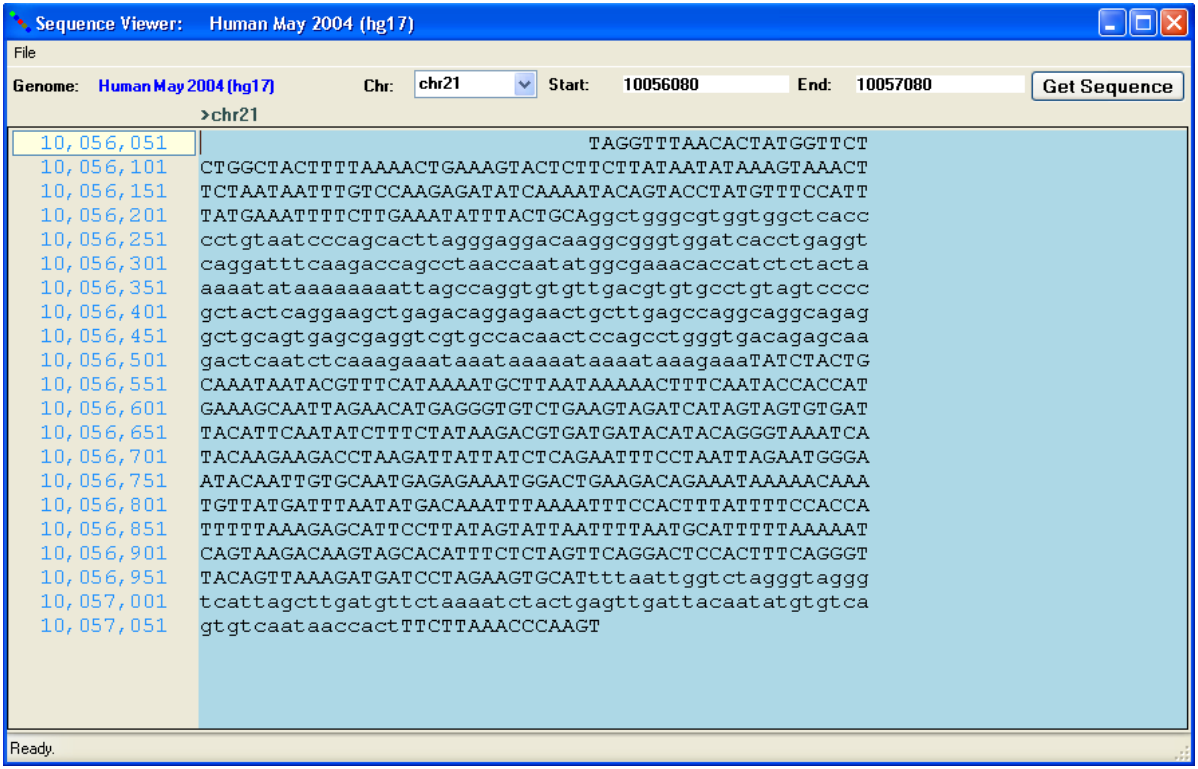


Figure 5-17 Illumina Sequence Viewer with Sequence Displayed

NOTES

illumina

Appendix

Troubleshooting Guide

Introduction

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

Frequently Asked Questions

Table A-1 lists frequently asked questions and associated responses.

Table A-1 Frequently Asked Questions

#	Question	Response
1	What is the data repository?	The **data repository** is a parent directory that contains the directories for the Sentrix array products you have scanned in your experiments. It contains the directories with IDAT files. BeadScan, the scanning software for the BeadArray Reader, will automatically create a subdirectory in the data repository you have defined for each Sentrix array product it scans. You should point BeadStudio to the same data repository.
2	What is the project repository?	The **project repository** is the parent directory that contains the information for BeadStudio projects.
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 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.

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