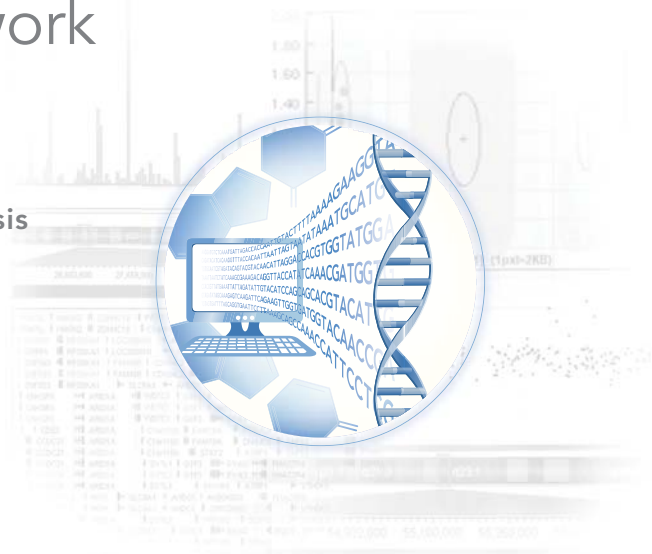


GenomeStudio™ 2008.1 Framework User Guide

An Integrated Platform for
Data Visualization and Analysis

FOR RESEARCH ONLY





Notice

This publication and its contents are proprietary to Illumina, Inc., and are intended solely for the contractual use of its customers and for no other purpose than to operate the system described herein. This publication and its contents shall not be used or distributed for any other purpose and/or otherwise communicated, disclosed, or reproduced in any way whatsoever without the prior written consent of Illumina, Inc.

For the proper operation of this system and/or all parts thereof, the instructions in this guide must be strictly and explicitly followed by experienced personnel. All of the contents of this guide must be fully read and understood prior to operating the system or any parts thereof.

FAILURE TO COMPLETELY READ AND FULLY UNDERSTAND AND FOLLOW ALL OF THE CONTENTS OF THIS GUIDE PRIOR TO OPERATING THIS SYSTEM, OR PARTS THEREOF, MAY RESULT IN DAMAGE TO THE EQUIPMENT, OR PARTS THEREOF, AND INJURY TO ANY PERSONS OPERATING THE SAME.

Illumina, Inc. does not assume any liability arising out of the application or use of any products, component parts or software described herein. Illumina, Inc. further does not convey any license under its patent, trademark, copyright, or common-law rights nor the similar rights of others. Illumina, Inc. further reserves the right to make any changes in any processes, products, or parts thereof, described herein without notice. While every effort has been made to make this guide as complete and accurate as possible as of the publication date, no warranty of fitness is implied, nor does Illumina accept any liability for damages resulting from the information contained in this guide.

© 2005-2008 Illumina, Inc. All rights reserved.

Illumina, Solexa, Making Sense Out of Life, Oligator, Sentrix, GoldenGate, DASL, BeadArray, Array of Arrays, Infinium, BeadXpress, VeraCode, IntelliHyb, iSelect, CSPro, iScan, and GenomeStudio are registered trademarks or trademarks of Illumina, Inc. All other brands and names contained herein are the property of their respective owners.



Revision History

Part Number	Revision	Title	Date
Part # 11318815	Rev. A	GenomeStudio 2008.1 Framework User Guide	November 2008
Part # 11285128	Rev. A	BeadStudio Framework v3.1.3 User Guide	December 2007
Part # 11204578	Rev. C	BeadStudio Framework User Guide	February 2007
Part # 11204578	Rev. B	BeadStudio Framework User Guide	March 2006
Part # 11204578	Rev. A	BeadStudio Framework User Guide	December 2005



Table of Contents

	Notice	iii
	Revision History.	v
	Table of Contents	vii
	List of Figures	xi
	List of Tables.	xv
Chapter 1	Overview	1
	Introduction.	2
	Audience and Purpose	2
	Installing GenomeStudio	3
	Starting GenomeStudio	7
	Verifying Your Version Numbers.	9
	Closing GenomeStudio.	10
Chapter 2	Detachable Docking Windows.	11
	Introduction.	12
	Working with Detachable Docking Windows	12
	Saving Your Window Configuration	17
	Reverting to the Default Window Configuration	17
Chapter 3	Tables	19
	Introduction.	20
	Working with Tables	20
	Selecting Rows	21
	Selecting Columns	22
	Showing & Hiding Columns	23

Searching Within Tables	24
Marking Table Rows	26
Clearing Marks	28
Exporting to a File.	29
Importing a Column	29
Sorting by Column	31
Creating a New Column	32
Editing a User-Defined Subcolumn.	35
Deleting a User-Defined Subcolumn	36
Filtering Rows	38
Locking Columns.	43
Unlocking Columns.	45

Chapter 4 Graphs 47

Introduction	49
Line Plots	49
Plotting a Single Column	49
Plotting Two Columns.	51
Changing the X-Axis Label	52
Changing the Plot Type	54
Scatter Plots.	57
Marking Samples.	58
Box Plots	60
Frequency Plots.	61
Pie Charts	63
Histograms	64
Adjusting the View Area	66
Changing the Number of Bins	66
Selecting Data.	66
Viewing Statistics.	68
Filtering Data	69
Clearing a Filter.	70
Marking a Selection	70
Heat Maps	72
Populating the Heat Map with Data	73
Clustering Heat Map Data	75
Similarities and Distances	76
Resizing Row Labels	79
Resizing the Column Tree Cluster.	79
Resizing Column Labels	79
Scrolling the Heat Map Area.	80

	Opening a Heat Map File	80
	Saving a Heat Map	80
	Exiting the Heat Map	81
	Editing Heat Map Properties	81
	Creating a Presentation Image	87
Chapter 5	Illumina Genome Viewer	89
	Introduction	90
	Using the IGV	90
	Selecting Data Plots	91
	Saving Data Plots	95
	Changing Viewing Modes	96
	Using the IGV Toolbar	100
	Using the IGV Menus	102
	Plotting Data in the IGV	103
	Viewing Data in the IGV	111
	Getting Data Files	113
Chapter 6	Illumina Chromosome Browser and Other Visualization Tools	121
	Introduction	123
	Using the ICB	123
	Using the ICB Toolbar	124
	Using the ICB Context Menu	127
	Changing the View Region	127
	Viewing Gene Information	128
	Viewing Project Manifest SNPs	131
	Finding Genes or Probes	133
	Loading Illumina-Provided Data Tracks	134
	Creating and Loading Your Own Data Tracks	137
	Using the Illumina Sequence Viewer	138
	Using the Chromosome Heat Map	140
	Clustering Data	141
	Binning Data	143
	Smoothing Data	144
Chapter 7	Bookmarking Tool	147
	Introduction	148
	Using the Bookmarking Tool	148
	Viewing Bookmarks in the ICB	149

Using the Bookmark Viewer	151
Manually Creating Bookmarks	155
Editing Bookmarks	157
Saving Bookmarks	158
Creating a Bookmark List	161
Editing Bookmark Types	162
Index	165

List of Figures

Figure 1	GenomeStudio Application Suite Unzipping	3
Figure 2	Selecting GenomeStudio Software Modules	4
Figure 3	License Agreement	5
Figure 4	Installing GenomeStudio	5
Figure 5	Installation Complete.	6
Figure 6	GenomeStudio Registration Dialog Box	7
Figure 7	GenomeStudio Installed	8
Figure 8	GenomeStudio Main Window	9
Figure 9	About GenomeStudio	10
Figure 10	Docking Icons.	13
Figure 11	Docking a Window Over Another Window	14
Figure 12	Docking a Window to the Right of Another Window.	15
Figure 13	Docking a Window to Occupy Side of Main Window	16
Figure 14	Restoring the Default View	17
Figure 15	Table without Subcolumns.	20
Figure 16	Table with Subcolumns	21
Figure 17	Table with Column Selected	22
Figure 18	Column Chooser	23
Figure 19	Tables Context Menu.	24
Figure 20	Find Row	24
Figure 21	Highlighted Table Row Containing Search Term.	25
Figure 22	Configure Marks Selected	26
Figure 23	Configuring Marks	27
Figure 24	Selecting Mark Name	27
Figure 25	Selecting Mark Color	27
Figure 26	Selected Rows Marked in Table.	28
Figure 27	Clearing Selected Marks from Table	28
Figure 28	Exporting Data	29
Figure 29	Importing Data.	30
Figure 30	Multiple-Column Sort	31
Figure 31	Creating a New Column.	32
Figure 32	Building an Equation	34

Figure 33	New Column Displayed for Every Sample	35
Figure 34	Editing a User-Defined Subcolumn	35
Figure 35	Changing Subcolumn Attributes	36
Figure 36	Deleting a Column	37
Figure 37	Delete Subcolumn	37
Figure 38	Subcolumn Deleted from the Table.	37
Figure 39	Filtering Table Rows	38
Figure 40	Creating a Simple Filter	39
Figure 41	Creating a More Complex Filter.	41
Figure 42	Entering a Name for a Filter.	41
Figure 43	Table with Two Locked Columns	43
Figure 44	Column Chooser, Display-Locked Columns.	44
Figure 45	Selecting Columns to Unlock	45
Figure 46	Selected Columns Unlocked	45
Figure 47	GenomeStudio Line Plot	50
Figure 48	Plotting a Single Column in a Line Plot	51
Figure 49	Plotting Two Columns in a Line Plot.	52
Figure 50	Line Plot with X-Axis Label "Address"	53
Figure 51	Line Plot Context Menu	54
Figure 52	Plot Settings	55
Figure 53	Bar Graph	56
Figure 54	Scatter Plot	57
Figure 55	Universal Scatter Plot	58
Figure 56	Scatter Plot Context Menu.	59
Figure 57	Select Mark Name	59
Figure 58	Selecting Mark Name & Color	59
Figure 59	Box Plot	60
Figure 60	Box Plot Displaying Data	61
Figure 61	Frequency Plot	62
Figure 62	Frequency Plot Displaying Data	62
Figure 63	Pie Chart Displaying Data	63
Figure 64	Pie Chart Displaying Column and Subcolumn Data.	64
Figure 65	Histogram	65
Figure 66	Histogram Displaying Data.	65
Figure 67	Histogram with Shaded Selection Area	67
Figure 68	Resizing Shaded Selection Area.	68
Figure 69	Filtering Selected Data.	69
Figure 70	Configure Marks.	70
Figure 71	Select Mark Name	70
Figure 72	Choosing Mark Name & Color.	71
Figure 73	Marked Data Displayed in a Histogram	71
Figure 74	Marked Data Displayed in Associated Table	72

Figure 75	Plot Sample Subcolumns in a Heat Map	73
Figure 76	Heat Map with Row Labels & Data Series	74
Figure 77	Cluster Options	75
Figure 78	Dendrogram, Similarity Example	77
Figure 79	Dendrogram, Showing Nodes	77
Figure 80	Heat Map, Display & Scrolling Properties	78
Figure 81	Heat Map Properties, Title Tab	82
Figure 82	Heat Map Properties, Legend Tab	83
Figure 83	Heat Map Properties, Row/Columns Tab	85
Figure 84	Heat Map Properties, Scroll Bars Tab	87
Figure 85	Presentation Image Setup	88
Figure 86	Launching the IGV	90
Figure 87	IGV Main Window, Whole Genome View Mode	91
Figure 88	IGV Data Workspace Context Menu	92
Figure 89	Add Favorite Data Plots Dialog Box	93
Figure 90	Favorite Data Plots Selected	94
Figure 91	Favorite Data Plots in the IGV Data Workspace	95
Figure 92	Data Plot, Whole Genome View Mode	97
Figure 93	Chromosome Slide Show View Options	98
Figure 94	Data Plot, Chromosome Slide Show Mode	98
Figure 95	Data Plot, Display Options Selected	100
Figure 96	IGV Toolbar	100
Figure 97	IGV Data Workspace, Microarray Modules	104
Figure 98	IGV Data Workspace, Sequencing Modules	105
Figure 99	IGV Data Workspace, Data Plots Tab	106
Figure 100	IGV Data Workspace, Stacked Alignment Plots Tab	106
Figure 101	IGV Data Workspace, Stacked Alignments Plots, Display Options	107
Figure 102	IGV Data Workspace, Data View Tab	108
Figure 103	Data Properties Tab	109
Figure 104	Display Properties Tab	111
Figure 105	Data View Tab	112
Figure 106	UCSC Genome Bioinformatics Web Page	113
Figure 107	Selected Genome Web Page (Human)	114
Figure 108	Annotation Database Web Page (Human)	115
Figure 109	Annotation Database Files Web Page, Files	116
Figure 110	Index (Data set by chromosome) Web Page	117
Figure 111	Preferences	118
Figure 112	Changing the Displayed Genome Species & Build	119
Figure 113	Continue Changing Displayed Genome	119
Figure 114	Selected Genome Shown in the IGV	120
Figure 115	ICB	124

Figure 116	ICB Toolbar.	125
Figure 117	Resizing the View Region	128
Figure 118	Viewing Gene Exons in Detail	130
Figure 119	Loading Project Manifest SNPs into the ICB	131
Figure 120	Zoomed View (Double-Clicking a SNP)	132
Figure 121	Add/Remove Data Tracks	134
Figure 122	Selected Data Tracks	135
Figure 123	Data Tracks Displayed in the ICB	136
Figure 124	ISV.	138
Figure 125	ISV Displaying a Selected Sequence	139
Figure 126	Chromosome Heat Map.	140
Figure 127	Chromosome Heat Map.	141
Figure 128	Cluster Options	142
Figure 129	Data Binning Options.	143
Figure 130	Smooth Sample Data Options	144
Figure 131	Bookmark Information Popup Window	149
Figure 132	Enable Manual Placement Checkbox Cleared	150
Figure 133	Bookmark Viewer	152
Figure 134	Bookmark Viewer with Bookmark Analysis Selected	152
Figure 135	Delete Selected Bookmarks.	154
Figure 136	Deleting Multiple Bookmarks	155
Figure 137	Add Bookmark	156
Figure 138	Bookmarks Displayed in the ICB	157
Figure 139	Moving View Region.	158
Figure 140	Add Bookmark	159
Figure 141	Bookmark Created at View Region.	160
Figure 142	New Bookmark List.	161
Figure 143	Add Bookmark, New Bookmark Visible	161
Figure 144	Edit Bookmark Types	162
Figure 145	Adding Comments to a Bookmark.	163



List of Tables

Table 1	Guidelines for Importing Columns or Subcolumns . . .	30
Table 2	New Column Operations & Descriptions.	33
Table 3	Histogram Statistics	69
Table 4	IGV Toolbar Buttons.	101
Table 5	IGV Menu Options & Descriptions.	102
Table 6	ICB Toolbar Elements	125
Table 7	Sample User-Created Data Track File	137



Overview

Topics

- 2 Introduction
- 2 Audience and Purpose
- 3 Installing GenomeStudio
- 7 Starting GenomeStudio
- 9 Verifying Your Version Numbers
- 10 Closing GenomeStudio

Introduction

GenomeStudio is a modular software platform that allows you to view and analyze data for a wide range of microarray and sequencing applications including:

Microarray Applications:

- ▶ Genotyping
- ▶ Gene Expression
- ▶ Methylation
- ▶ Protein Analysis

Sequencing Applications:

- ▶ ChIP Sequencing
- ▶ DNA Sequencing
- ▶ RNA Sequencing

The GenomeStudio platform includes the GenomeStudio Framework, various analysis software modules, and additional plug-ins that you can download from the GenomeStudio Portal.

The GenomeStudio Framework includes a common user interface and tools that are available in all GenomeStudio modules. In addition, each GenomeStudio module allows you to perform specific types of analyses. To use the GenomeStudio Framework, you must also install at least one licensed GenomeStudio software module.

Audience and Purpose

This guide is written for researchers who want to use GenomeStudio to view and analyze data for various microarray and sequencing applications.

This guide provides procedures and user interface information for the GenomeStudio Framework, the set of features and tools common to all GenomeStudio modules. For information about the features specific to each GenomeStudio application, refer to the user guide for that module:

- ▶ *GenomeStudio Genotyping Module User Guide*
- ▶ *GenomeStudio Gene Expression Module User Guide*
- ▶ *GenomeStudio Methylation Module User Guide*
- ▶ *GenomeStudio Protein Analysis Module User Guide*
- ▶ *GenomeStudio ChIP Sequencing Module User Guide*
- ▶ *GenomeStudio DNA Sequencing Module User Guide*
- ▶ *GenomeStudio RNA Sequencing Module User Guide*

Installing GenomeStudio

To install the GenomeStudio Framework:

1. Put the GenomeStudio CD into your CD drive.

If the Illumina GenomeStudio Installation screen appears (Figure 2), continue to Step 3.

If the CD does not load automatically, double-click the *GenomeStudio<version>.exe* icon in the **GenomeStudio** folder on the CD.

The GenomeStudio application suite unzips (Figure 1).

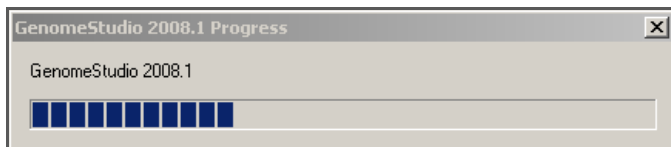


Figure 1 GenomeStudio Application Suite Unzipping

The Illumina GenomeStudio Installation dialog box appears (Figure 2).

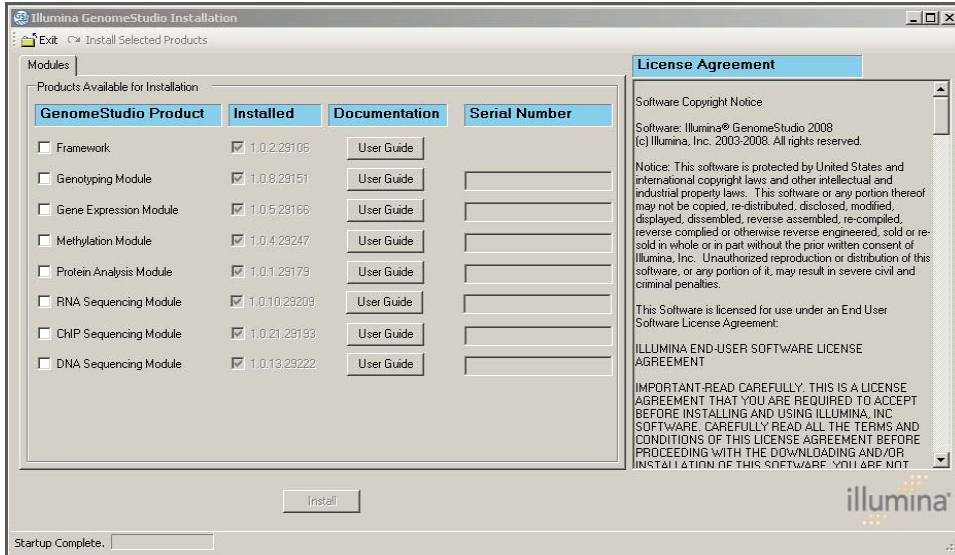


Figure 2 Selecting GenomeStudio Software Modules

2. Read the software license agreement in the right-hand side of the Illumina GenomeStudio Installation dialog box.
3. In the GenomeStudio Product area, select **Framework**.



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

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



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

5. **[Optional]** Enter the serial numbers for additional GenomeStudio modules if you have licenses for additional GenomeStudio modules and want to install them now.
6. Click **Install**.

The Software License Agreement dialog box appears (Figure 3).



Figure 3 License Agreement

7. Click **Yes** to accept the software license agreement. The GenomeStudio Framework is installed on your computer, along with any GenomeStudio modules you selected (Figure 4).

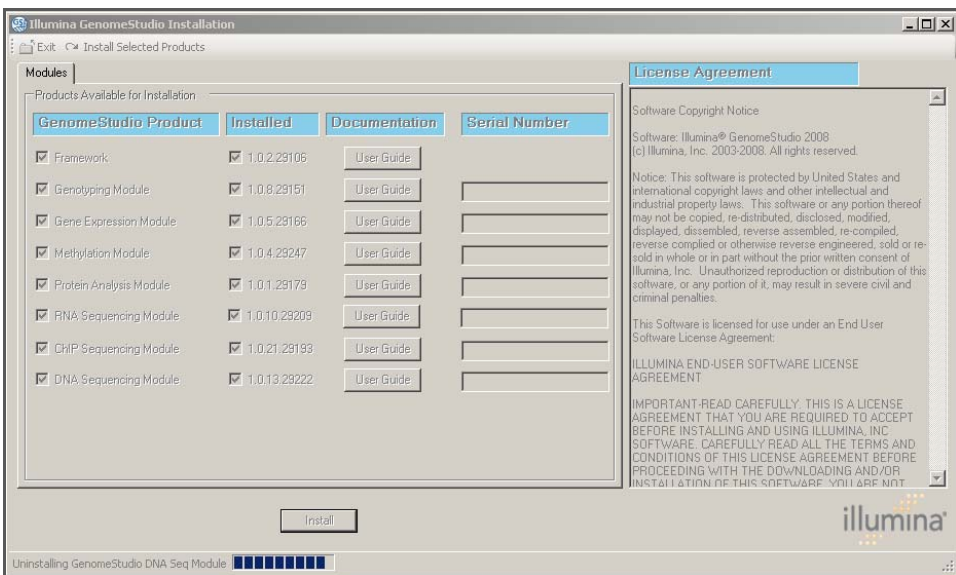


Figure 4 Installing GenomeStudio

The Installation Progress dialog box notifies you that installation is complete (Figure 5).

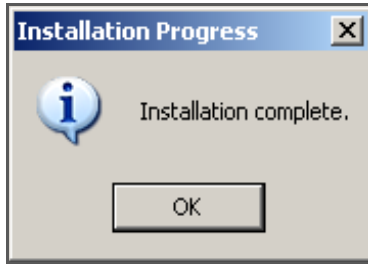


Figure 5 Installation Complete

8. Click **OK**.
9. In the Illumina GenomeStudio Installation dialog box (Figure 4), click **Exit**.

You can now start a new GenomeStudio project using any GenomeStudio module you have installed along with the Framework.

Starting GenomeStudio

Perform the following steps to start GenomeStudio:

1. Do one of the following:

▶ Select **Start | Program Files | Illumina | GenomeStudio**.

▶ Double-click the GenomeStudio icon on your desktop.



GenomeStudio opens to the Registration dialog box (Figure 6).

A screenshot of the 'Registration' dialog box. The title bar says 'Registration'. On the left is the 'illumina' logo. On the right, text reads: 'To better serve customers Illumina, Inc. is requesting to register the Genome Studio application on this machine. To register the application, please enter your user name and domain below and then select the Register button.' Below this are six input fields: 'User Name:', 'Domain:' (with 'ILLUMINA' entered), 'Full Name:', 'Email:', 'Company Name:', and 'Phone Number:'. At the bottom right are 'Register' and 'Cancel' buttons.

Figure 6 GenomeStudio Registration Dialog Box



GenomeStudio automatically recognizes your user name and domain.

2. Enter the following information into the appropriate fields in the Registration dialog box (Figure 7):
 - Full Name
 - Email Address
 - Company Name
 - Phone Number

Registration

illumina

To better serve customers Illumina, Inc. is requesting to register the Genome Studio application on this machine. To register the application, please enter your user name and domain below and then select the Register button.

User Name: Domain:

Full Name: Email:

Company Name: Phone Number:

All fields are required. Please enter all highlighted fields and select Register.

Figure 7 GenomeStudio Installed

3. Click **Register**.
GenomeStudio is installed on your computer.
4. Click **OK**.
GenomeStudio launches and opens to the main window (Figure 8).

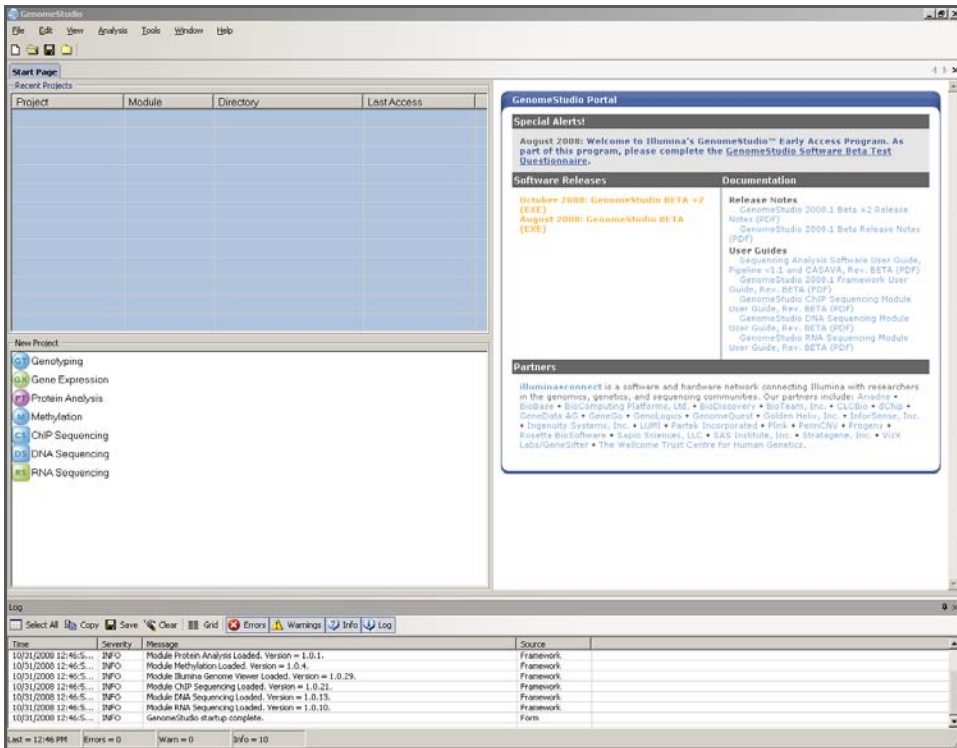


Figure 8 GenomeStudio Main Window

Verifying Your Version Numbers

To verify the version number of the GenomeStudio Framework and any GenomeStudio modules you have installed, do the following:

- ▶ Select **Help | About GenomeStudio**.

The GenomeStudio About dialog box appears (Figure 9).

The version number of the GenomeStudio Framework appears in the top portion of the GenomeStudio About dialog box.

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

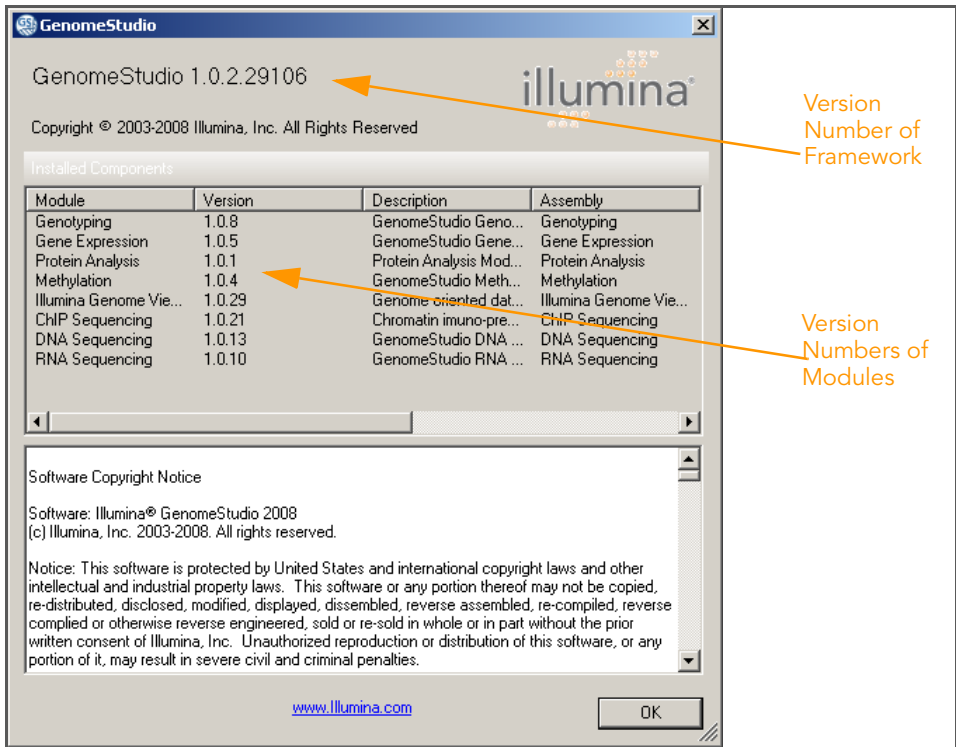


Figure 9 About GenomeStudio

Closing GenomeStudio

To exit GenomeStudio, do either of the following:

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

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 GenomeStudio 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 GenomeStudio interface, known as detachable docking windows. All GenomeStudio modules have detachable docking windows.

Working with Detachable Docking Windows

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

Use the docking icons shown in Figure 10 through Figure 13 to reposition GenomeStudio's detachable docking windows.

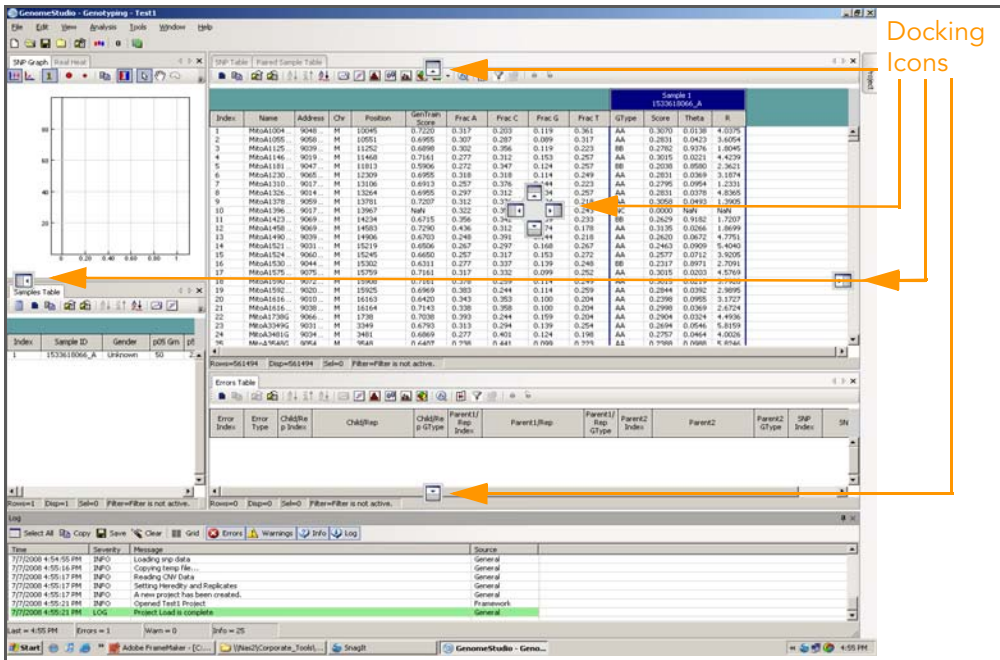


Figure 10 Docking Icons

Use the following techniques to detach and dock a window:

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

GenomeStudio displays icons (shown in Figure 10) that indicate potential docking positions and locations for the window. When you drag a window over a docking icon, GenomeStudio 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.

GenomeStudio'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 GenomeStudio 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 11) 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.

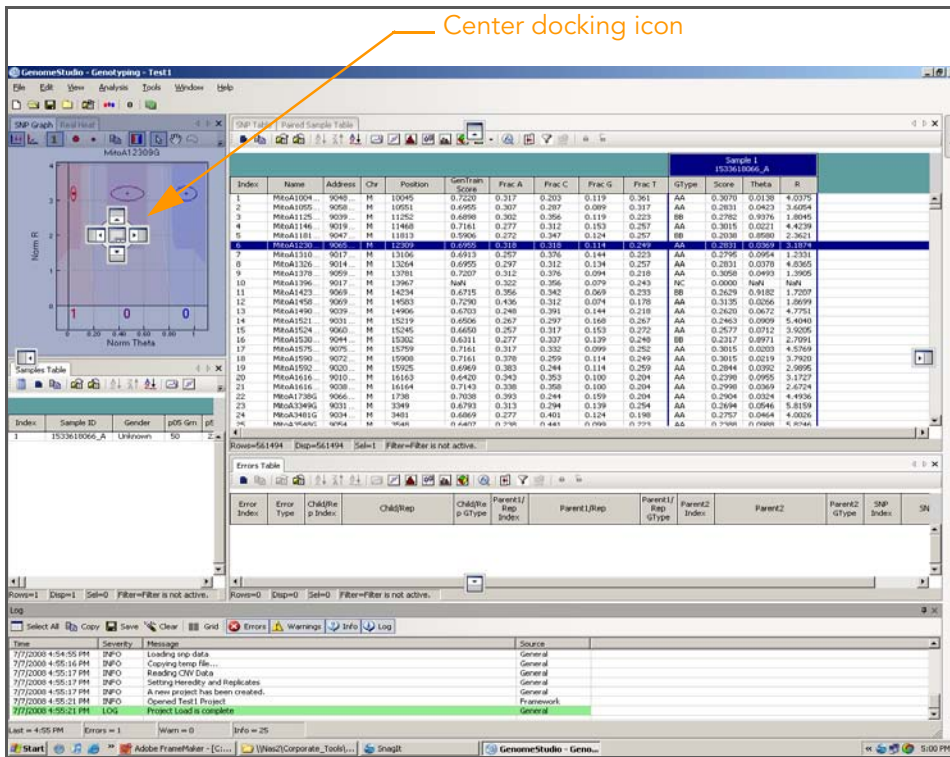



Figure 11 Docking a Window Over Another Window

To dock a window to the right of another window:

- ▶ Drag the mouse over the right docking icon , as shown in Figure 12, and release the mouse button.

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

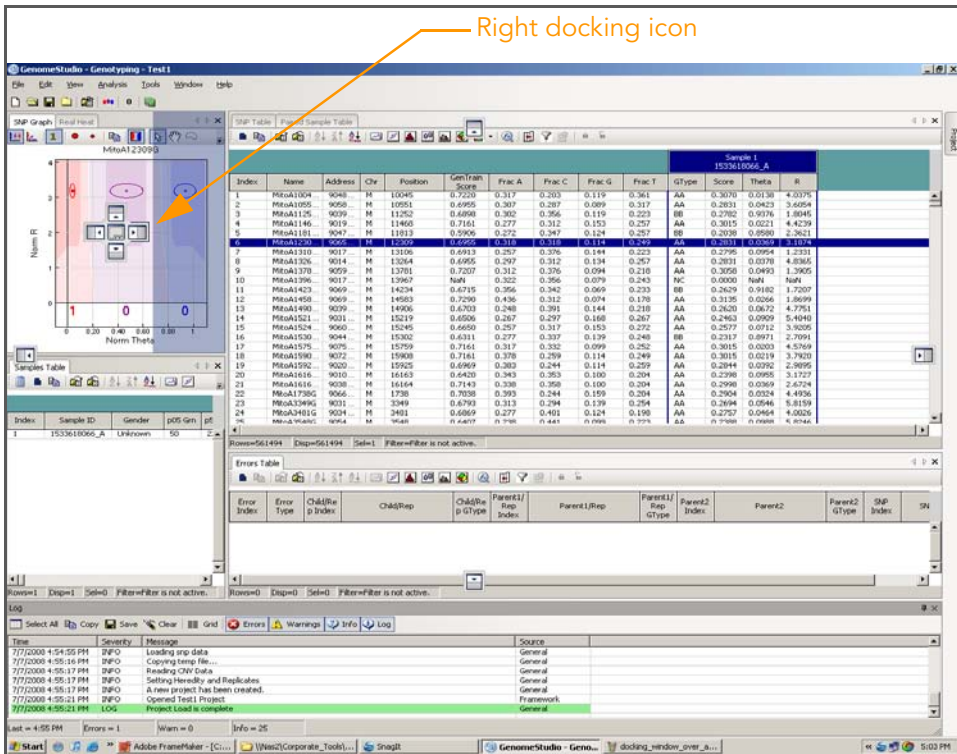



Figure 12 Docking a Window to the Right of Another Window

To dock a window to the left of another window:

- ▶ Drag the mouse over the left docking icon , as shown in Figure 12, 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 13, and release the mouse button.

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

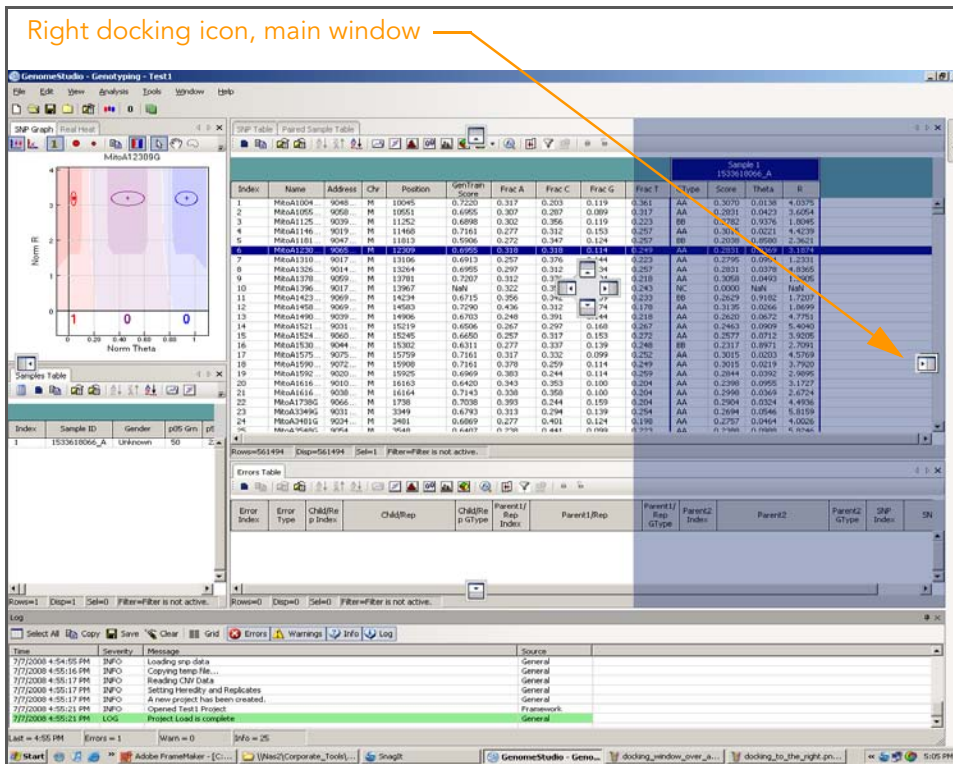


Figure 13 Docking a Window to Occupy Side of Main Window

Saving Your Window Configuration

When you quit GenomeStudio, your window configuration is saved. The next time you start GenomeStudio, 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 14).

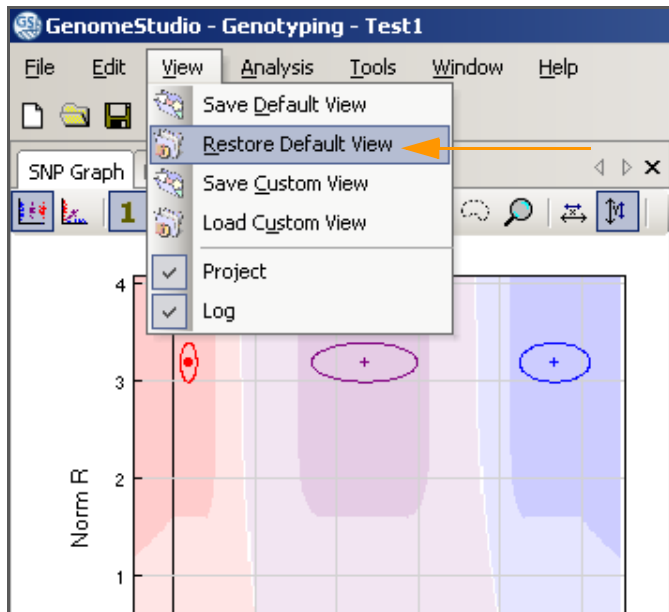


Figure 14 Restoring the Default View

Tables

Topics

- 20 Introduction
- 20 Working with Tables
 - 21 Selecting Rows
 - 22 Selecting Columns
 - 24 Searching Within Tables
 - 26 Marking Table Rows
 - 28 Clearing Marks
 - 29 Exporting to a File
 - 29 Importing a Column
 - 31 Sorting by Column
 - 32 Creating a New Column
 - 35 Editing a User-Defined Subcolumn
 - 36 Deleting a User-Defined Subcolumn
 - 38 Filtering Rows
 - 43 Locking Columns
 - 45 Unlocking Columns

Introduction

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

Working with Tables

There are two types of tables: with and without subcolumns. For example, in the GenomeStudio Genotyping Module, the **SNP Table** (Figure 15) does not have subcolumns. The **Full Data Table** (Figure 16) does have subcolumns.



Tables with subcolumns have the same subcolumns for each column.

Index	Name	Chr	Position	ChiTest100	Het Excess	AA Freq	AB Freq	BB Freq	Call Freq	Minor Freq	Aux	P-C Errors	P-P-C Errors	Rep Errors	10% GC	50% GC
1	MikoA1004...	M	10045	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
2	MikoA1055...	M	10551	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
3	MikoA1125...	M	11252	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
4	MikoA1146...	M	11468	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
5	MikoA1181...	M	11813	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
6	MikoA1230...	M	12309	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
7	MikoA1310...	M	13106	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
8	MikoA1326...	M	13264	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
9	MikoA1378...	M	13781	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
10	MikoA1396...	M	13967	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
11	MikoA1423...	M	14234	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
12	MikoA1458...	M	14583	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
13	MikoA1490...	M	14906	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
14	MikoA1521...	M	15219	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
15	MikoA1524...	M	15245	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
16	MikoA1530...	M	15302	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
17	MikoA1575...	M	15759	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
18	MikoA1590...	M	15908	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
19	MikoA1592...	M	15925	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
20	MikoA1616...	M	16163	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
21	MikoA1616...	M	16164	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
22	MikoA1738G	M	1738	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
23	MikoA3349G	M	3349	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
24	MikoA3481G	M	3481	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000
25	MikoA3548G	M	3548	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0.0000	0	0	0	0	0.0000	0.0000

Figure 15 Table without Subcolumns


In Figure 16, the subcolumns for each sample column include **GType**, **Score**, **Theta**, and **R**.

										Sample 1 1533616066_A			
Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MitoA11004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MitoA11055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331
8	MitoA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2631	0.0378	4.8365
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905
10	MitoA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	MitoA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	MitoA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	MitoA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751
14	MitoA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040
15	MitoA1524...	9060...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	MitoA1530...	9044...	M	15302	0.6311	0.277	0.337	0.139	0.248	BB	0.2317	0.8971	2.7091
17	MitoA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MitoA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920
19	MitoA1592...	9020...	M	15925	0.6969	0.383	0.244	0.114	0.259	AA	0.2844	0.0392	2.9895
20	MitoA1616...	9010...	M	16163	0.6420	0.343	0.353	0.100	0.204	AA	0.2398	0.0955	3.1727
21	MitoA1616...	9038...	M	16164	0.7143	0.338	0.358	0.100	0.204	AA	0.2998	0.0369	2.6724
22	MitoA1738G	9066...	M	1738	0.7038	0.393	0.244	0.159	0.204	AA	0.2904	0.0324	4.4936
23	MitoA3349G	9031...	M	3349	0.6793	0.313	0.294	0.139	0.254	AA	0.2694	0.0546	5.8159
24	MitoA3481G	9034...	M	3481	0.6869	0.277	0.401	0.124	0.198	AA	0.2757	0.0464	4.0026
25	MitoA3548C	9054...	M	3548	0.6407	0.298	0.441	0.099	0.223	AA	0.2988	0.0688	5.8246

Figure 16 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 17).


The screenshot shows a software window titled 'Full Data Table' with a toolbar and a data table. The table has columns for Index, Name, Address, Chr, Position, GenTrain Score, Frac A, Frac C, Frac G, Frac T, GType, Score, Theta, and R. The 'GType' column is highlighted in yellow. The data rows show various mitochondrial DNA (MitoA) samples with their corresponding scores and fractions.

Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331
8	MitoA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2831	0.0378	4.8365
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905
10	MitoA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	MitoA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	MitoA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	MitoA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751
14	MitoA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040
15	MitoA1524...	9060...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	MitoA1530...	9044...	M	15302	0.6311	0.277	0.337	0.139	0.246	BB	0.2317	0.8971	2.7091
17	MitoA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MitoA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920
19	MitoA1592...	9020...	M	15925	0.6969	0.383	0.244	0.114	0.259	AA	0.2844	0.0392	2.9895
20	MitoA1616...	9010...	M	16163	0.6420	0.343	0.353	0.100	0.204	AA	0.2398	0.0955	3.1727
21	MitoA1616...	9038...	M	16164	0.7143	0.338	0.358	0.100	0.204	AA	0.2998	0.0369	2.6724
22	MitoA1738G	9066...	M	1738	0.7038	0.393	0.244	0.159	0.204	AA	0.2904	0.0324	4.4936
23	MitoA3349G	9031...	M	3349	0.6793	0.313	0.294	0.139	0.254	AA	0.2694	0.0546	5.8159
24	MitoA3481G	9034...	M	3481	0.6869	0.277	0.401	0.124	0.198	AA	0.2757	0.0464	4.0026
25	MitoA3548C	9054...	M	3548	0.6407	0.238	0.441	0.094	0.223	AA	0.2388	0.0688	5.8246

Figure 17 Table with 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 18).

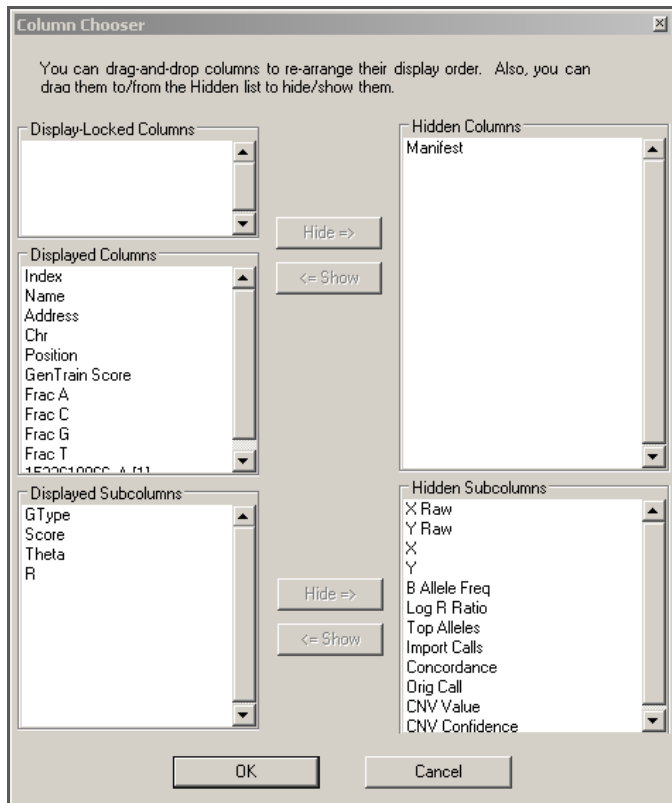


Figure 18 Column Chooser

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 and Hidden Columns areas.

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 19).

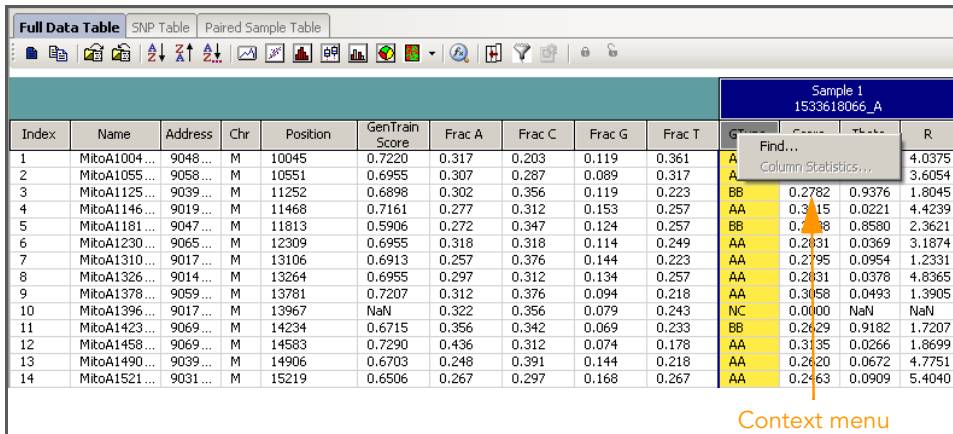


Figure 19 Tables Context Menu

3. Select **Find** from the context menu.
The Find Row dialog box appears (Figure 20).

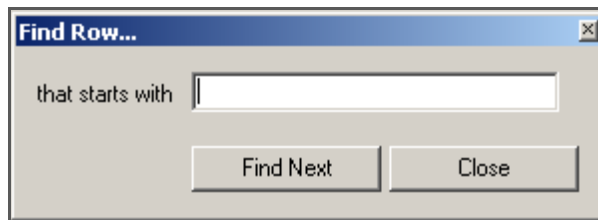


Figure 20 Find Row

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 21).

that starts with 0.91

Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MitoA1055...	9058...	M	10045	0.7220	0.317	0.203	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MitoA1125...	9039...	M	10045	0.7220	0.317	0.203	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MitoA1146...	9019...	M	10045	0.7220	0.317	0.203	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MitoA1181...	9047...	M	10045	0.7220	0.317	0.203	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MitoA1230...	9065...	M	10045	0.7220	0.317	0.203	0.114	0.249	AA	0.2831	0.0369	3.1874
7	MitoA1310...	9017...	M	10045	0.7220	0.317	0.203	0.144	0.223	AA	0.2795	0.0954	1.2331
8	MitoA1326...	9014...	M	10045	0.7220	0.317	0.203	0.134	0.257	AA	0.2831	0.0378	4.8365
9	MitoA1378...	9059...	M	10045	0.7220	0.317	0.203	0.094	0.218	AA	0.3058	0.0493	1.3905
10	MitoA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	MitoA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	MitoA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	MitoA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.672	4.7751
14	MitoA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.909	5.4040
15	MitoA1524...	9060...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	MitoA1530...	9044...	M	15302	0.6311	0.277	0.337	0.139	0.248	BB	0.2317	0.8971	2.7091
17	MitoA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MitoA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920
19	MitoA1592...	9020...	M	15925	0.6969	0.383	0.244	0.114	0.259	AA	0.2844	0.0392	2.9895
20	MitoA1616...	9010...	M	16163	0.6420	0.343	0.353	0.100	0.204	AA	0.2395	0.0955	3.1727
21	MitoA1616...	9038...	M	16164	0.7143	0.338	0.358	0.100	0.204	AA	0.2968	0.0369	2.6724
22	MitoA1738G	9066...	M	1738	0.7038	0.393	0.244	0.159	0.204	AA	0.2994	0.0324	4.4936
23	MitoA3349G	9031...	M	3349	0.6793	0.313	0.294	0.139	0.254	AA	0.2794	0.0546	5.8159
24	MitoA3481G	9034...	M	3481	0.6869	0.277	0.401	0.124	0.198	AA	0.2757	0.0464	4.0026
25	MitoA3548C	9054...	M	3548	0.6407	0.238	0.441	0.099	0.223	AA	0.2788	0.0488	5.8246

Rows=561494 Disp=561494 Sel=1 Filter=Filter is not active.

Search term = 0.91

First instance of 0.91

Figure 21 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 22).

Full Data Table											Sample 1 1533618066_A		
Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874
7	M			106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331
8	M			264	0.6955	0.297	0.312	0.134	0.257	AA	0.2831	0.0378	4.8365
9	M			781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905
10	M			967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	M			234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	M			583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	M			906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751
14	M			219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040
15	M			245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	M			302	0.6311	0.277	0.337	0.139	0.248	BB	0.2317	0.8971	2.7091
17	MitoA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MitoA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920
19	MitoA1592...	9020...	M	15925	0.6969	0.363	0.244	0.114	0.259	AA	0.2844	0.0392	2.9895

Figure 22 Configure Marks Selected

The Configure Marks dialog box appears (Figure 23).

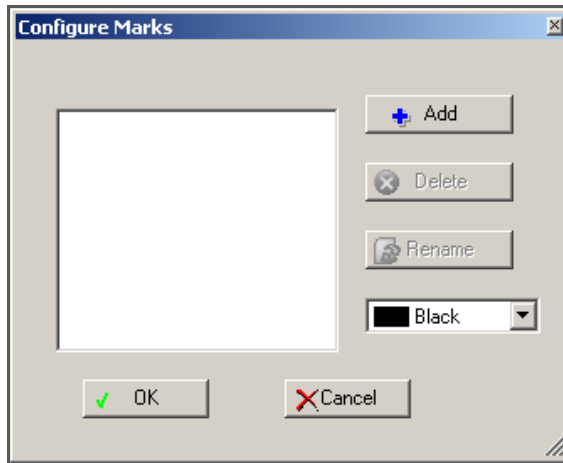


Figure 23 Configuring Marks

4. Click **Add**.
The Select Mark Name dialog box appears (Figure 24).

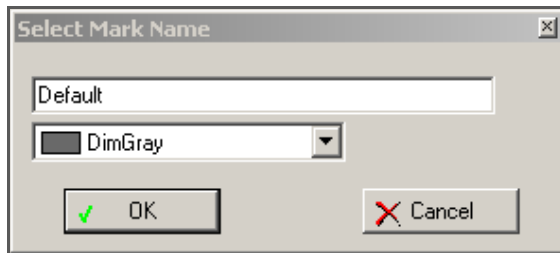


Figure 24 Selecting Mark Name

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

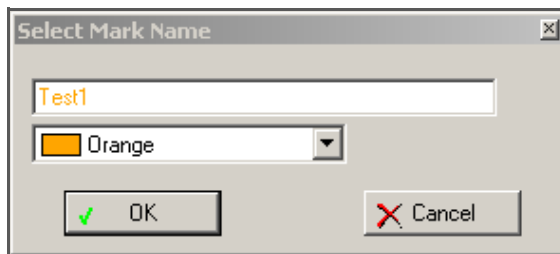


Figure 25 Selecting Mark Color

7. Click **OK**.

The mark is displayed in the color you chose (Figure 26).

Full Data Table											Sample 1 1533618066_A			
Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R	
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375	
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054	
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045	
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239	
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621	
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874	
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331	
8	MitoA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2831	0.0378	4.8365	
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905	

Figure 26 Selected Rows Marked in Table

Clearing Marks

To clear any marks in a table:

1. Select **Clear Marks** from the context menu (Figure 27).

Full Data Table											Sample 1 1533618066_A			
Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R	
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375	
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054	
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045	
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239	
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621	
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874	
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331	
8	MitoA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2831	0.0378	4.8365	
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905	
10	MitoA1397...	9067...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN	
11	MitoA1423...	9071...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207	
12	MitoA1458...	9079...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699	
13	MitoA1490...	9073...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751	
14	MitoA1521...	9076...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040	
15	MitoA1524...	9077...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205	
16	MitoA1530...	9078...	M	15302	0.6311	0.277	0.337	0.139	0.248	BB	0.2317	0.8971	2.7091	
17	MitoA1716...	9081...	M	17161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769		
18	MitoA1716...	9081...	M	17161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920		
19	MitoA1592...	9020...	M	15921	0.6969	0.383	0.244	0.114	0.259	AA	0.2844	0.0392	2.9895	
20	MitoA1616...	9010...	M	16163	0.6420	0.343	0.353	0.100	0.204	AA	0.2398	0.0955	3.1727	
21	MitoA1616...	9038...	M	16164	0.7143	0.338	0.358	0.100	0.204	AA	0.2998	0.0369	2.6724	

Figure 27 Clearing Selected Marks from 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 GenomeStudio 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** field, enter a name for the file.
3. Click **Save**.

If there are rows selected in the table, the following prompt appears:

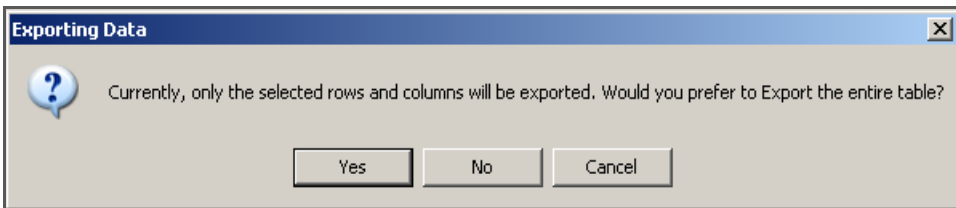


Figure 28 Exporting Data

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 GenomeStudio from a preexisting file.

To import a column or a subcolumn:

1. Click  **Import Columns**.

The Import dialog box appears (Figure 29).

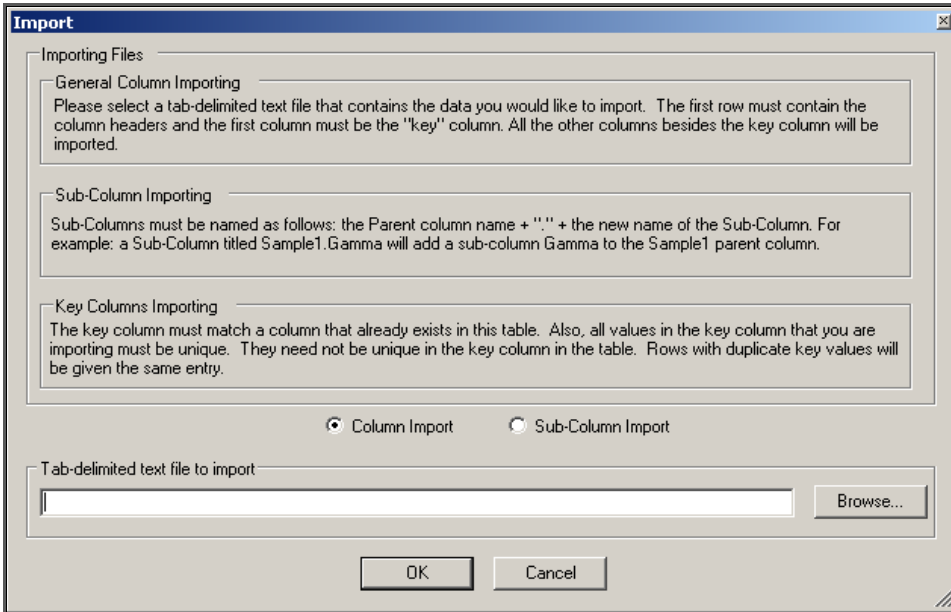


Figure 29 Importing Data

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 appears 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 open the Sort dialog box (Figure 30).

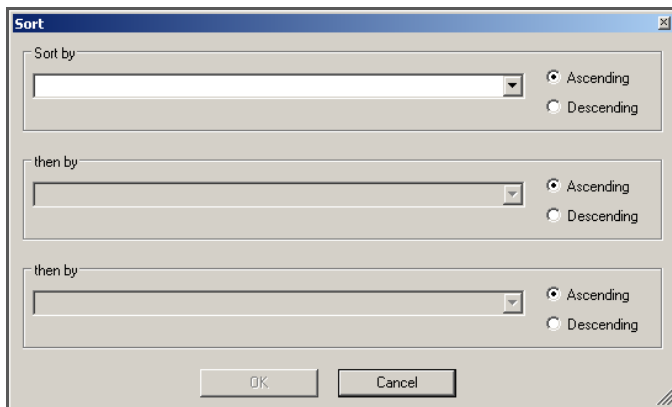


Figure 30 Multiple-Column Sort

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 GenomeStudio Framework allows you to create new columns and add them to a table. 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 31).

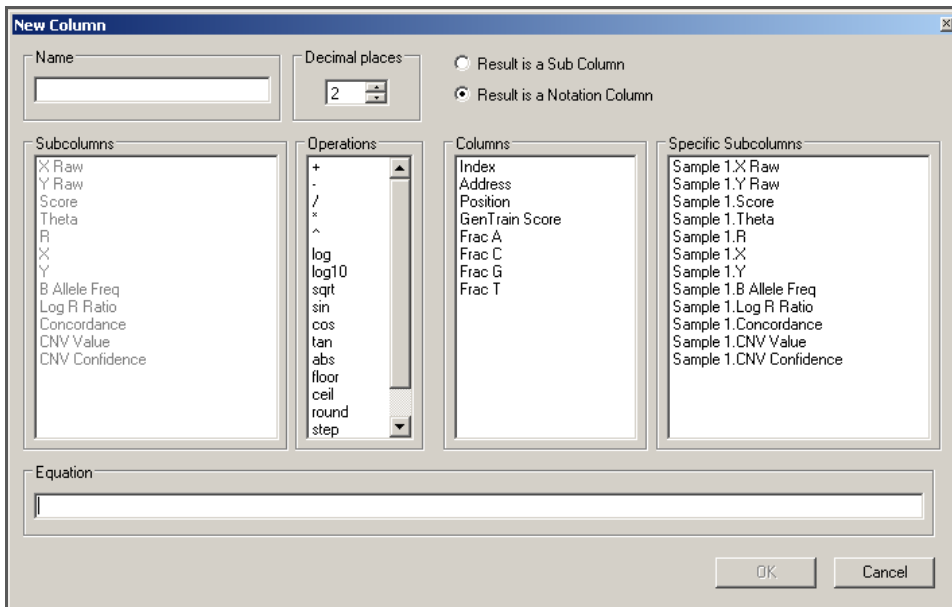


Figure 31 Creating a New Column

2. In the **Name** 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
sqrt	Square Root
sin	Sine
cos	Cosine
tan	Tangent
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

Table 2 New Column Operations & Descriptions

Operation	Description
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 field (Figure 32).

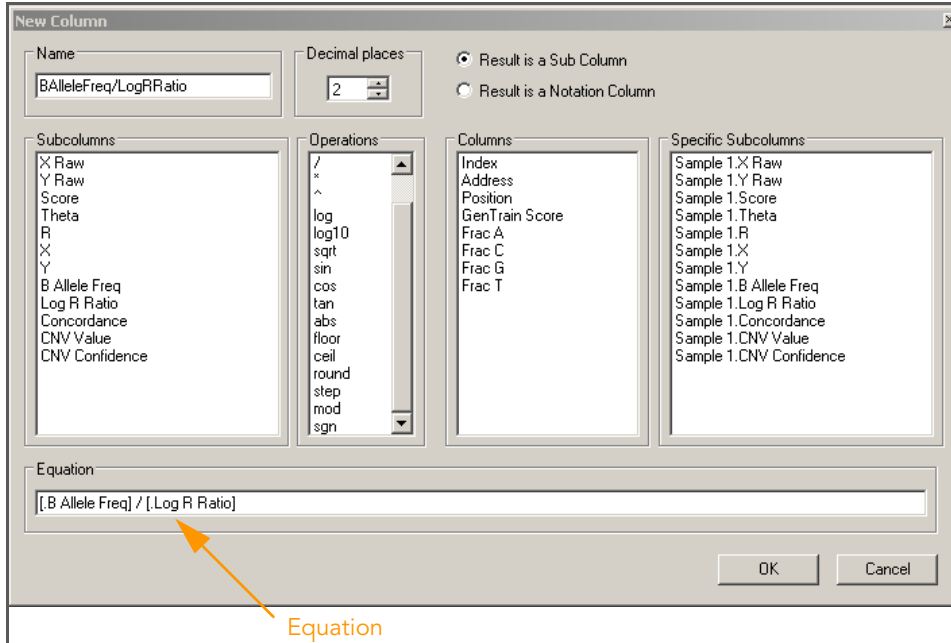


Figure 32 Building an Equation

6. Click **OK**.

The new column is added to the table for every sample (Figure 33).

New Subcolumn

										Sample 1 1533618066_A				
Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R	AlleleFreq/LogR Ratio
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375	NaN
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054	NaN
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045	Infinity
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239	NaN
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621	Infinity
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874	NaN
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331	NaN
8	MitoA1335...	9017...	M	13264	0.6955	0.297	0.312	0.184	0.257	AA	0.2331	0.0378	4.8365	NaN
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905	NaN
10	MitoA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN	NaN
11	MitoA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207	Infinity
12	MitoA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699	NaN
13	MitoA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751	NaN
14	MitoA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040	NaN

Figure 33 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 34) from the context menu.

Sample 1 1533618066_A				
GType	Score	Theta	R	AlleleFreq/LogR Ratio
AA	0.30			
AA	0.28			
BB	0.27			
AA	0.30			
BB	0.20			
AA	0.28			
AA	0.2795	0.0954	1.2331	NaN
AA	0.2831	0.0378	4.8365	NaN
AA	0.3058	0.0493	1.3905	NaN
NC	0.0000	NaN	NaN	NaN
BB	0.2629	0.9182	1.7207	Infinity
AA	0.3135	0.0266	1.8699	NaN
AA	0.2620	0.0672	4.7751	NaN
AA	0.2463	0.0909	5.4040	NaN

Figure 34 Editing a User-Defined Subcolumn

The Edit Column dialog box appears (Figure 35).

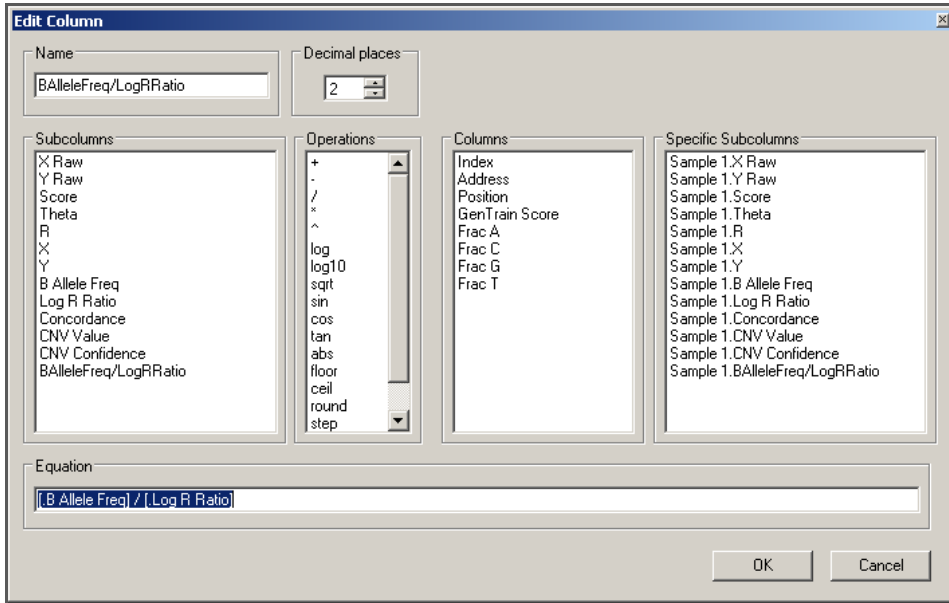


Figure 35 Changing Subcolumn Attributes

3. In the Edit Column dialog box, edit the subcolumn by changing the subcolumn attributes.
4. Click **OK**.
The changes you made to the subcolumn are 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 1533618066_A			
GType	Score	Theta	R
AA	0.3070	0.0138	4.0375
AA	0.2831	0.0423	3.6054
BB	0.2782	0.9376	1.8045
AA	0.3015	0.0221	4.4239
BB	0.2038	0.8580	2.3621
AA	0.2831	0.0369	3.1874
AA	0.2795	0.0954	1.2331
AA	0.2831	0.0378	4.8365
AA	0.3058	0.0493	1.3905
NC	0.0000	NaN	NaN
BB	0.2629	0.9182	1.7207

Figure 36 Deleting a Column

A dialog box appears (Figure 37).

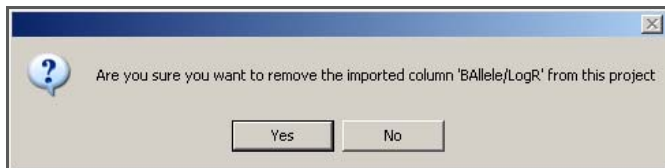


Figure 37 Delete Subcolumn

3. Click **Yes**.

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

Sample 1 1533618066_A			
GType	Score	Theta	R
AA	0.3070	0.0138	4.0375
AA	0.2831	0.0423	3.6054
BB	0.2782	0.9376	1.8045
AA	0.3015	0.0221	4.4239
BB	0.2038	0.8580	2.3621
AA	0.2831	0.0369	3.1874
AA	0.2795	0.0954	1.2331
AA	0.2831	0.0378	4.8365
AA	0.3058	0.0493	1.3905
NC	0.0000	NaN	NaN
BB	0.2629	0.9182	1.7207

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).

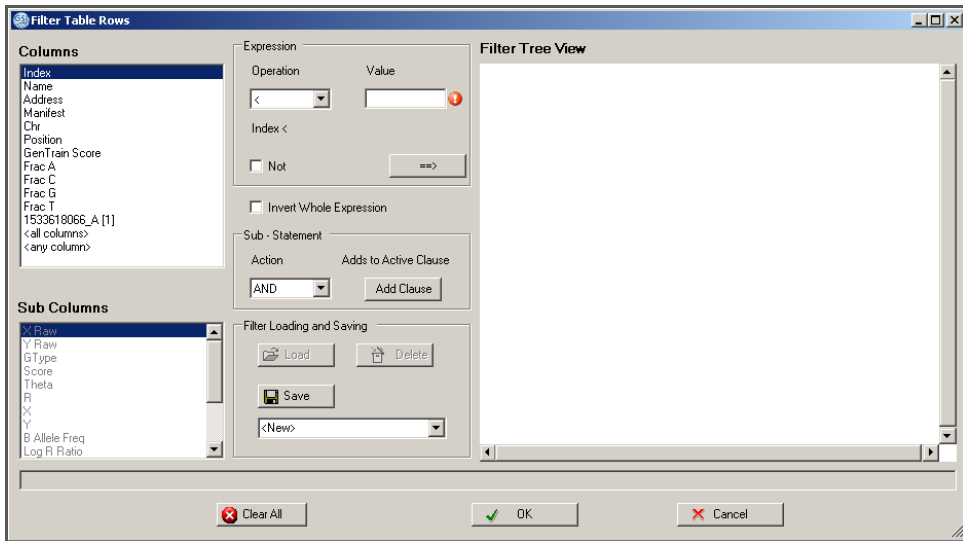
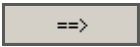


Figure 39 Filtering Table Rows

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 list.
4. Enter a value in the **Value** 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**.

**NOTE**

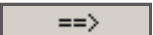
You may enter multiple expressions linked by AND, OR, etc.

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** field.
4. Click .

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

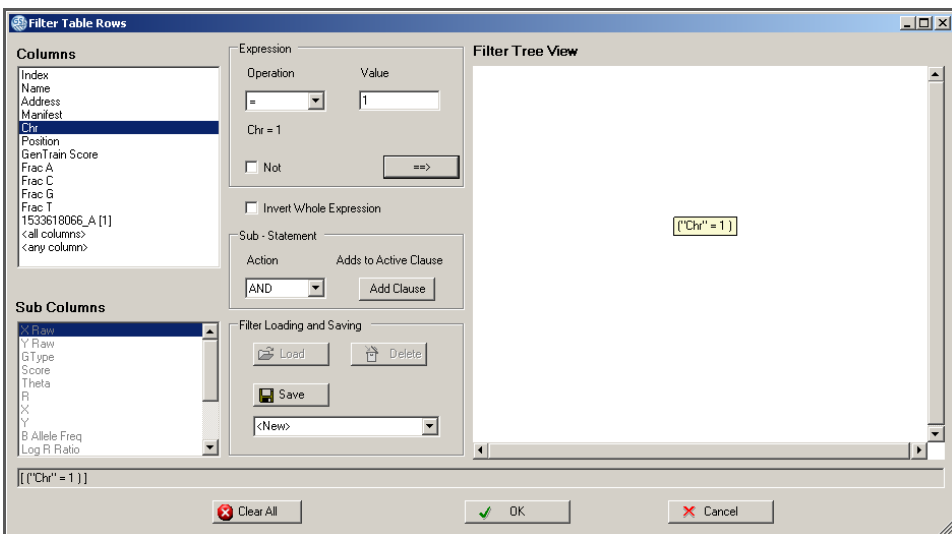

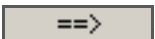


Figure 40 Creating a Simple Filter

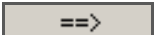
Creating a More Complex Filter

You can also create more complex boolean expressions.

For example, you can select all SNPs on chromosome 1 for which the position is less than 50000000 and the GenTrain score is less than 0.8:

1. Select the **Chr** column.
2. Select the **=** operation.
3. Enter 1 in the **Value** field.
4. Click .
5. Select **AND** from the **Action** pulldown menu.
6. Select **Position** from the **Columns** listbox.
7. Select **<** from the **Operation** listbox.
8. Type **50000000** in the **Value** field.
9. Click .

The **AND** clause you created is added to the filter.

10. Select **GenTrain Score** from the **Columns** listbox.
11. Select **>** in the **Operation** pulldown menu.
12. Type **0.8** in the **Value** field.
13. 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 ("Position" < 50000000 ) AND ("GenTrain Score" > 0.8 ) ]
```



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 only appears once you add a clause.

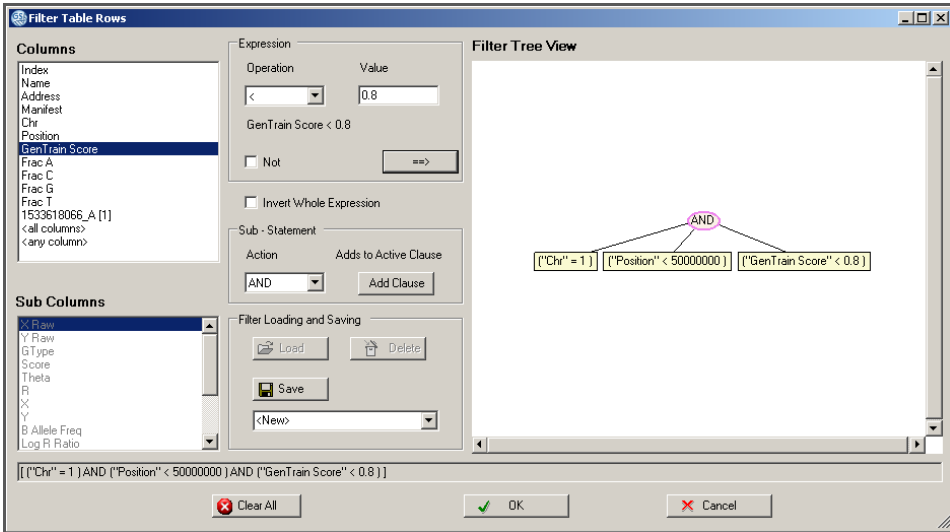


Figure 41 Creating a More Complex Filter

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 dialog box, click **Save**.

The Enter Name for Filter dialog box appears (Figure 42).

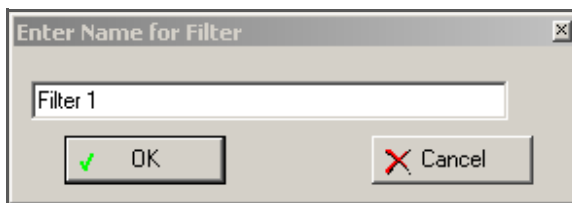


Figure 42 Entering a Name for a Filter

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



NOTE

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

4. Click **OK**.

Your filter is saved to *C:\Documents and Settings\\Application Data\Illumina\GenomeStudio\<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 dialog box, select the filter you want to use from the dropdown list.

The **Load** button becomes active.

2. Click **Load**.

Your filter is loaded and ready to use.

Clearing a Filter

To clear the filter:

- ▶ Click  **Clear Filter**.


The filter is cleared.

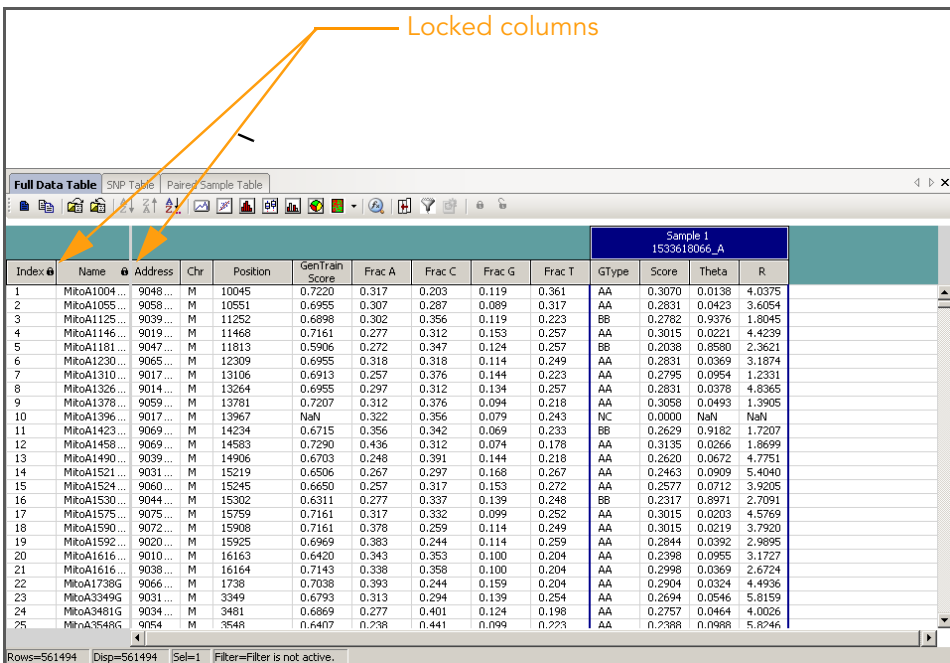
Filtering Notes

- ▶ 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 dialog box.
- ▶ 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 43).



Locked columns

Full Data Table											Sample 1 1533618066_A		
Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MiroA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MiroA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MiroA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MiroA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MiroA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MiroA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874
7	MiroA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331
8	MiroA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2931	0.0378	4.8365
9	MiroA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905
10	MiroA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	MiroA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	MiroA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	MiroA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751
14	MiroA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040
15	MiroA1524...	9060...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	MiroA1530...	9044...	M	15302	0.6311	0.277	0.337	0.139	0.246	BB	0.2317	0.8971	2.7091
17	MiroA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MiroA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920
19	MiroA1592...	9020...	M	15925	0.6969	0.383	0.244	0.114	0.259	AA	0.2844	0.0392	2.9895
20	MiroA1616...	9010...	M	16163	0.6420	0.343	0.353	0.100	0.204	AA	0.2398	0.0955	3.1727
21	MiroA1616...	9038...	M	16164	0.7143	0.338	0.358	0.100	0.204	AA	0.2998	0.0369	2.6724
22	MiroA1738G	9066...	M	1738	0.7038	0.393	0.244	0.159	0.204	AA	0.2904	0.0324	4.4936
23	MiroA3349G	9031...	M	3349	0.6793	0.313	0.294	0.139	0.254	AA	0.2694	0.0546	5.8159
24	MiroA3481G	9034...	M	3481	0.6869	0.277	0.401	0.124	0.198	AA	0.2757	0.0464	4.0026
25	MiroA3548G	9054...	M	3548	0.6407	0.238	0.441	0.099	0.223	AA	0.2388	0.0488	5.8246

Rows=561494 Disp=561494 Sel=1 Filter=Filter is not active.

Figure 43 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 44).

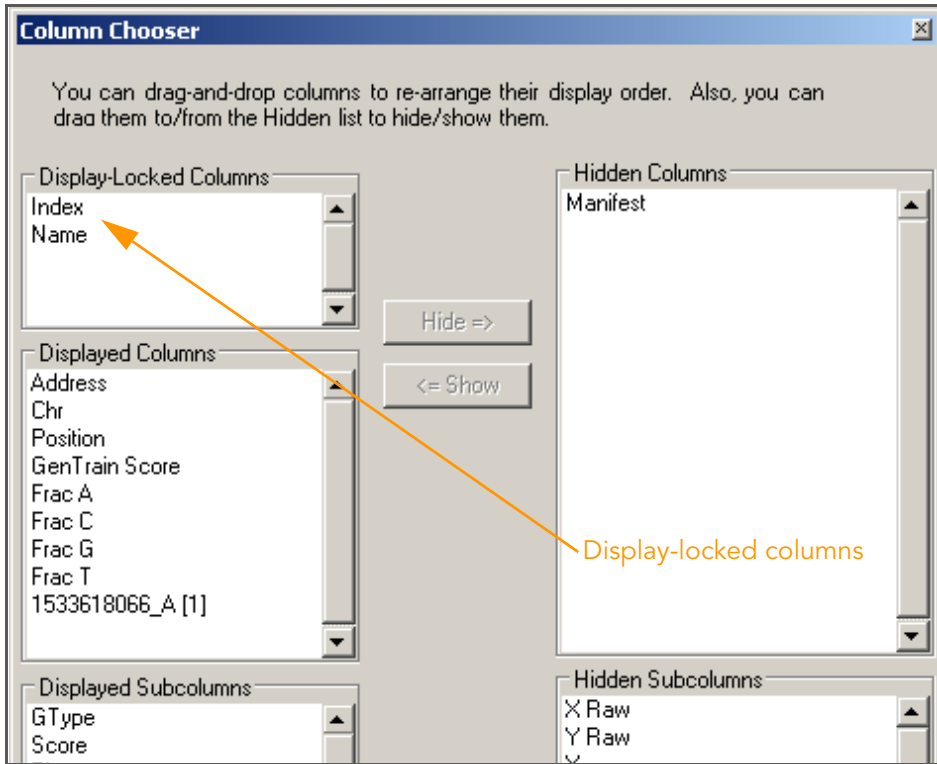


Figure 44 Column Chooser, Display-Locked Columns

Unlocking Columns

To unlock columns:

1. Select the column header of each locked column you want to unlock (Figure 45).

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

Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331
8	MitoA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2831	0.0378	4.8365
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905
10	MitoA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	MitoA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	MitoA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	MitoA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751
14	MitoA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040
15	MitoA1524...	9060...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	MitoA1530...	9044...	M	15302	0.6311	0.277	0.337	0.139	0.248	BB	0.2317	0.8971	2.7091
17	MitoA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MitoA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920

Figure 45 Selecting Columns to Unlock

2. Click  **Unlock**.

The selected columns are unlocked (Figure 46).

The selected columns are unlocked.

Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GType	Score	Theta	R
1	MitoA1004...	9048...	M	10045	0.7220	0.317	0.203	0.119	0.361	AA	0.3070	0.0138	4.0375
2	MitoA1055...	9058...	M	10551	0.6955	0.307	0.287	0.089	0.317	AA	0.2831	0.0423	3.6054
3	MitoA1125...	9039...	M	11252	0.6898	0.302	0.356	0.119	0.223	BB	0.2782	0.9376	1.8045
4	MitoA1146...	9019...	M	11468	0.7161	0.277	0.312	0.153	0.257	AA	0.3015	0.0221	4.4239
5	MitoA1181...	9047...	M	11813	0.5906	0.272	0.347	0.124	0.257	BB	0.2038	0.8580	2.3621
6	MitoA1230...	9065...	M	12309	0.6955	0.318	0.318	0.114	0.249	AA	0.2831	0.0369	3.1874
7	MitoA1310...	9017...	M	13106	0.6913	0.257	0.376	0.144	0.223	AA	0.2795	0.0954	1.2331
8	MitoA1326...	9014...	M	13264	0.6955	0.297	0.312	0.134	0.257	AA	0.2831	0.0378	4.8365
9	MitoA1378...	9059...	M	13781	0.7207	0.312	0.376	0.094	0.218	AA	0.3058	0.0493	1.3905
10	MitoA1396...	9017...	M	13967	NaN	0.322	0.356	0.079	0.243	NC	0.0000	NaN	NaN
11	MitoA1423...	9069...	M	14234	0.6715	0.356	0.342	0.069	0.233	BB	0.2629	0.9182	1.7207
12	MitoA1458...	9069...	M	14583	0.7290	0.436	0.312	0.074	0.178	AA	0.3135	0.0266	1.8699
13	MitoA1490...	9039...	M	14906	0.6703	0.248	0.391	0.144	0.218	AA	0.2620	0.0672	4.7751
14	MitoA1521...	9031...	M	15219	0.6506	0.267	0.297	0.168	0.267	AA	0.2463	0.0909	5.4040
15	MitoA1524...	9060...	M	15245	0.6650	0.257	0.317	0.153	0.272	AA	0.2577	0.0712	3.9205
16	MitoA1530...	9044...	M	15302	0.6311	0.277	0.337	0.139	0.248	BB	0.2317	0.8971	2.7091
17	MitoA1575...	9075...	M	15759	0.7161	0.317	0.332	0.099	0.252	AA	0.3015	0.0203	4.5769
18	MitoA1590...	9072...	M	15908	0.7161	0.378	0.259	0.114	0.249	AA	0.3015	0.0219	3.7920

Figure 46 Selected Columns Unlocked

Graphs

Topics

- 49 Introduction
- 49 Line Plots
 - 49 Plotting a Single Column
 - 51 Plotting Two Columns
 - 52 Changing the X-Axis Label
 - 54 Changing the Plot Type
- 57 Scatter Plots
 - 58 Marking Samples
- 60 Box Plots
- 61 Frequency Plots
- 63 Pie Charts
- 64 Histograms
- 72 Heat Maps
 - 73 Populating the Heat Map with Data
 - 75 Clustering Heat Map Data
 - 76 Similarities and Distances
 - 79 Resizing Row Labels
 - 79 Resizing Row Labels

- 79 Resizing the Column Tree Cluster
- 79 Resizing Column Labels
- 80 Scrolling the Heat Map Area
- 80 Opening a Heat Map File
- 80 Saving a Heat Map
- 81 Exiting the Heat Map
- 81 Editing Heat Map Properties
- 87 Creating a Presentation Image

Introduction

The graphing functions of GenomeStudio allow you to plot data displayed in tables in a number of ways:

- ▶ Line plots
- ▶ Scatter plots
- ▶ Histograms
- ▶ Box plots
- ▶ Frequency plots
- ▶ Pie charts
- ▶ Heat maps

The following sections include information about how to use each of these graphing tools.


Line Plots

Line plots are linked to data tables. Data appears in the same order in a line plot as it does in the associated data 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 Line Plot (Figure 47).

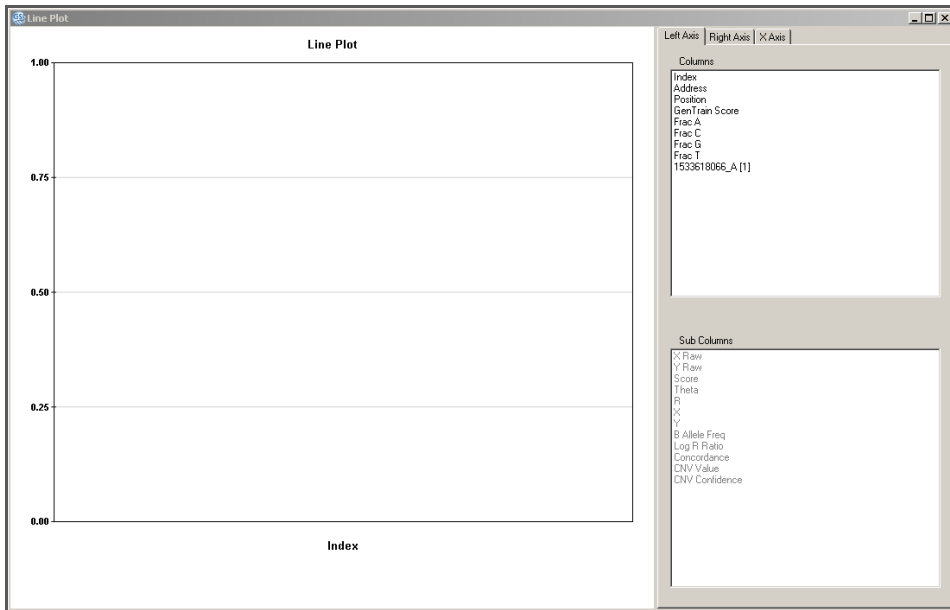


Figure 47 GenomeStudio Line Plot

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 choose (Figure 48).

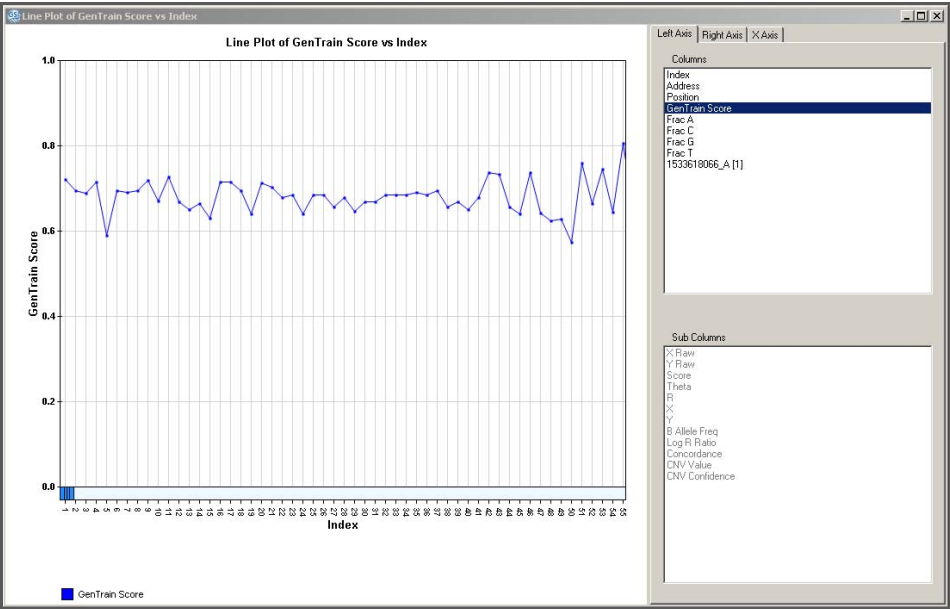


Figure 48 Plotting a Single Column in a Line Plot

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 48.
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 49).

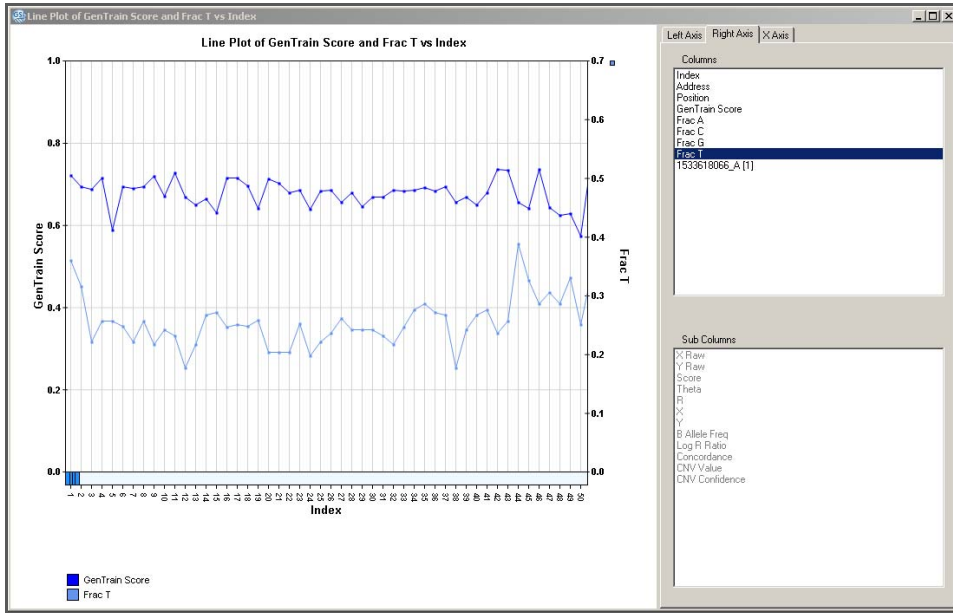


Figure 49 Plotting Two Columns in a Line Plot

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 GenomeStudio Column Plot dialog box, 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 50).

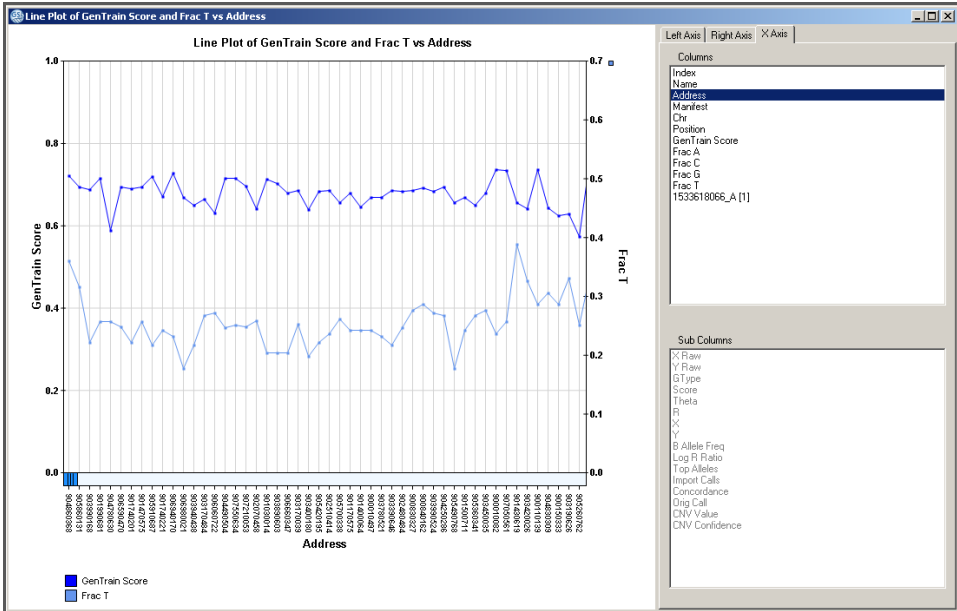


Figure 50 Line Plot with X-Axis Label "Address"

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 51).

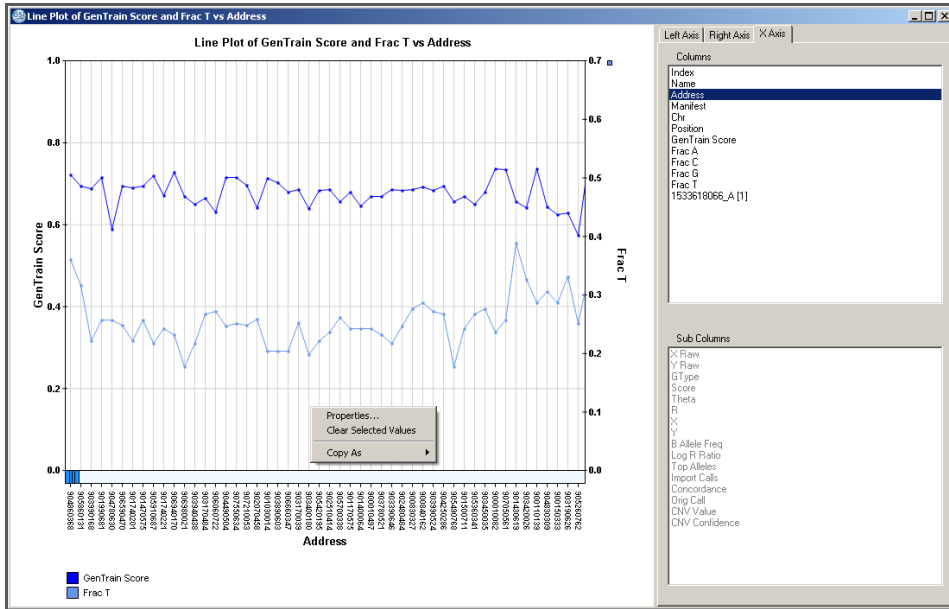


Figure 51 Line Plot Context Menu

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

3. The Plot Settings dialog box appears (Figure 52).

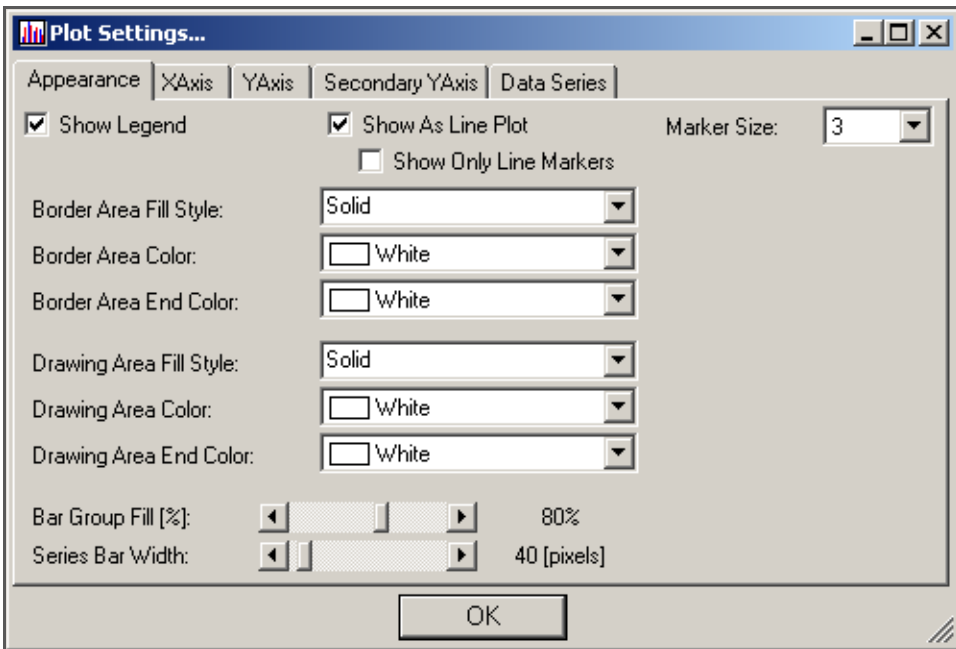


Figure 52 Plot Settings

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 53).

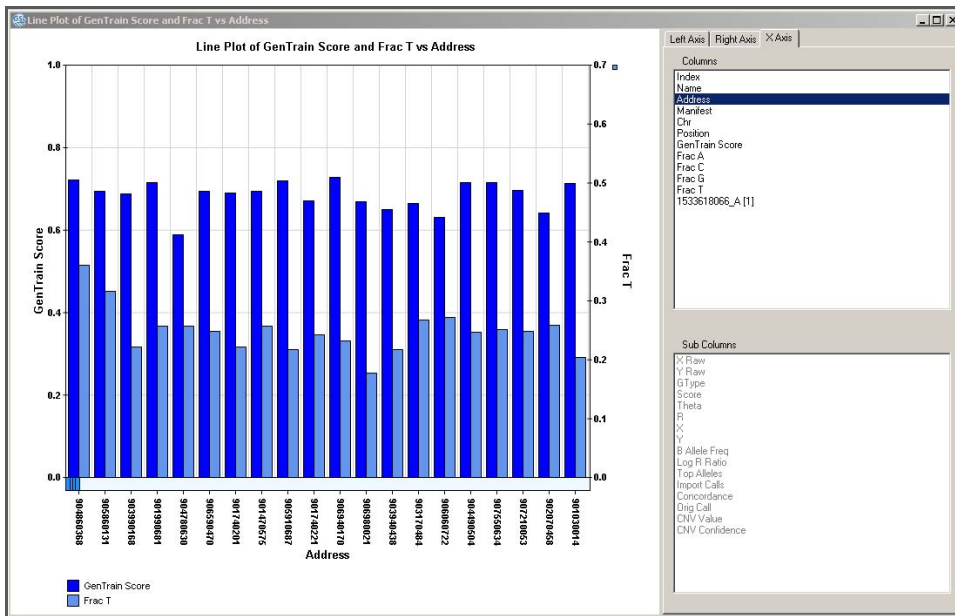



Figure 53 Bar Graph

Scatter Plots

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

1. Click  **Scatter Plot** in the table to open the Scatter Plot dialog box (Figure 54).

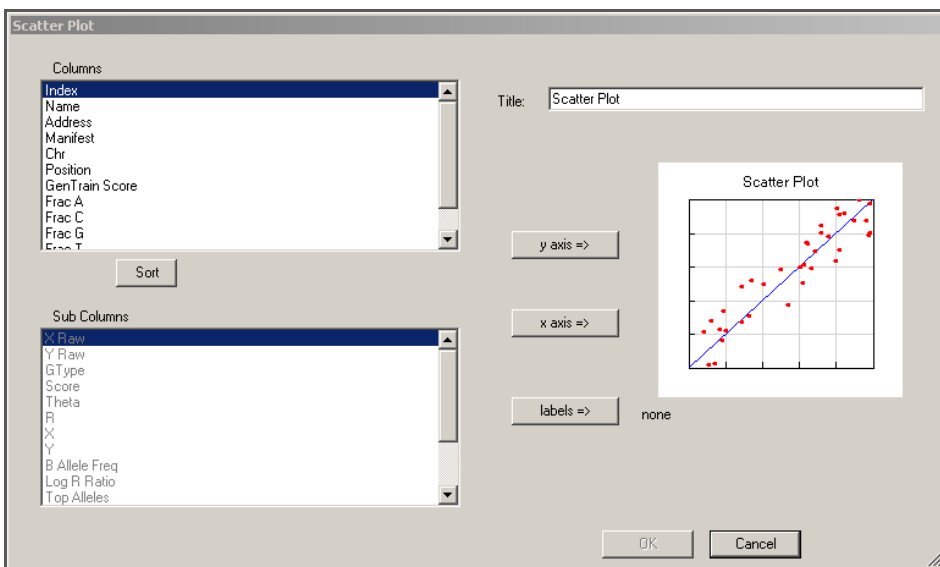


Figure 54 Scatter Plot

2. Choose graphing options from the **Columns** and **Subcolumns** dropdown lists.
3. Click **OK**.

The scatter plot appears with the parameters you selected (Figure 55).

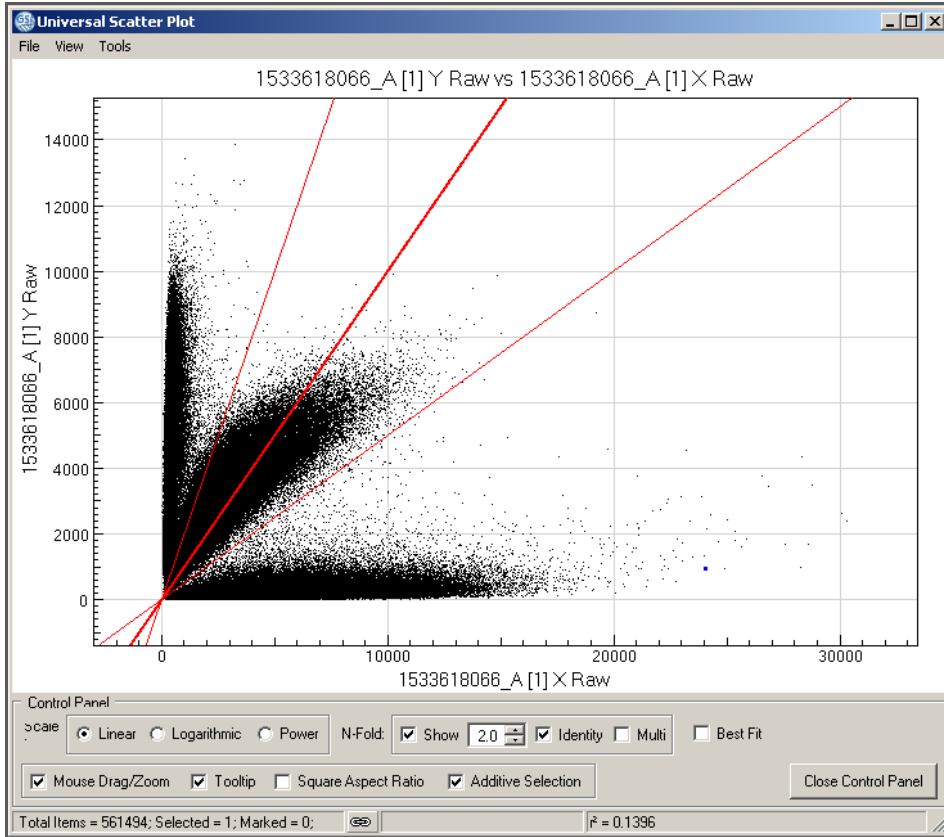


Figure 55 Universal Scatter Plot

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 56).

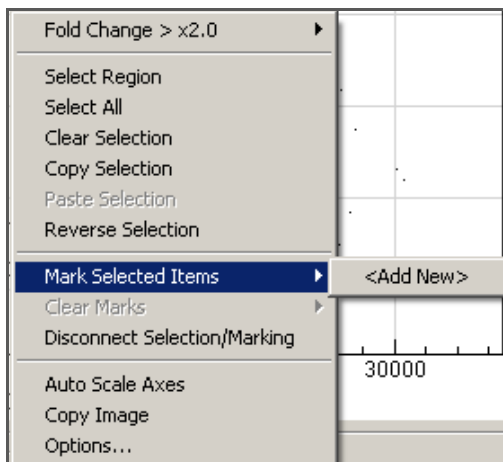


Figure 56 Scatter Plot Context Menu

The Select Mark Name dialog box appears (Figure 57).

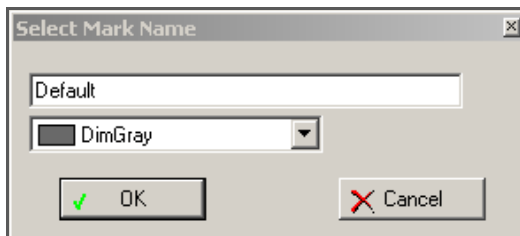


Figure 57 Select Mark Name

3. Enter a name for your mark in the field.
4. Select a color for your mark from the dropdown list (Figure 58).

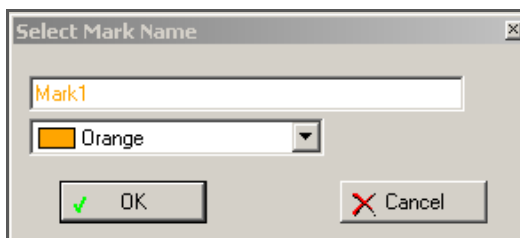



Figure 58 Selecting Mark Name & Color

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.

Box Plots

A box plot is a graphical summary of a distribution of values. Use the box plot to plot either column or subcolumn values.

1. In the table toolbar, click  **Box Plot**.

The Table Box Plot dialog box appears (Figure 59).

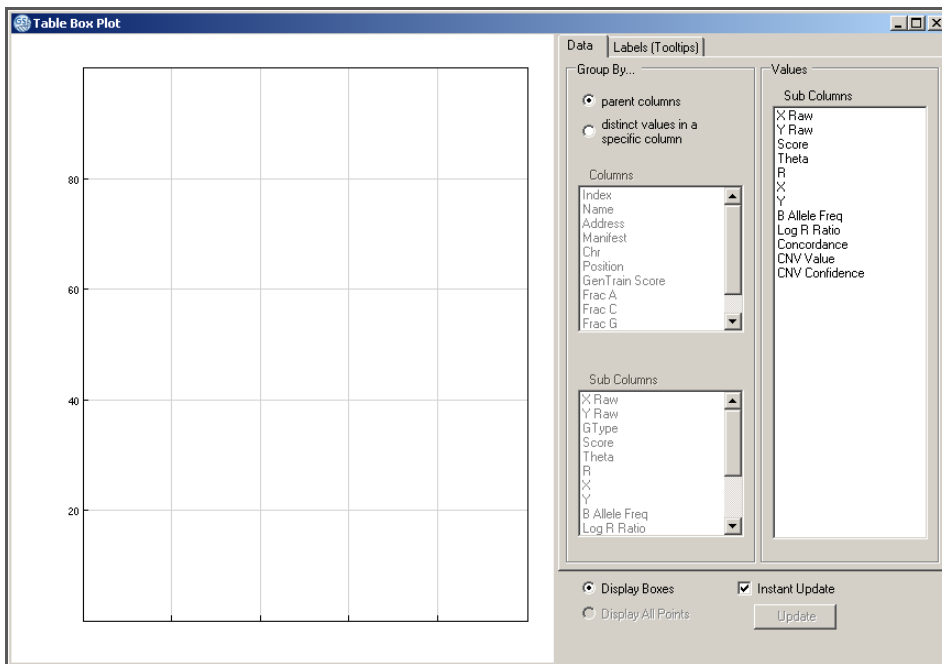


Figure 59 Box Plot

2. Click a subcolumn.
GenomeStudio calculates the distribution statistics that are displayed graphically in the box plot.
The box plot appears (Figure 60).

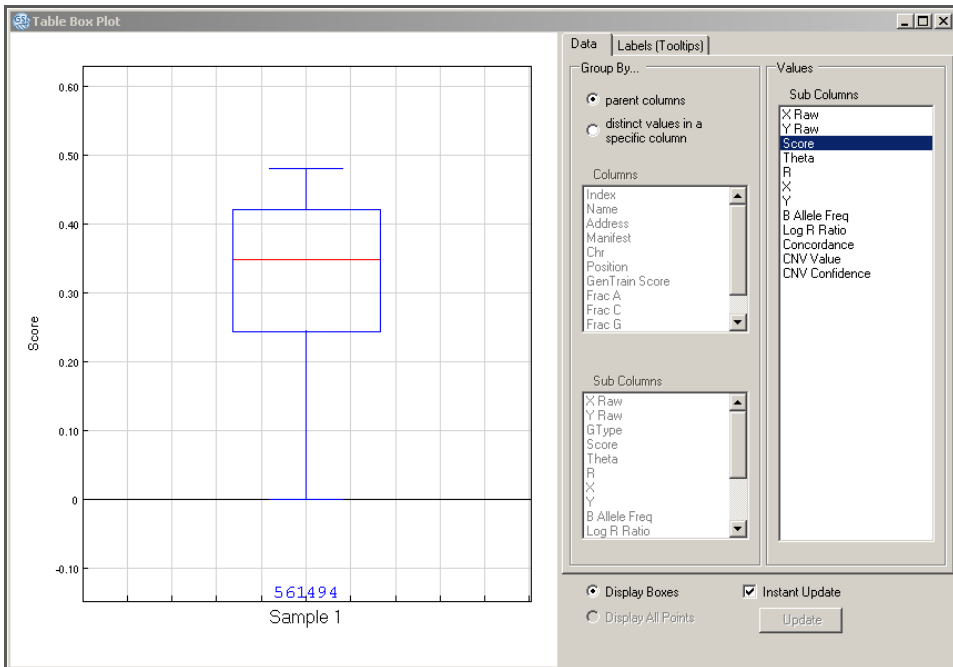



Figure 60 Box Plot Displaying Data

Frequency Plots

A frequency plot is a graphical display of how often data values occur. Use the Frequency Plot option to display a range of data values in a graphical format.

1. In the table toolbar, click  **Frequency Plot**.

The Frequency Plot dialog box appears (Figure 61).

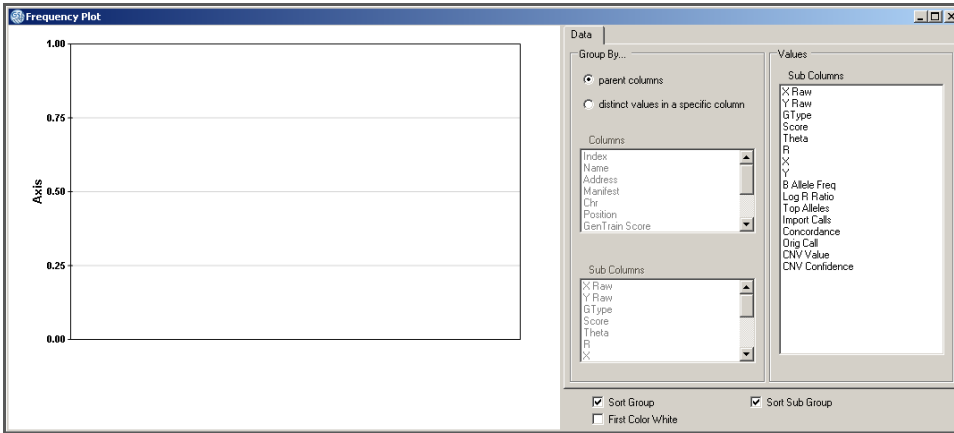


Figure 61 Frequency Plot

2. Select grouping and values options.

The Frequency Plot automatically displays your data with the selections you have chosen (Figure 62).

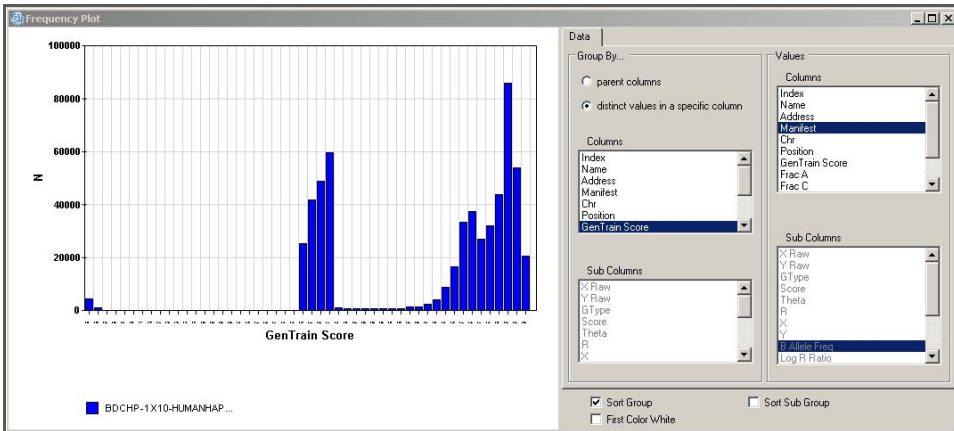



Figure 62 Frequency Plot Displaying Data

Pie Charts

A pie chart is a circular chart, divided into sectors, that illustrates the relative magnitudes of frequencies or percentages. Use the Pie Chart option to display table data in a graphical format.

1. In the table toolbar, click  **Pie Chart**.

The Pie Chart dialog box appears (Figure 63).

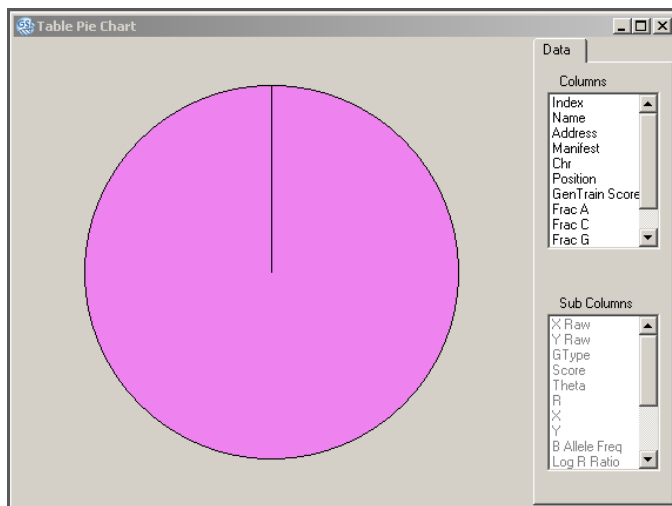


Figure 63 Pie Chart Displaying Data

2. Select the columns and subcolumns you want to display. The pie chart automatically displays your data (Figure 64).

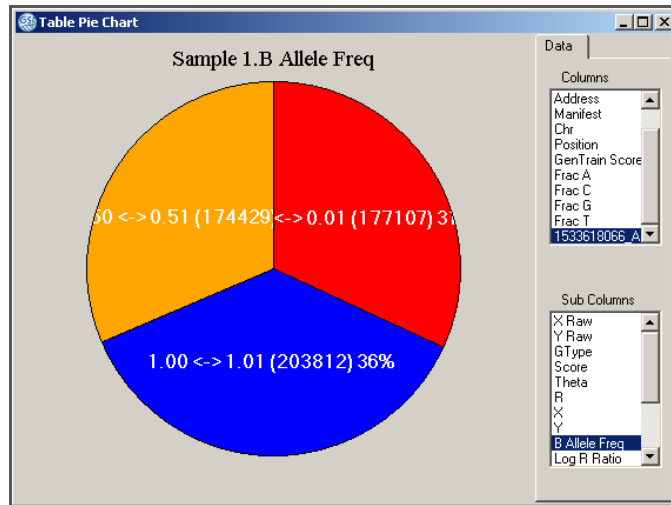



Figure 64 Pie Chart Displaying Column and Subcolumn Data

Histograms

A histogram is a graphical display of tabulated frequencies. Use the **Histogram** option to plot the distribution of values for a particular column.

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

The GenomeStudio Histogram dialog box appears (Figure 65).

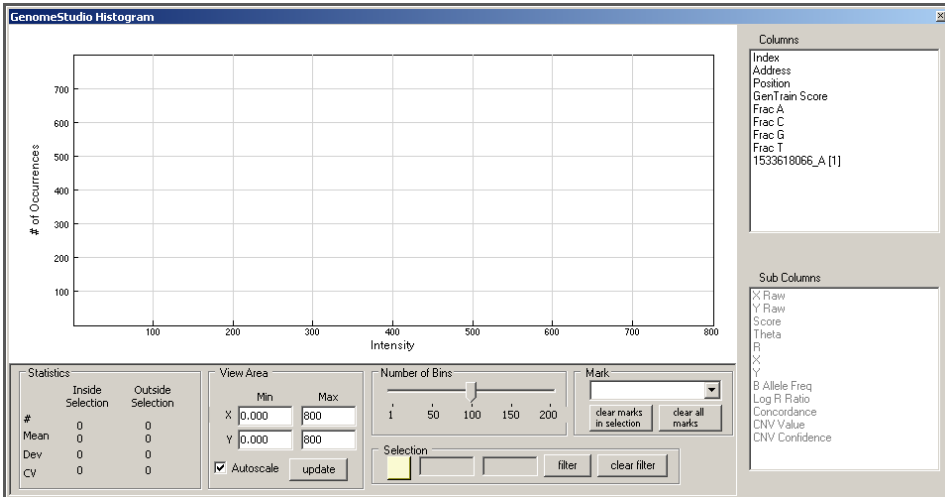


Figure 65 Histogram

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

A histogram displaying the options you selected appears (Figure 66).

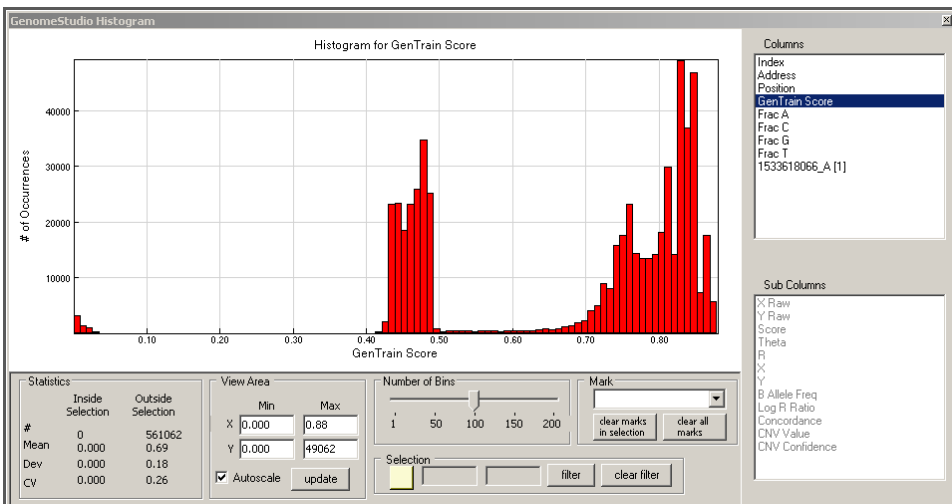


Figure 66 Histogram Displaying Data

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 GenomeStudio Histogram dialog box, 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 67).

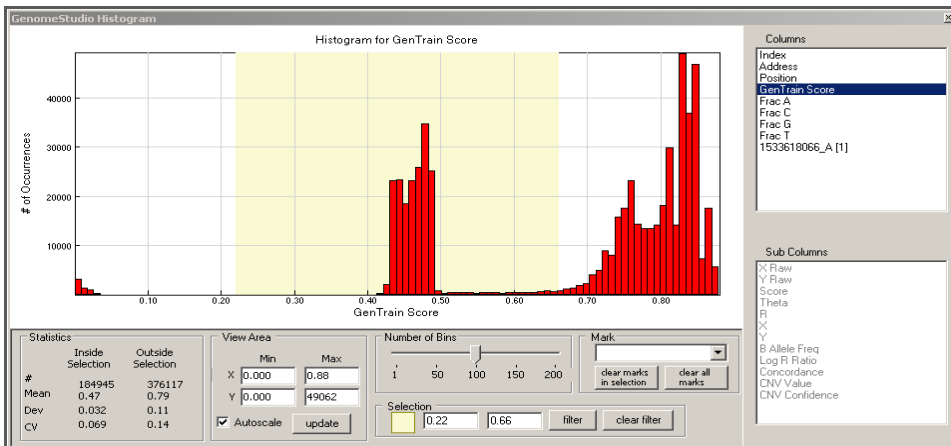


Figure 67 Histogram with Shaded Selection Area

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: <-|->.
 - 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 68).

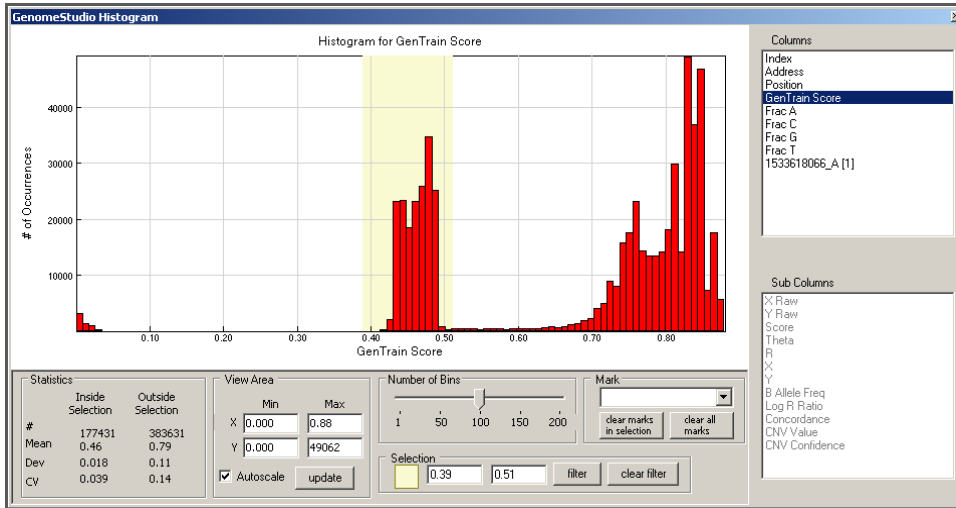


Figure 68 Resizing Shaded Selection Area

- ▶ 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 GenomeStudio Histogram dialog box. 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

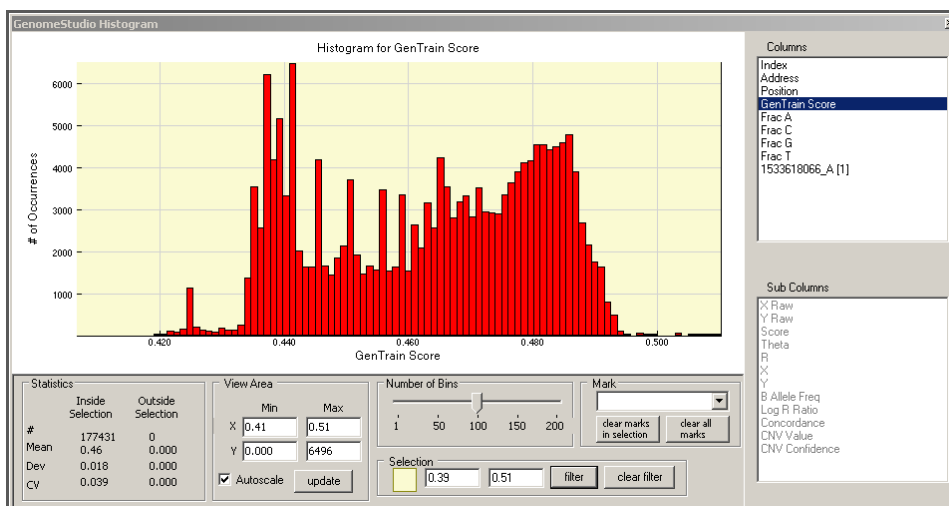
Statistic	Description
#	Number of occurrences
Mean	Mean values
Dev	Standard deviation
CV	Coefficient of variation

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 66.
2. In the Selection area of the GenomeStudio Histogram dialog box, click **Filter**.

The table data is filtered according to the shaded selection area. The histogram displays only the filtered data (Figure 69).

**Figure 69** 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 GenomeStudio Histogram, select **Configure Marks** from the dropdown list.

The Configure Marks dialog box appears (Figure 70).

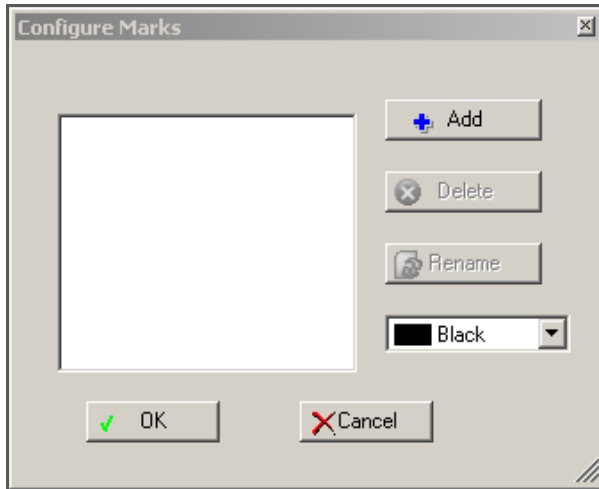


Figure 70 Configure Marks

2. Click **Add**.
3. The Select Mark Name dialog box appears (Figure 71).

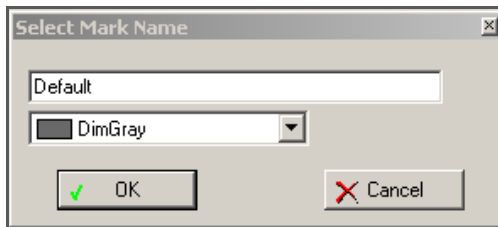


Figure 71 Select Mark Name

4. Type a name for your mark in the field.

5. Select a color for your mark from the dropdown list (Figure 72).

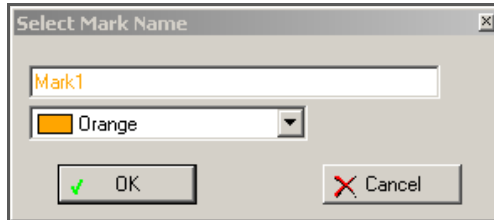


Figure 72 Choosing Mark Name & Color

6. Click **OK**.
7. In the Configure Marks dialog box, click **OK**.
The mark you created is displayed in the Mark area dropdown list.
8. Select the mark you want to apply to your data from the Mark area dropdown list.
After a few seconds, the marked data is displayed in the histogram with the properties of the mark you applied (Figure 73).

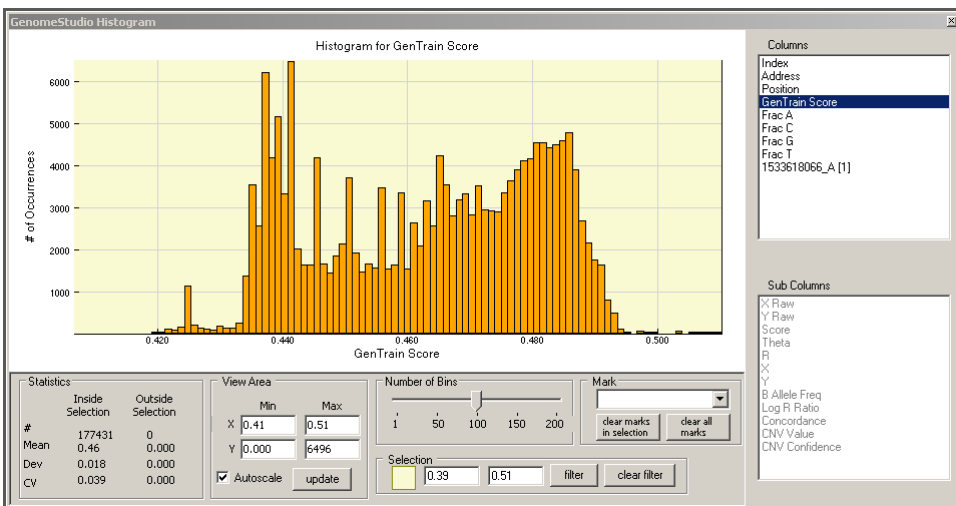


Figure 73 Marked Data Displayed in a Histogram

The data are also marked in the associated table (Figure 74).

Index	Name	Address	Chr	Position	GenTrain Score	Frac A	Frac C	Frac G	Frac T	GTtype	Score	Theta	R
182	rs10000010	9074...	4	21294943	0.4772	0.284	0.216	0.176	0.324	AB	0.2441	0.6265	0.5300
183	rs10000023	3310...	4	96091084	0.4416	0.333	0.333	0.069	0.265	AB	0.2096	0.5115	2.0802
184	rs10000030	9006...	4	103731334	0.4391	0.381	0.124	0.208	0.287	AB	0.2073	0.7168	0.7029
185	rs10000041	1042...	4	165979560	0.4243	0.225	0.206	0.402	0.167	AB	0.1945	0.7344	0.5345
187	rs10000081	5073...	4	17024632	0.4695	0.333	0.186	0.294	0.186	AB	0.2362	0.6248	0.8684
189	rs10000121	6220...	4	187931640	0.4880	0.343	0.088	0.206	0.363	AB	0.2554	0.5555	0.3524
196	rs10000169	3054...	4	77713425	0.4697	0.441	0.176	0.167	0.216	AB	0.2364	0.6062	0.9766
199	rs1000022	1042...	13	96259220	0.4790	0.235	0.206	0.275	0.284	AB	0.2459	0.5500	0.8097
201	rs1000025	5033...	6	194127012	0.4596	0.243	0.218	0.198	0.342	AB	0.2264	0.4590	1.1899
206	rs10000300	6001...	4	40307475	0.4857	0.294	0.216	0.196	0.294	AB	0.2530	0.5378	0.6029
209	rs10000388	5063...	4	185024103	0.4417	0.343	0.245	0.147	0.265	AB	0.2096	0.4960	1.7733
210	rs1000040	6066...	15	23302034	0.4675	0.265	0.265	0.314	0.157	AB	0.2342	0.6350	0.8015
214	rs10000438	2053...	4	61602528	0.4450	0.373	0.118	0.255	0.255	AB	0.2127	0.6935	0.6501
216	rs10000487	3780...	4	35121853	0.4825	0.363	0.167	0.167	0.304	AB	0.2496	0.5544	0.8641
217	rs10000490	2006...	4	129749665	0.4823	0.225	0.245	0.216	0.314	AB	0.2494	0.5503	0.4813
219	rs10000502	5068...	4	141222362	0.4915	0.297	0.228	0.163	0.312	AB	0.2593	0.6594	1.2694
222	rs10000543	5044...	4	31047057	0.4875	0.412	0.118	0.216	0.255	AB	0.2550	0.5574	0.3923
225	rs1000060	7610...	4	186214648	0.4819	0.412	0.108	0.255	0.225	AB	0.2490	0.5426	0.7100
229	rs10000708	4070...	4	42316280	0.4454	0.324	0.216	0.206	0.255	AB	0.2130	0.4893	1.2517
232	rs10000726	3610...	4	120568367	0.4866	0.314	0.127	0.176	0.382	AB	0.2540	0.5023	0.9270
235	rs10000785	7064...	4	187061194	0.4571	0.363	0.157	0.206	0.275	AB	0.2240	0.6632	0.4620
238	rs10000856	1006...	4	185721352	0.4908	0.157	0.255	0.284	0.304	AB	0.2585	0.6901	1.4922
240	rs10000901	8054...	4	52977704	0.4358	0.225	0.186	0.275	0.314	AB	0.2043	0.7223	0.5768
242	rs10000929	6007...	4	131654629	0.4406	0.245	0.186	0.245	0.324	AB	0.2087	0.4880	1.5246

Figure 74 Marked Data Displayed in Associated Table

Heat Maps

A heat map is a graphical representation of data in which the values represented by the variables in a two dimensional map are represented with different colors. Use the **Heat Map** option to plot data from any subcolumn across all columns.



NOTE

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 75).

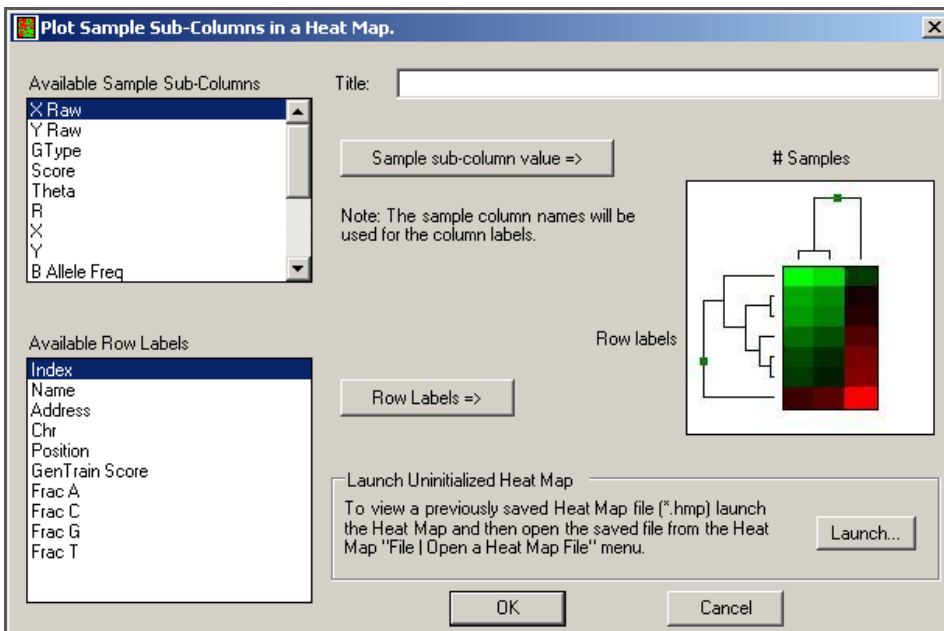


Figure 75 Plot Sample Subcolumns in a Heat Map

2. In the Title 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 76).

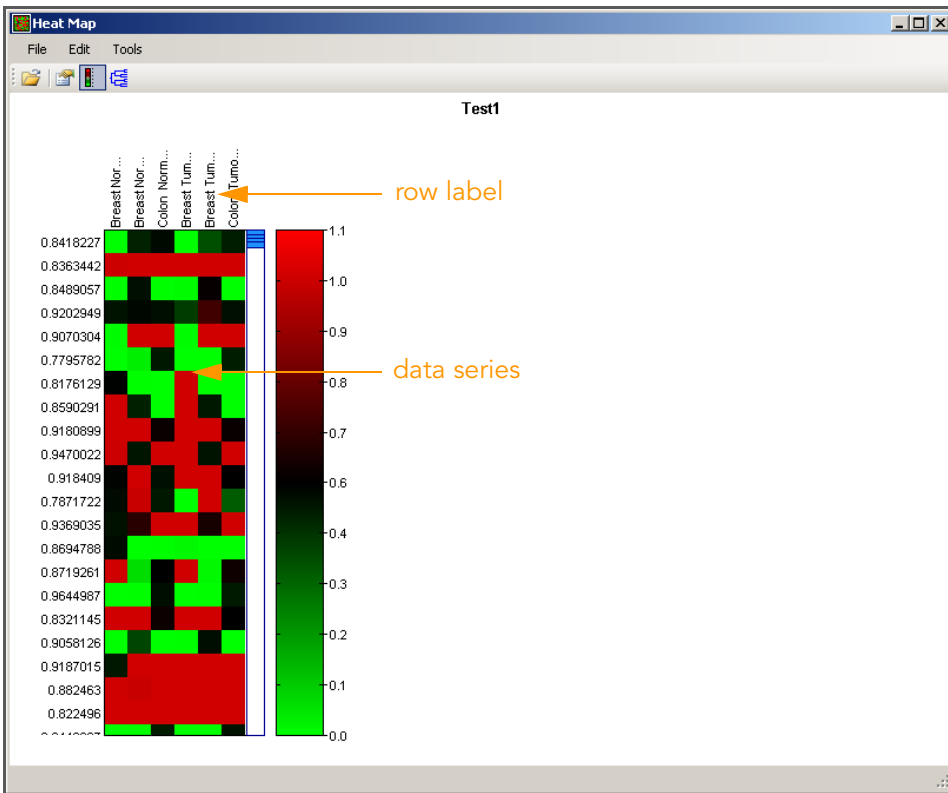



Figure 76 Heat Map with Row Labels & Data Series

Clustering Heat Map Data



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 dialog box, click  **Cluster Heat Map Rows and/or Columns.**

The Cluster Options dialog box appears (Figure 77).

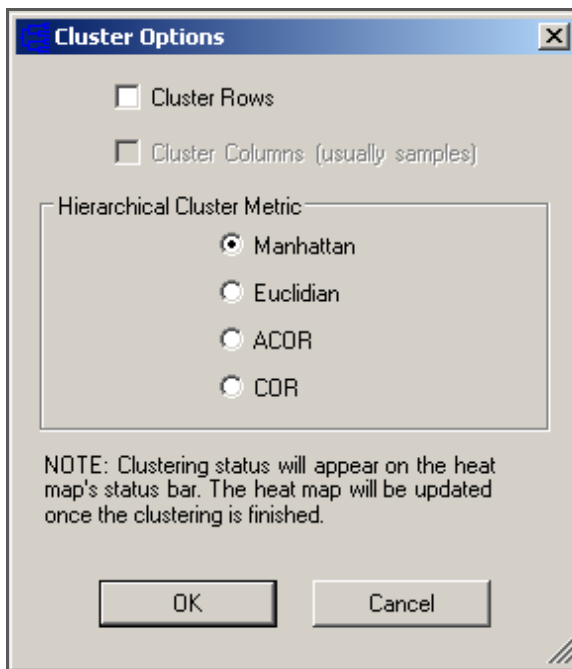


Figure 77 Cluster Options

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

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 80).

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. GenomeStudio also uses Manhattan ($\sum |X_i - Y_i|$) and squared Euclidian ($\sum (X_i - Y_i)^2$) distances.

GenomeStudio 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 78).

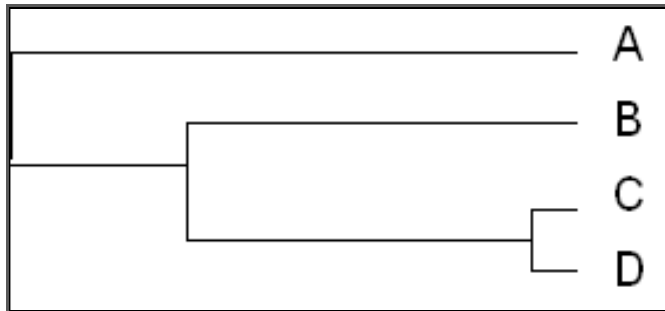


Figure 78 Dendrogram, Similarity Example

After clustering, nodes are reordered starting near the top to ensure that node "ar" is closer to "B" than node "al" is, and node "bl" is closer to "A" than node "br" is (Figure 79).

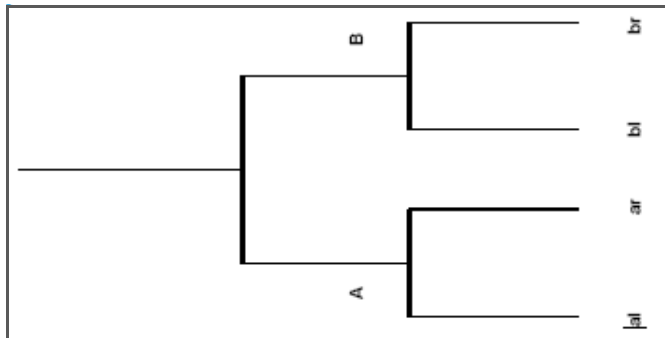


Figure 79 Dendrogram, Showing Nodes

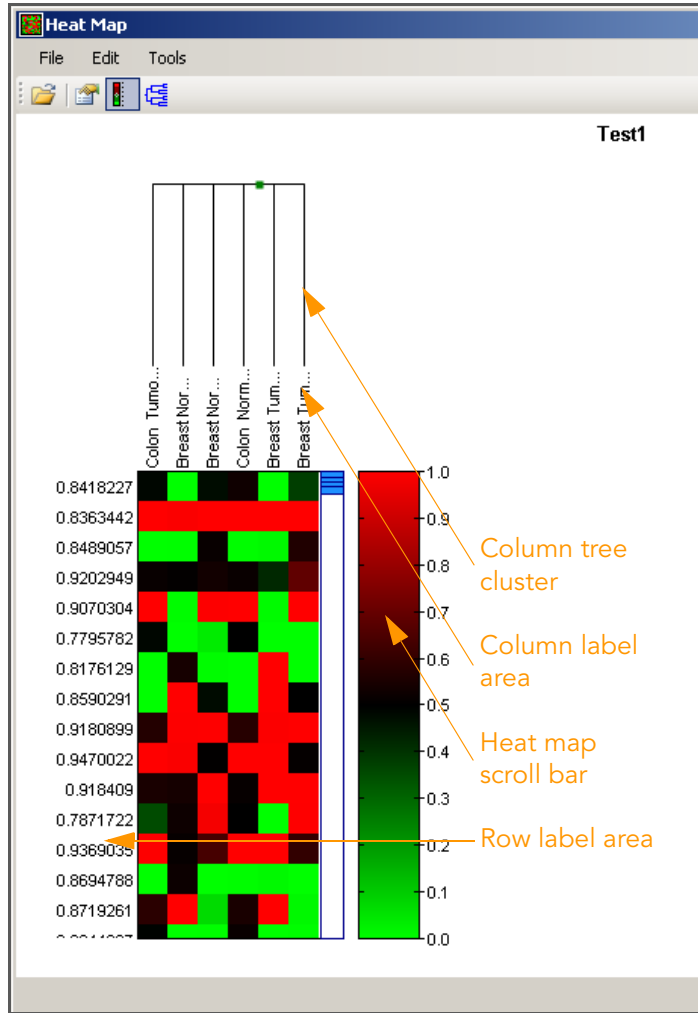


Figure 80 Heat Map, Display & Scrolling Properties

Resizing Row Labels

The row label area is shown in Figure 80.

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 80.

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 80.

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 80.

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** field.
4. Click **Save**.
Your heat map is saved in the location you specified.

Exiting the Heat Map

To exit the heat map:

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 81).

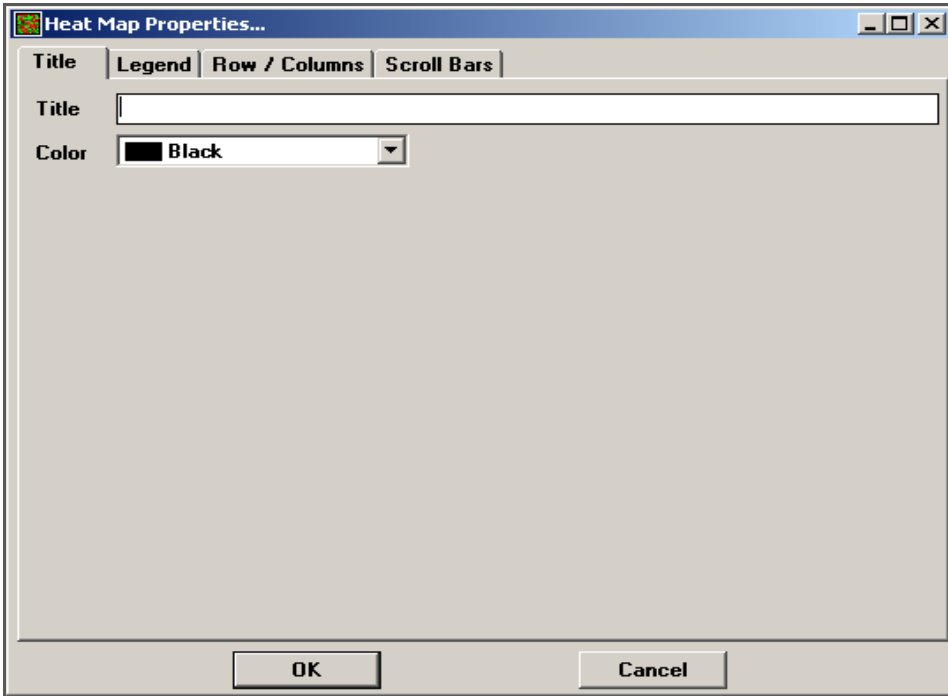


Figure 81 Heat Map Properties, Title Tab

2. In the Title tab, enter a title for your heat map in the **Title** 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 82).

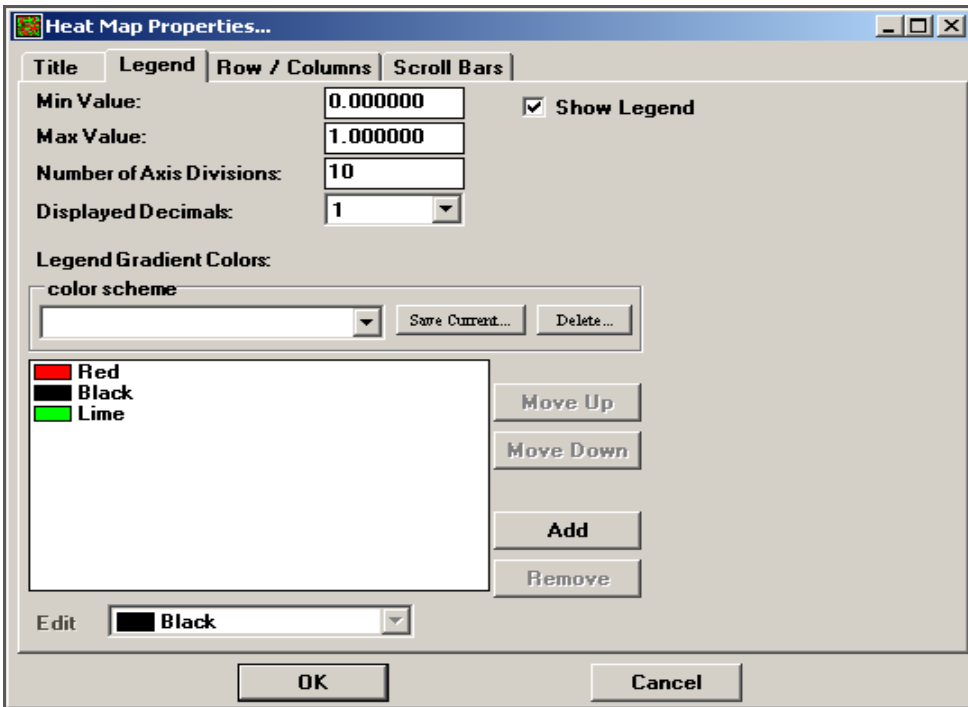


Figure 82 Heat Map Properties, Legend Tab

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 82). 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 list (Figure 82).
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**.
2. Choose a color from the dropdown list.
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 the heat map displays without the color you removed.

Row/Column Properties

You can change heat map row and column properties (Figure 83) to suit your preferences.

Showing/Hiding Labels

To show row labels:

- ▶ In the Rows area, select the **Show Labels** checkbox.

To hide row labels:

- ▶ In the Rows area, clear the **Show Labels** checkbox.

To show column labels:

- ▶ in the Columns area, select the **Show Labels** checkbox.

To hide column labels:

- ▶ in the Columns area, clear the **Show Labels** checkbox.

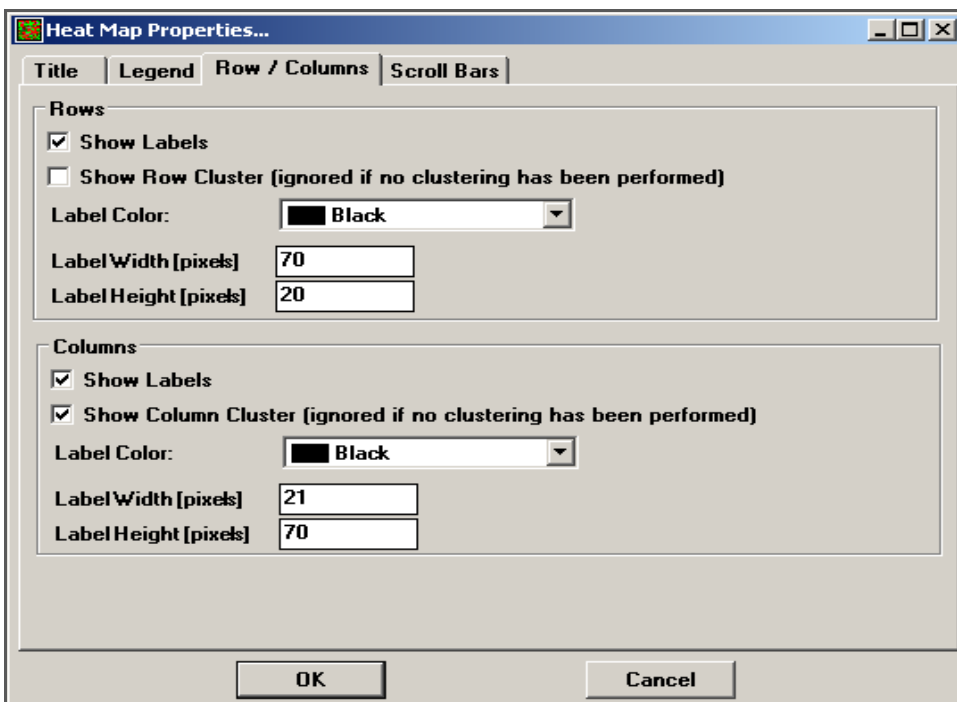


Figure 83 Heat Map Properties, Row/Columns Tab

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 list.

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

Scroll Bar Properties

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

To change scroll bar properties (Figure 84), select new colors from the **Scroll Area Color**, **Scroll Bar Color**, and **Scroll Border Color** dropdown lists.

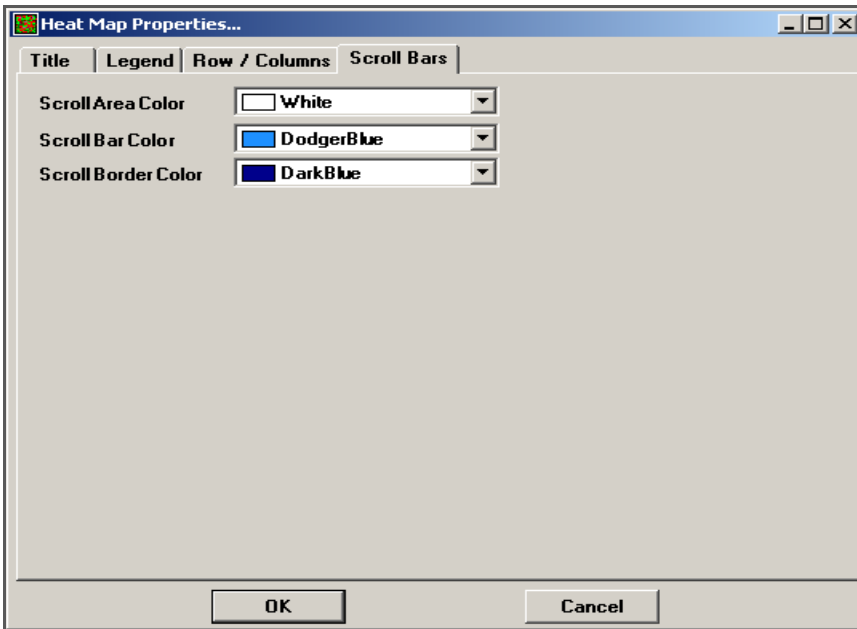


Figure 84 Heat Map Properties, Scroll Bars Tab

Creating a Presentation Image

To create a presentation image:

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

The Presentation Image Setup dialog box appears (Figure 85).

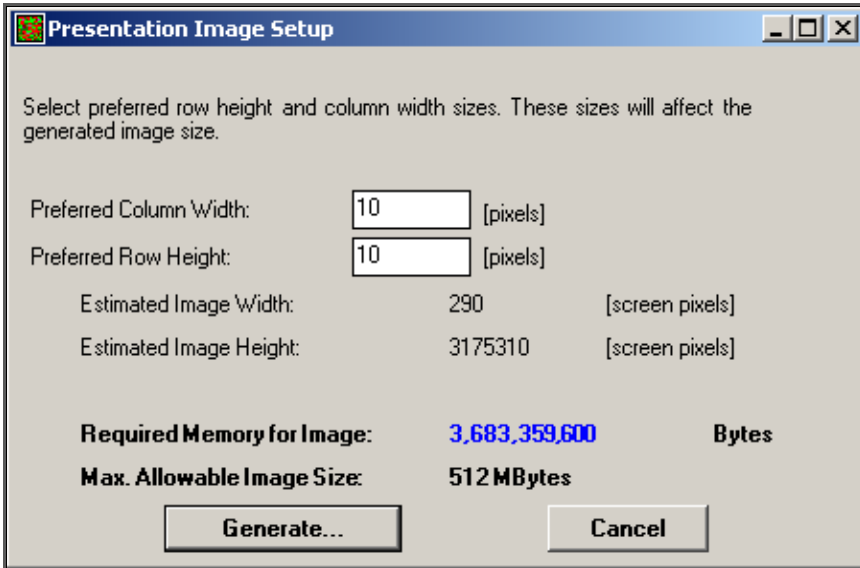


Figure 85 Presentation Image Setup

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 dialog box appears with a static heat map image that can be viewed in its entirety and saved to a file.

Illumina Genome Viewer

Topics

- 90 Introduction
- 90 Using the IGV
 - 91 Selecting Data Plots
 - 95 Saving Data Plots
 - 96 Changing Viewing Modes
 - 100 Using the IGV Toolbar
 - 102 Using the IGV Menus
 - 103 Plotting Data in the IGV
 - 111 Viewing Data in the IGV
 - 113 Getting Data Files

Introduction

This chapter describes how to use the IGV (Illumina Genome Viewer).

Using the IGV

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

If you are using a GenomeStudio sequencing module (ChIP Seq, DNA Seq, RNA Seq), your project opens to the IGV by default.

If you are using a GenomeStudio microarray module (GT, GX, M, PT), launch the IGV by doing the following:

- ▶ In the GenomeStudio main window, select **Tools | Show Genome Viewer** (Figure 86).

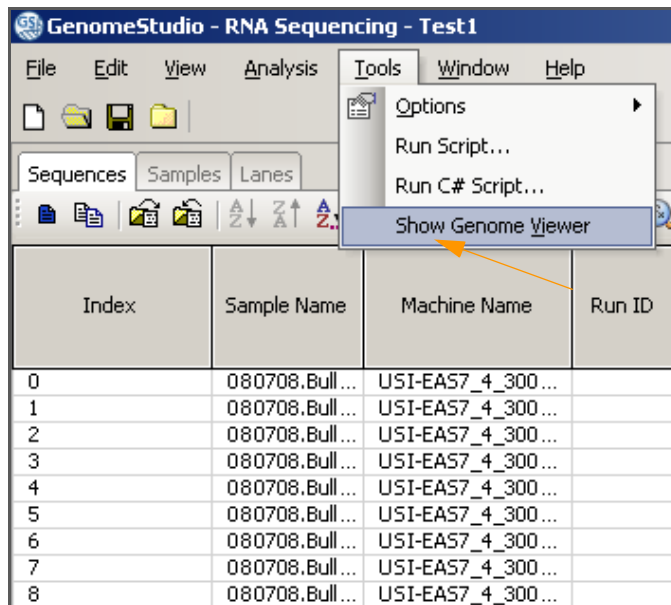


Figure 86 Launching the IGV

The IGV main window appears (Figure 87).

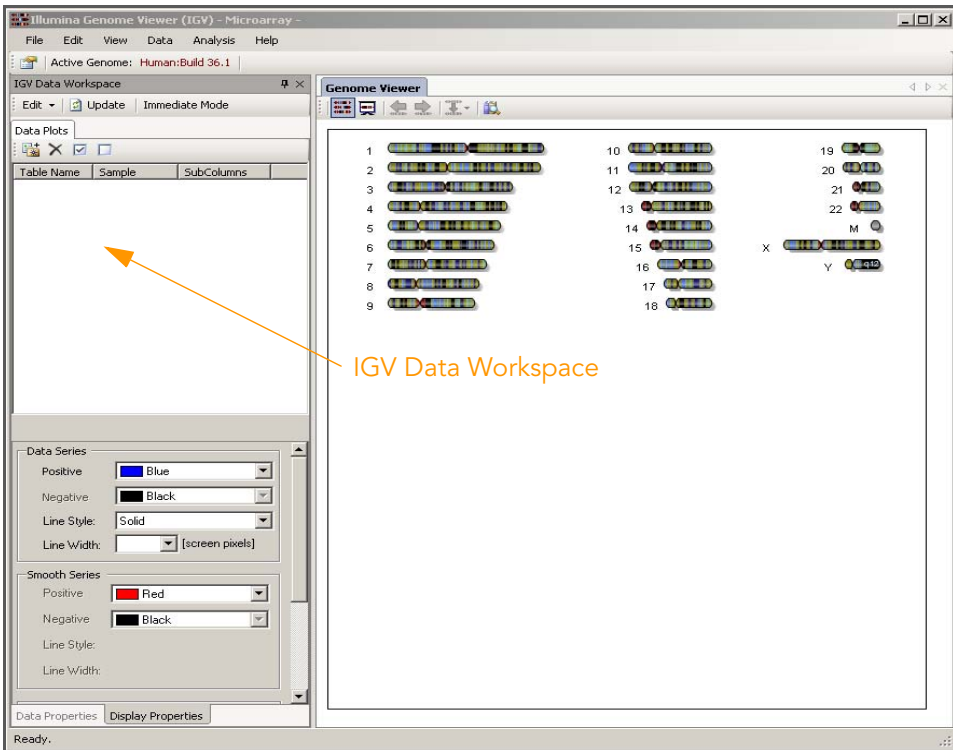


Figure 87 IGV Main Window, Whole Genome View Mode

Selecting Data Plots

When you first create a project and launch the IGV, the IGV Data Workspace in the left panel is empty. However, you can load and store your “favorite” data plots into the IGV Data Workspace for quick and easy viewing.

Perform the following steps to add data plots to the IGV Data Workspace, which you can then view in the IGV:

1. In the IGV Data Workspace area, position the cursor over the large white area under the Data Plots tab and right-click. A context menu appears (Figure 88).

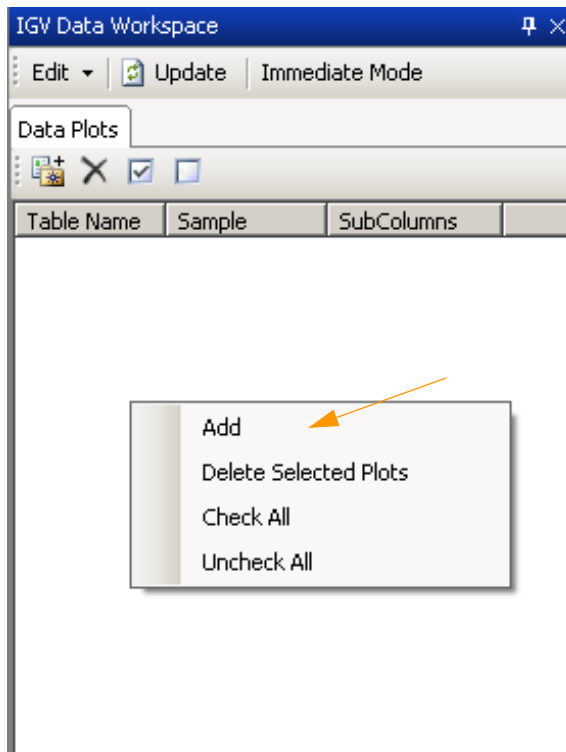


Figure 88 IGV Data Workspace Context Menu

2. Select **Add** from the context menu.
The Add Favorite Data Plots dialog box appears (Figure 89).

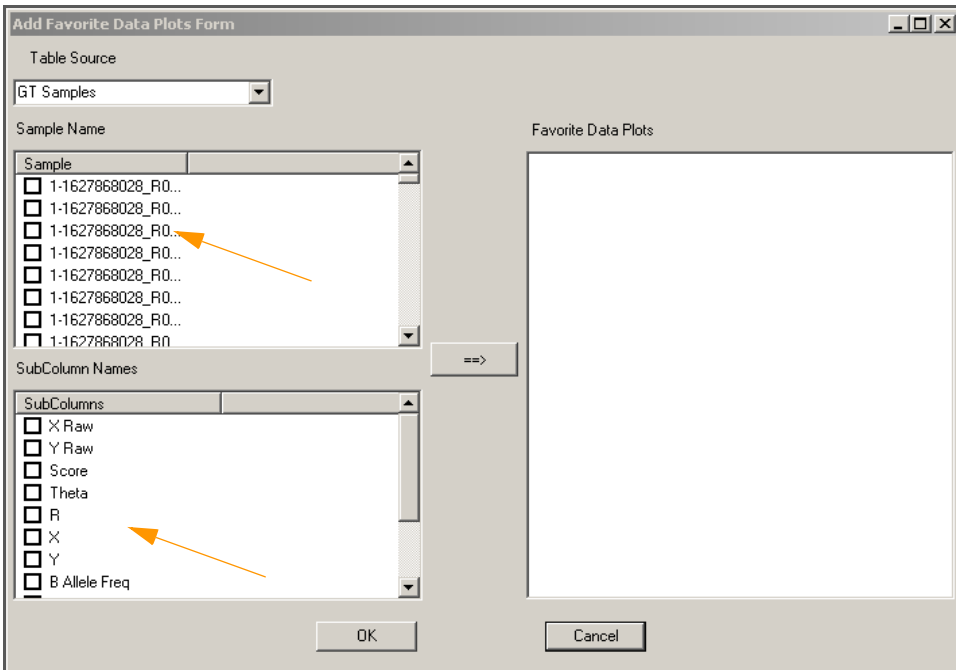


Figure 89 Add Favorite Data Plots Dialog Box

3. In the Table Source area, select the table that contains the data you want to display in the IGV.
4. In the Samples Name area, select the samples you want to display in the IGV.
5. In the Subcolumn Names area, select the subcolumns you want to display in the IGV.
6. Click ==> to activate your selections.

Your selections are moved to the Favorite Data Plots area (Figure 90).

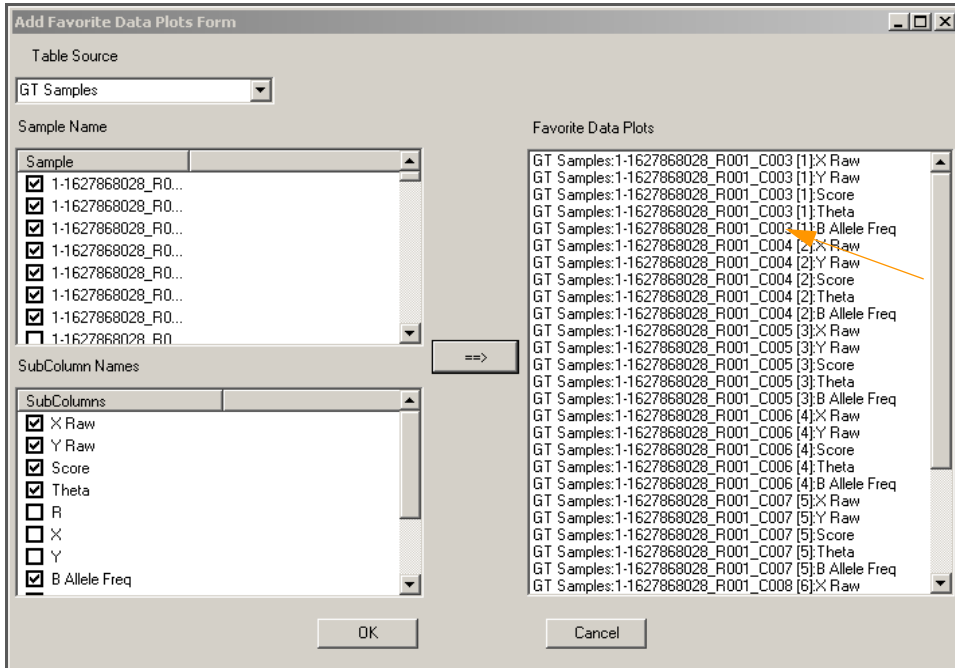


Figure 90 Favorite Data Plots Selected

7. Click **OK**.

The data you selected appears in the data plots area in the IGV Data Workspace (Figure 91).

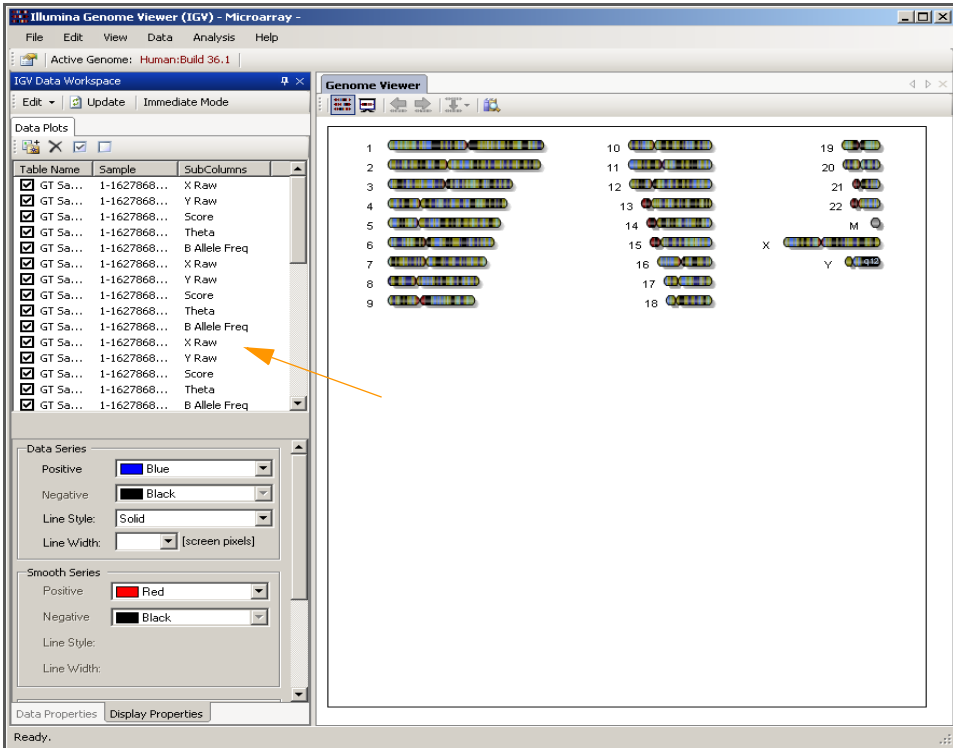


Figure 91 Favorite Data Plots in the IGV Data Workspace



CAUTION

If you close the current project without saving the data plots in the IGV Data Workspace, the next time you open the project the area will be blank and you will need to select the data plots again.

Saving Data Plots

You can save the plots listed in the IGV Data Workspace for quick access to them the next time you open this project.

To save selected data plots in the IGV Data Workspace, perform the following steps:

- ▶ In the IGV Data Workspace, click **Edit | Plot Settings | Save**.

The listed data plots are saved in the IGV Data Workspace of this project. These plots will be available from the IGV Data Workspace the next time you open this project.

Changing Viewing Modes

In Illumina sequencing modules, the IGV has a per-chromosome view. However, in Illumina microarray modules, the IGV has two viewing modes:

- ▶ **Whole Genome View Mode**
- ▶ **Chromosome Slide Show Mode**

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

In Chromosome Slide Show mode, the IGV can display up to four plots at a time for one chromosome. In Whole Genome View mode, the IGV can display up to two plots at a time for all chromosomes.



NOTE

To reduce loading time, every project with more than 500,000 SNPs automatically displays in Chromosome Slide Show mode.

To display a plot in Whole Genome View mode, perform the following steps:

1. In the Data Plots area, select the checkbox for the data table(s) you want to view.
2. Click **Update**.
3. **[Optional]** Select the **Immediate Mode** checkbox if you want the plots you select to be automatically updated in the IGV.



NOTE

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 92).

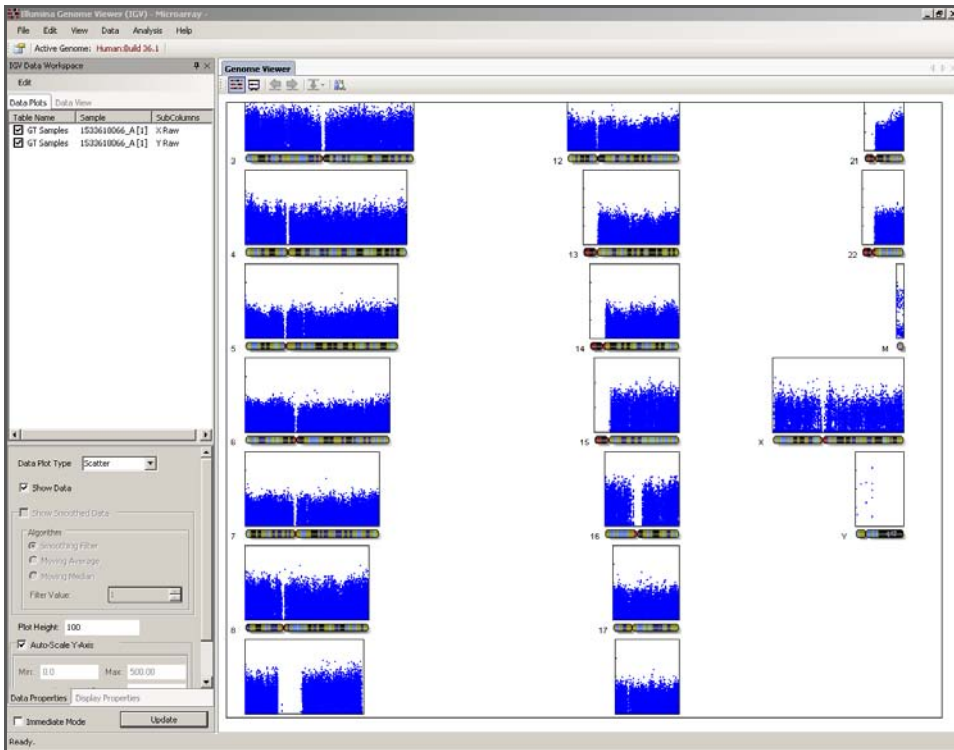


Figure 92 Data Plot, Whole Genome View Mode




NOTE

If SNPs are selected (highlighted or marked) in the Full Data Table, they appear in the IGV as different-colored squares.

In Chromosome Slide Show 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 Slide Show Mode:

1. In the Data Plots area, select the checkboxes for the plots you want to display.
2. In the IGV, click  **Chromosome Slide Show Mode**.

The Chromosome Slide Show View Options dialog box appears (Figure 93).

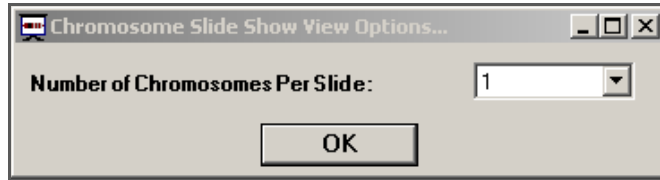


Figure 93 Chromosome Slide Show View Options

3. In the dropdown list, select the number of chromosomes per slide you want to display in the IGV.
4. Click **OK**.
The plots appear in the IGV main window (Figure 94).

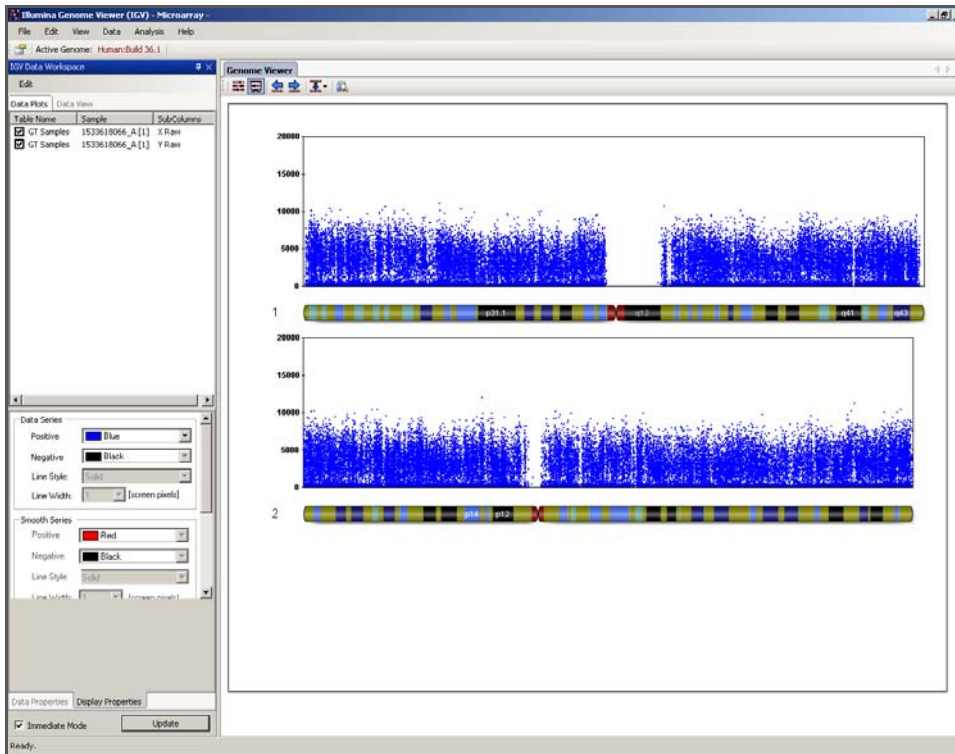


Figure 94 Data Plot, Chromosome Slide Show Mode

To change the display options of your plots, perform the following steps:

1. In the upper half of the Data Plots area, choose the samples for which you want to display data.
2. In the lower half of the Data Plots area, in the Data Properties tab, select an option for data plot type:
 - Scatter
 - Line
 - Bar
3. If the Show Smoothed Data option is available, select an algorithm and a filter value.
4. Enter a value in the Plot Height area.
5. Select **Auto-Scale Y-Axis** and enter autoscaling values in the following fields:
 - Min
 - Max
 - Number of Steps
 - Number of Decimals to Display
6. Click the Display Properties tab and select the display options you prefer.
7. Select the **Immediate Mode** checkbox if you want the plots you have chosen to be automatically updated in the IGV. The plot(s) you selected appear in the IGV with the parameters you have chosen (Figure 95).

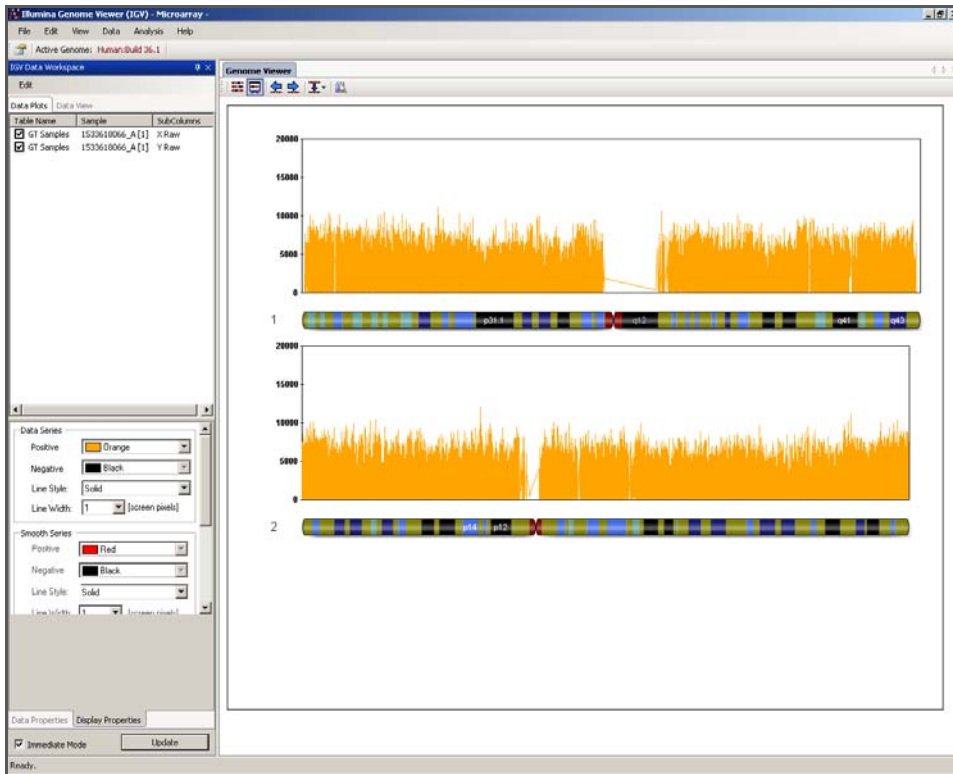


Figure 95 Data Plot, Display Options Selected

Using the IGV Toolbar







Figure 96 shows the IGV toolbar.



Figure 96 IGV Toolbar

Table 4 includes descriptions of IGV toolbar buttons and their functions.

Table 4 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/Hide Bookmarks	Toggles between showing and hiding bookmark files you can select to display with the current plot(s).

Using the IGV Menus

Table 5 describes IGV menu elements and their functions.

Table 5 IGV Menu Options & Descriptions

Option	Description
File Menu	
Exit	Closes the IGV main 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 on top of the IGV main window.
Chromosome Heat Map	Displays a chromosome heat map for the current chromosome.
Sequence Viewer	Launches the ISV on top of the IGV main window.
Genome Viewer	Toggles the IGV on and off.
IGV Data Workspace	Toggles the IGV Data Workspace on and off.
CNV Analysis as Bookmarks	Converts the project's CNV analysis results to bookmarks.
ChIP Seq Region as Bookmarks	Converts the project's ChIP Seq regions to bookmarks.
Bookmark Viewer	Displays bookmark files for the current chromosome.
Data Menu	
Add/Remove Data Tracks	Allows you to add or remove data tracks for use with the ICB.

Table 5 IGV Menu Options & Descriptions

Option	Description
Analysis Menu	
Run Auto-Bookmark	Opens the Auto-Bookmark Analysis dialog box, from which you can select Auto-Bookmark Analysis algorithms to apply, and save your selections with comments as a new analysis.
Help Menu	
About	Displays version, copyright, and reference information for the IGV.

Plotting Data in the IGV

The IGV has access to the sample columns in the Full Data Table. If any paired samples have been created, it also has access to the sample columns in the Paired Sample Table.

The samples appear in the IGV Data Workspace to the left of the IGV main window. Figure 97 shows the IGV Data Workspace as it appears for microarray modules.

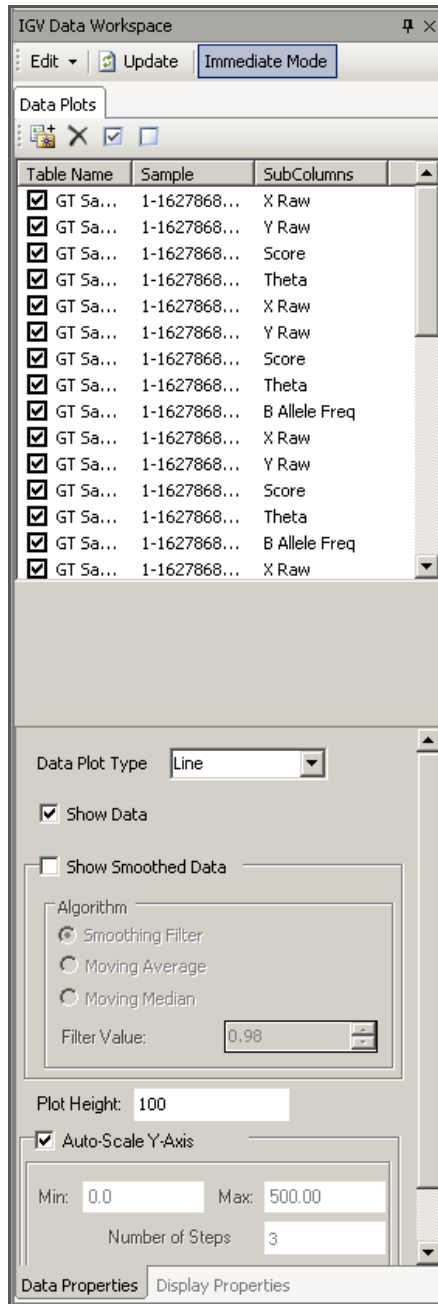


Figure 97 IGV Data Workspace, Microarray Modules

Figure 94 shows the IGV Data Workspace as it appears for sequencing modules. Note the Stacked Alignment Plots tab.

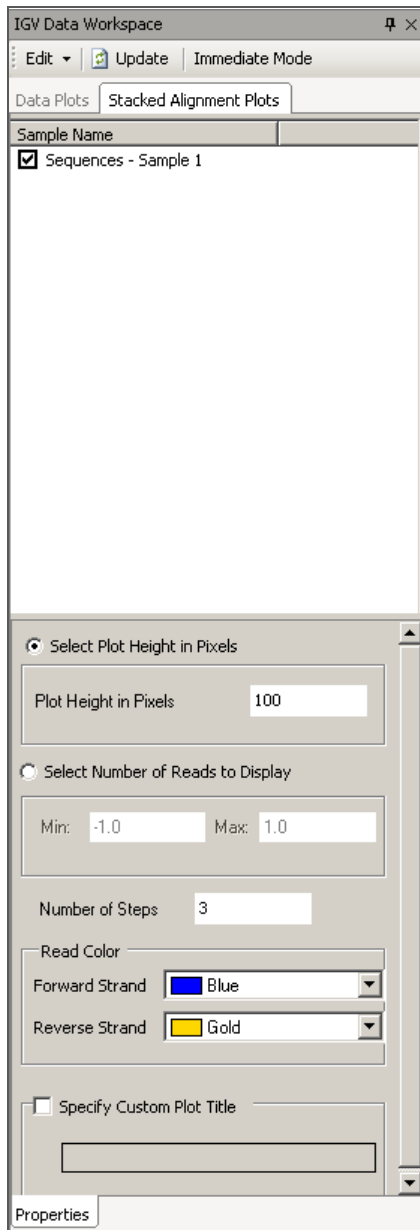


Figure 98 IGV Data Workspace, Sequencing Modules

Selecting Options in the Data Plots Tab

In the IGV Data Workspace, the Data Plots tab displays by default.

The checkboxes in the upper half of the Data Plots tab toggle a particular plot's visibility. Leaving a checkbox cleared hides the associated plot.

For example, in Figure 99, both data plots are selected, and the subcolumns to be plotted are X Raw and Y Raw.

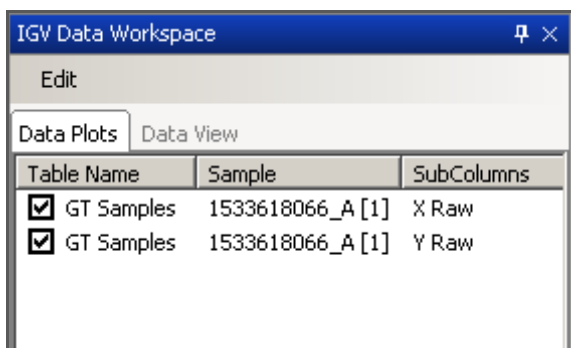


Figure 99 IGV Data Workspace, Data Plots Tab

Selecting Options in the Stacked Alignment Plots Tab

In the Stacked Alignment Plots tab, you can select plots to display. In Figure 100, only the first sequence file is selected.

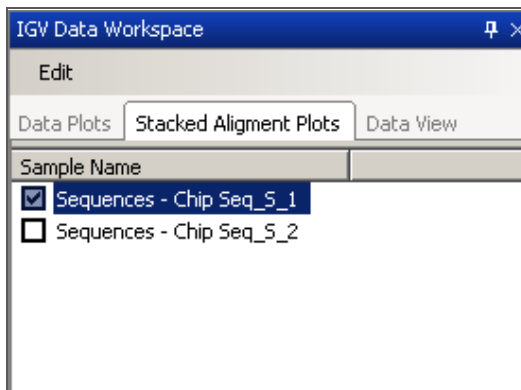


Figure 100 IGV Data Workspace, Stacked Alignment Plots Tab

From the area below (Figure 101), you can also change the appearance of the stacked alignments plots. Select options for the following variables:

- ▶ **Select Plot Height in Pixels**
 - **Plot Height in Pixels**—Vertical height of the stacked alignment plots
 - **Number of Steps**—Number of steps or “ticks” to display along the Y-axis
- ▶ **Select Number of Reads to Display**
 - **Min**—Minimum number of reads to display along the Y-axis
 - **Max**—Maximum number of reads to display along the Y-axis
- ▶ **Read Color**
 - **Forward Strand**—Display color of the forward strand
 - **Reverse Strand**—Display color of the reverse strand.
- ▶ **Specify Custom Plot Title**
Enter a title in the field.

The screenshot shows a dialog box with the following settings:

- Select Plot Height in Pixels**
 - Plot Height in Pixels: 100
- Select Number of Reads to Display**
 - Min: -1.0, Max: 1.0
 - Number of Steps: 3
- Read Color**
 - Forward Strand: Blue
 - Reverse Strand: Gold
- Specify Custom Plot Title**
 - Plot1

Figure 101 IGV Data Workspace, Stacked Alignments Plots, Display Options

Selecting Options in the Data View Tab

In the Data View tab, you can select loaded data tracks, bookmark analyses, or genome annotations to display (Figure 102).

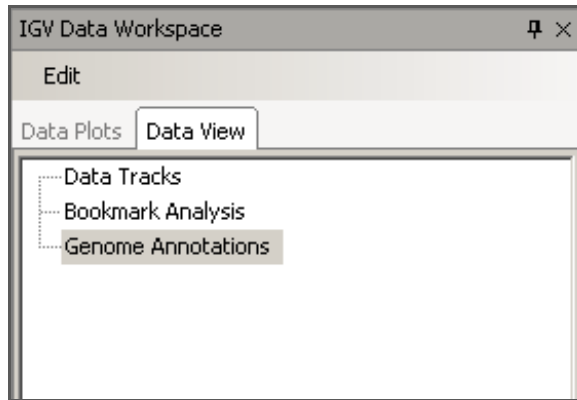


Figure 102 IGV Data Workspace, Data View Tab

Selecting Options in the Data Properties Tab

In the Data Properties tab, you can choose which samples to plot and which display options to use (Figure 103).

The screenshot shows the 'Data Properties' tab in the IGV interface. The 'Data Plot Type' is set to 'Scatter'. The 'Show Data' checkbox is checked. The 'Show Smoothed Data' checkbox is unchecked. Under 'Show Smoothed Data', the 'Algorithm' section has three radio buttons: 'Smoothing Filter' (selected), 'Moving Average', and 'Moving Median'. The 'Filter Value' is set to 1. The 'Plot Height' is set to 200. The 'Auto-Scale Y-Axis' checkbox is checked. The 'Min' value is 0 and the 'Max' value is 20000. The 'Number of Steps' is set to 3. The 'Number of Decimals to Display' is set to 0. The 'Data Properties' tab is selected, and the 'Display Properties' tab is also visible. The 'Immediate Mode' checkbox is checked, and the 'Update' button is present.

Figure 103 Data Properties Tab

Selecting Options in the Display Properties Tab

In the Display Properties tab (Figure 104), you can change the appearance of each plot. Select options for the following variables:

- ▶ **Data Series**
 - **Positive**—Select a color for positive values.
 - **Negative**—Select a color for negative values.
 - **Line Style**—Select a line style.
 - **Line Width**—Select a line width in screen pixels.
- ▶ **Smooth Series**
 - **Positive**—Select a color for positive values.
 - **Negative**—Select a color for negative values.
 - **Line Style**—Select a line style.
 - **Line Width**—Select a line width in screen pixels.
- ▶ **Specify Custom Plot Title**—A default title populates automatically in the field; however, you can change the plot title to whatever you like.
- ▶ **Specify Y-Axis Custom Label**—Optionally, enter a Y-Axis custom label.
- ▶ **Font Color**—Select a font color.
- ▶ **Show Grid**—Select whether you would like to display a grid for your plots.

To apply changes to your data plots, select **Immediate Mode** and click **Update Plots**.

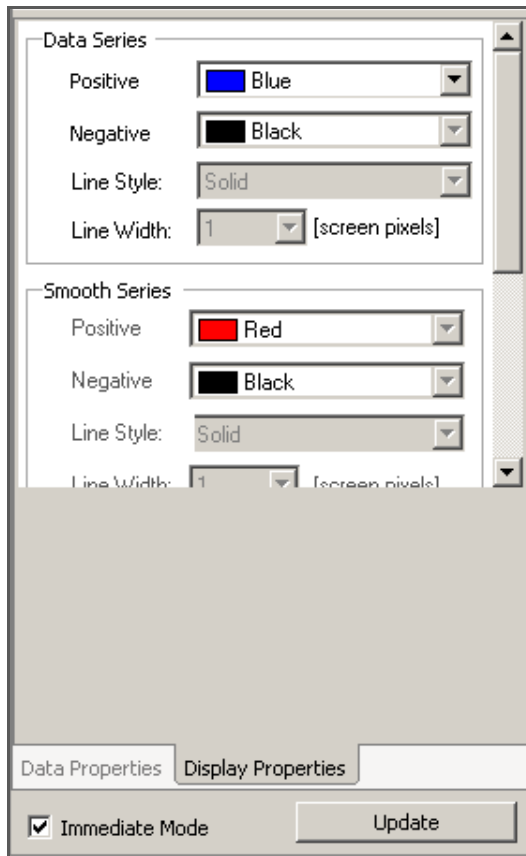


Figure 104 Display Properties Tab

Viewing Data in the IGV

The Data View tab has a navigation tree with branches from which you can view available genomes, view loaded data tracks, and view or change the active bookmark analysis (Figure 105).

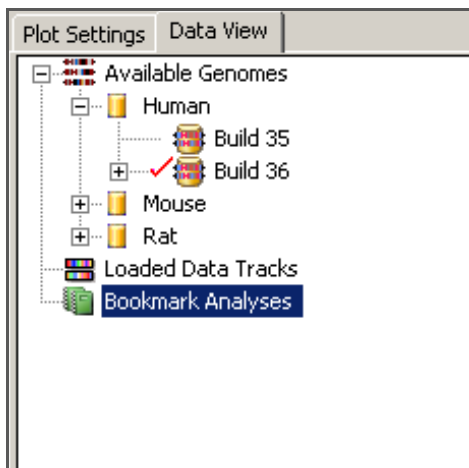


Figure 105 Data View Tab

The Available Genomes branch allows you to drill down through available species and genomes. For more information about viewing data files in the IGV, see “Getting Data Files” on page 113.

The Loaded Data Tracks branch allows you to drill down through loaded data tracks. For more information about loading data tracks, see “Loading Illumina-Provided Data Tracks” on page 134, or see “Creating and Loading Your Own Data Tracks” on page 137.

The Bookmark Analyses branch allows you to drill down through available bookmark analyses. For more information about bookmark analyses, see “Using the Bookmark Viewer” on page 151.



NOTE

When you open the IGV, no bookmark analysis is active by default. To activate a bookmark analysis, double-click a bookmark analysis entry in the “Bookmark Analyses” branch of the Data View tree.

Getting Data Files

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

If you want to load builds of genomes 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> (Figure 106).

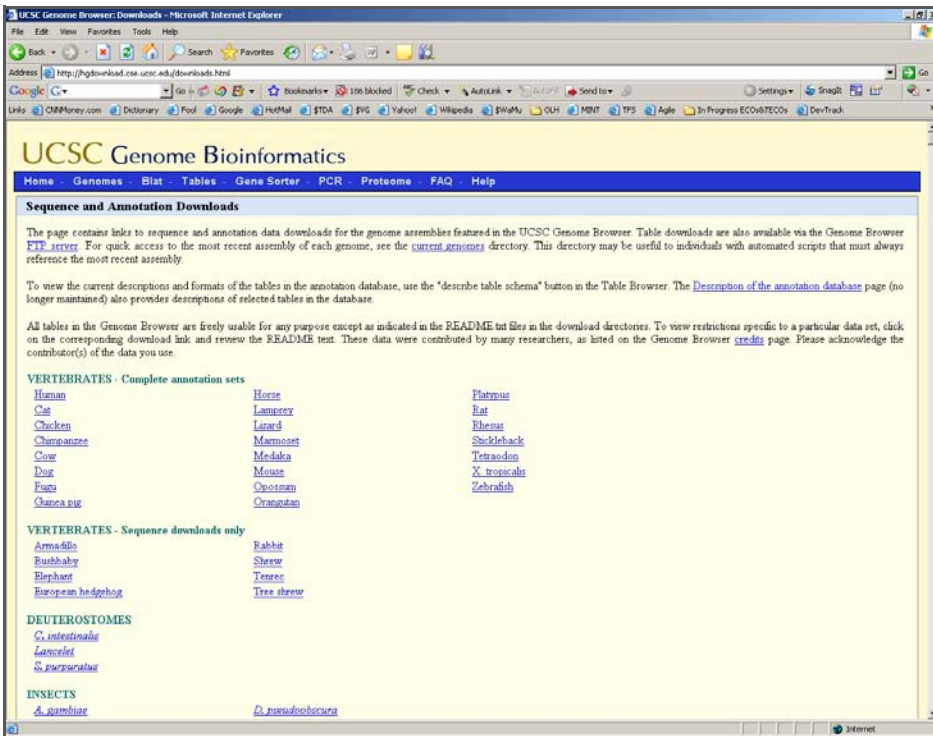


Figure 106 UCSC Genome Bioinformatics Web Page

2. Click the species name of the genome you want to download; e.g., **Human**.

The web page for the genome you selected appears (Figure 107).

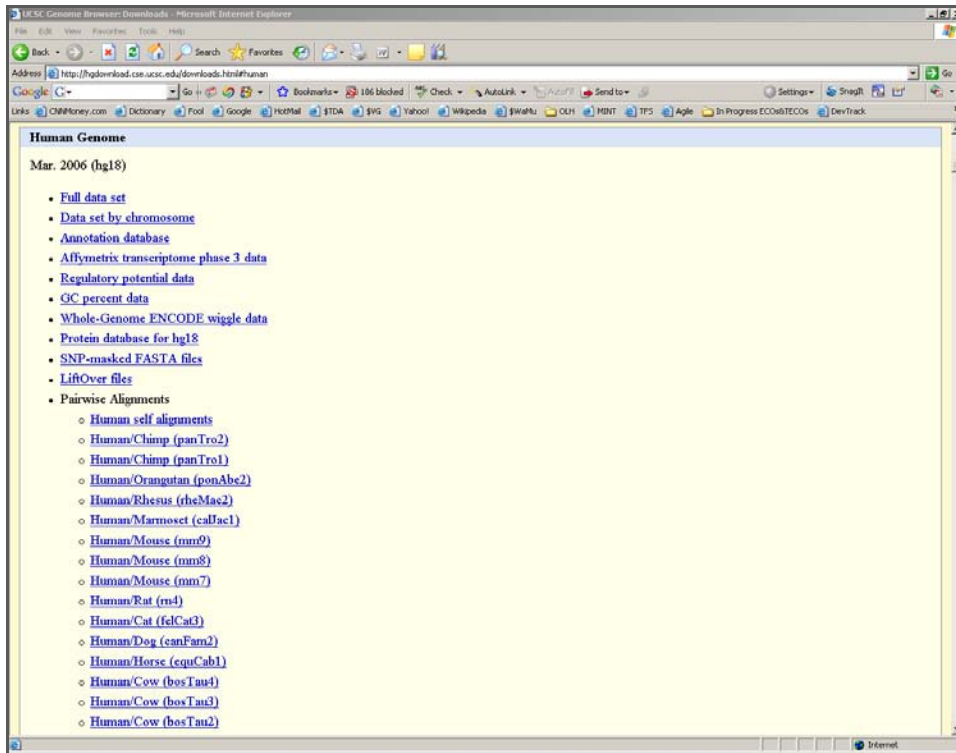


Figure 107 Selected Genome Web Page (Human)

3. Click **Annotation database**.

The annotation database for your selected species appears (Figure 108).

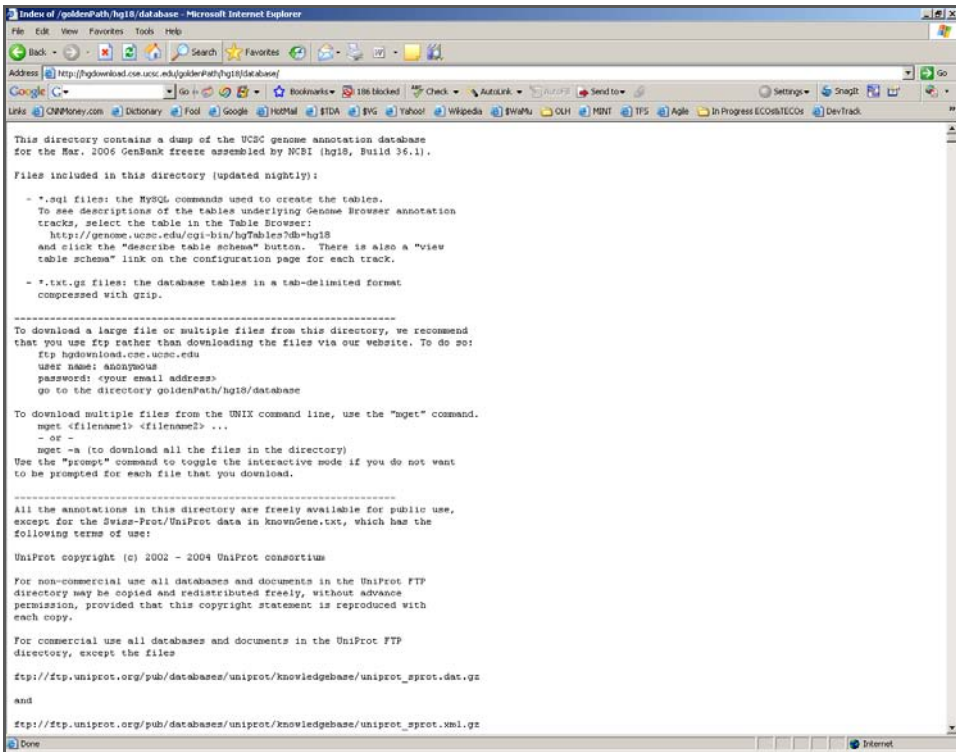


Figure 108 Annotation Database Web Page (Human)

4. On the Annotation Database web page, scroll down until you find files available to download (Figure 109).

8. Click **Data set by chromosome**.

The **Index** (Data set by chromosome) web page for the genome you selected appears (Figure 110).

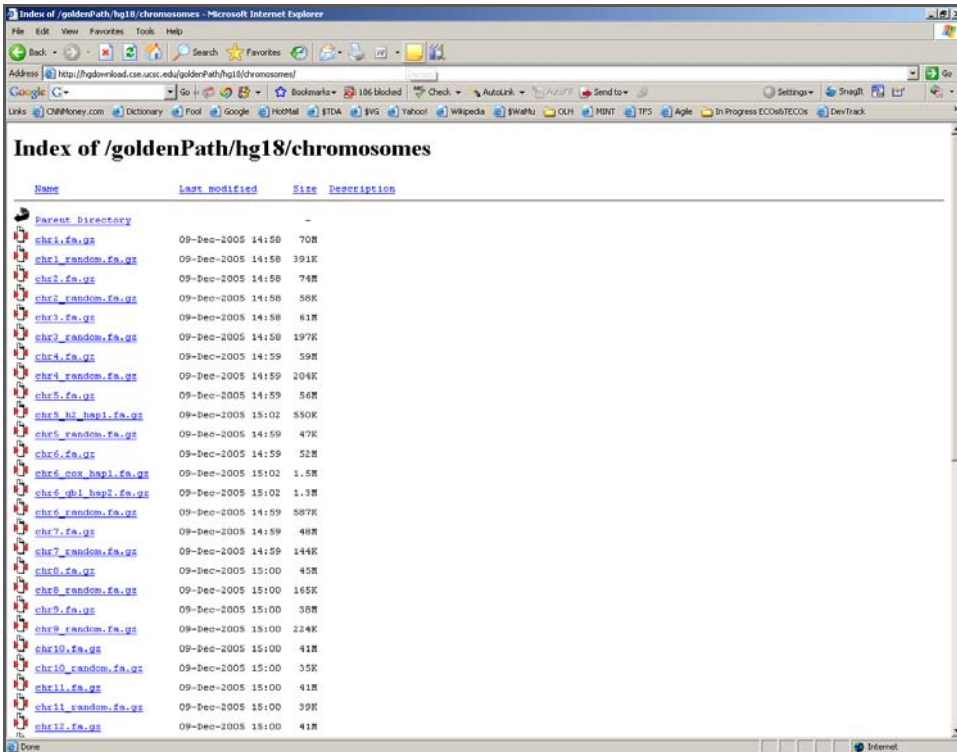


Figure 110 Index (Data set by chromosome) Web Page

9. Download each of the sequence files shown on the Index web page into the folder you created on your desktop in Step 5.
10. Navigate to `C:\Program Files\Illumina\GenomeStudio\Modules\Genome Viewer\Genomes\`, and create a `Species\Build` folder for the genome you selected.
For example, create a folder called `Human\Build 36.1`.
The complete path to this folder is `C:\Program Files\Illumina\GenomeStudio\Modules\Genome Viewer\Genomes\Human\Build 36.1`.

11. Unzip the annotation files from Step 5 into the folder you created in Step 10.
12. Create a *Sequence* subfolder inside the folder you created in Step 10.
Example: `C:\Program Files\Illumina\GenomeStudio\Modules\Genome Viewer\Genomes\Human\Build 36.1\Sequence`
13. Unzip the sequence files from Step 9 into the *Sequence* subfolder you created in Step 12.

The selected genome is now available to be viewed in the IGV.

To view the selected genome in the IGV, perform the following steps:

1. In the IGV, click **Edit Preferences**.
2. The **Preferences** dialog box appears (Figure 111).

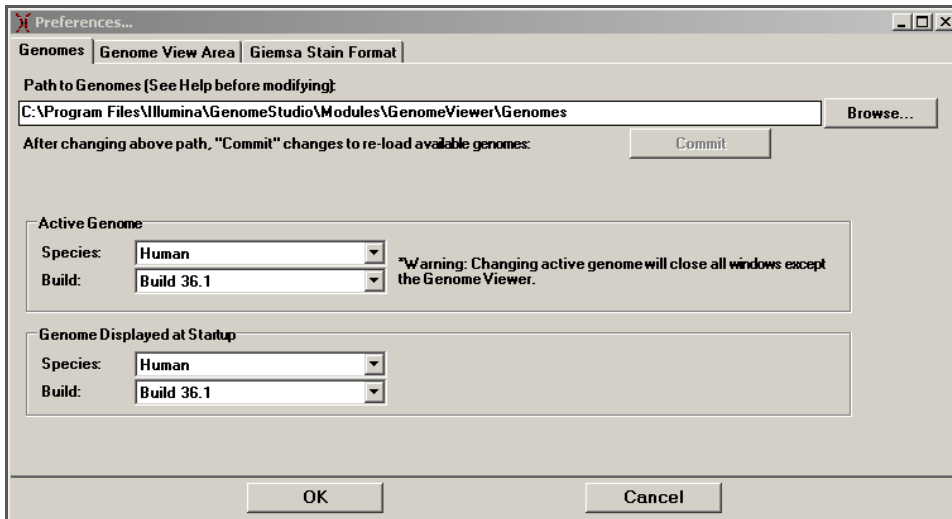


Figure 111 Preferences

3. In the Active Genome area, change the species and build to the genome you want to display in the IGV (Figure 112).

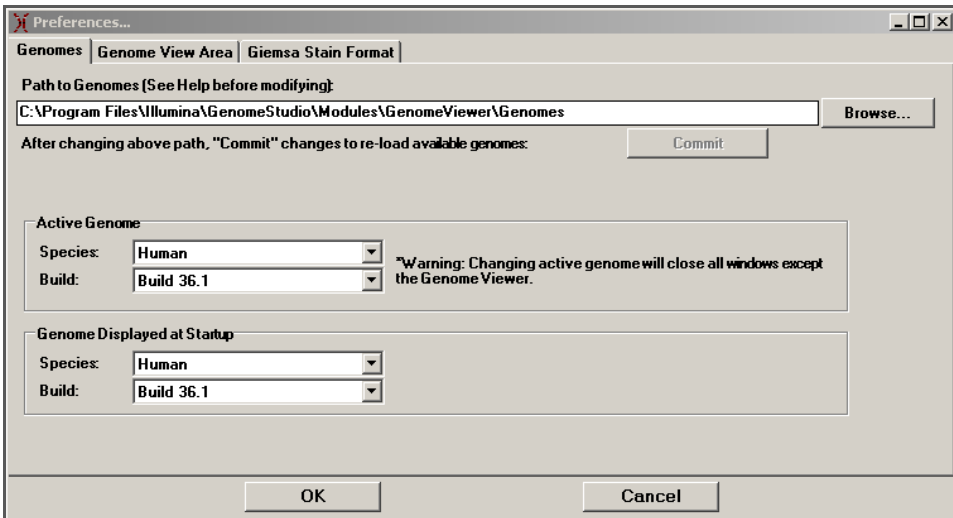


Figure 112 Changing the Displayed Genome Species & Build

4. **[Optional]** If you like, at this point you can change the species and build that displays in the IGV on startup.
5. Click **OK**.
The **Continue Changing Displayed Genome?** dialog box appears (Figure 113).

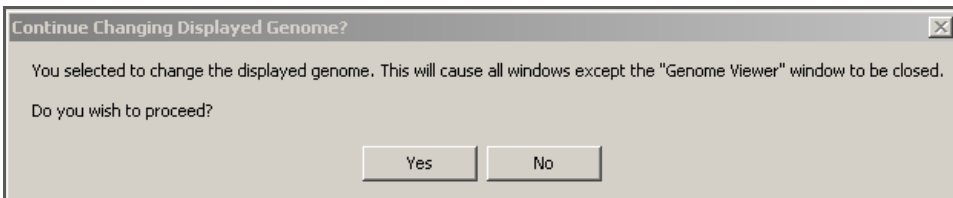


Figure 113 Continue Changing Displayed Genome

6. Click **Yes**.
The genome you selected is now shown in the IGV (Figure 114).

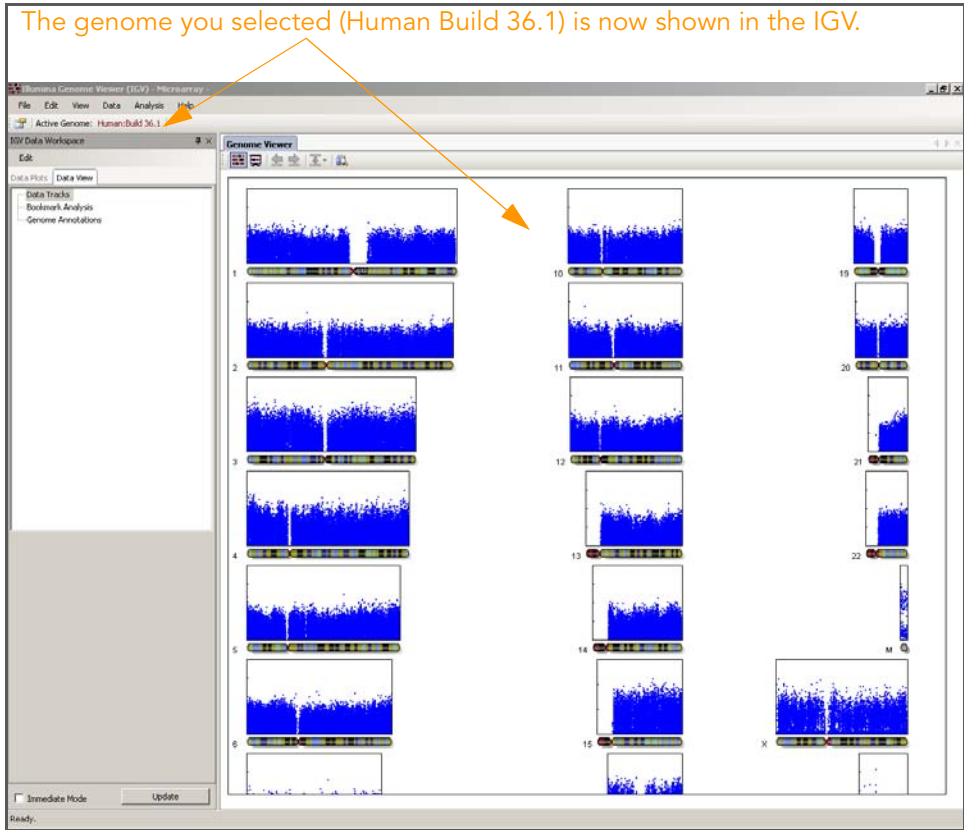


Figure 114 Selected Genome Shown in the IGV

Illumina Chromosome Browser and Other Visualization Tools

Topics

- 123 Introduction
- 123 Using the ICB
 - 124 Using the ICB Toolbar
 - 127 Using the ICB Context Menu
 - 127 Changing the View Region
 - 128 Viewing Gene Information
 - 131 Viewing Project Manifest SNPs
 - 133 Finding Genes or Probes
 - 134 Loading Illumina-Provided Data Tracks
 - 137 Creating and Loading Your Own Data Tracks
- 138 Using the Illumina Sequence Viewer
- 140 Using the Chromosome Heat Map
 - 141 Clustering Data
 - 143 Binning Data

144 Smoothing Data

Introduction

This chapter describes how to use the following GenomeStudio data visualization tools:

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

Using the ICB

The ICB allows you to explore data by chromosome or by gene. The following sections describe how to work with the ICB.

To launch the ICB:

- ▶ Double-click a chromosome in the IGV.
The ICB appears (Figure 115).

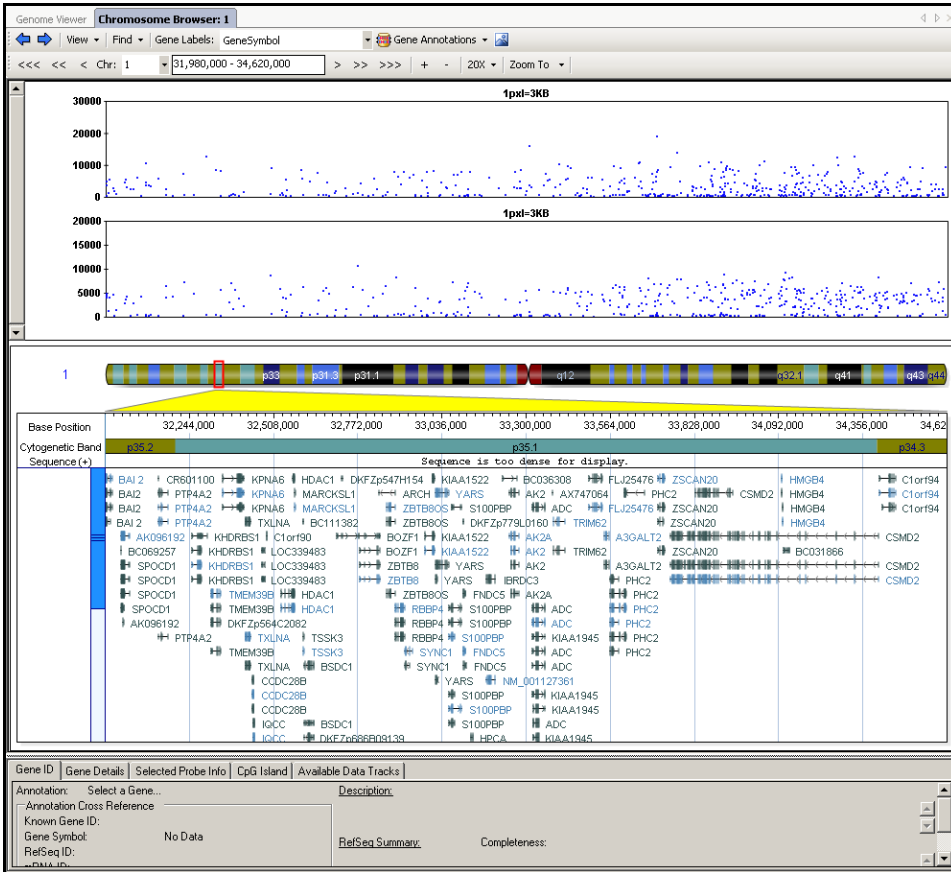


Figure 115 ICB

Using the ICB Toolbar

Figure 116 shows the ICB toolbar.

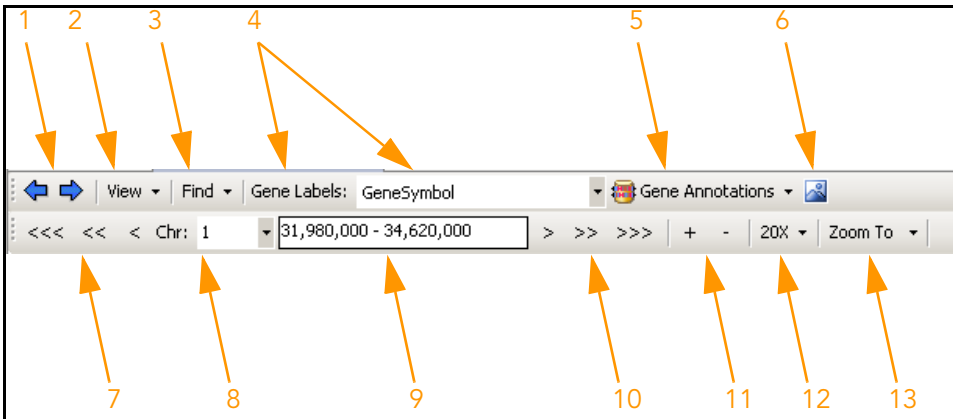


Figure 116 ICB Toolbar

Table 6 includes descriptions of ICB toolbar elements and their functions.

Table 6 ICB Toolbar Elements

Number	Element	Description
1	Navigate Back/Forward	Behaves like Back/Forward in an internet browser; takes you back to and forward from previous views.
2	View	Allows you to select from among many views, including: <ul style="list-style-type: none"> • Dense Gene View • Zoom Plot to Current View • CpG Islands • Manifest Probes SNPs • Consensus Sequence • Bookmark Viewer
3	Find	Allows you to find genes or probes .

Table 6 ICB Toolbar Elements (continued)

Number	Element	Description
4	Gene Labels	<p>Toggles how the gene IDs are shown in the gene region. Genes can be viewed using the following identifiers:</p> <ul style="list-style-type: none"> • KnownGeneID • mRNAID • Swiss-Prot Accession • Swiss-Prot Display ID • Gene Symbol • RefSeq ID • NCBI Protein Accession • None
5	Gene Annotations	<p>Choose one of the following:</p> <ul style="list-style-type: none"> • Edit—displays the Gene Annotation dialog box, 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. • MiRBASE—displays MiRBASE information in the ICB in red text.
6	Show Bookmarks	Toggles bookmark display on and off
7	Navigate Back	Navigates backward by 10%, 50%, or 100% of the current view.
8	Chr	Shows the current chromosome displayed for the current genome. You can choose a different chromosome to view from the dropdown list.
9	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.
10	Navigate Forward	Navigates forward by 10%, 50%, or 100% of the current view.

Table 6 ICB Toolbar Elements (continued)

Number	Element	Description
11	Zoom in/out	Allows you to zoom in or out from the current chromosome view.
12	Magnification	Allows you to set the magnification of the zoom tool.
13	Zoom to Base Level/Whole Chromosome	Allows you to zoom directly to the base level or whole chromosome level.

Using the ICB Context Menu

In addition to toolbar options, the ICB has a context menu which allows you to copy chromosome plots to the clipboard for use in documents.

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:

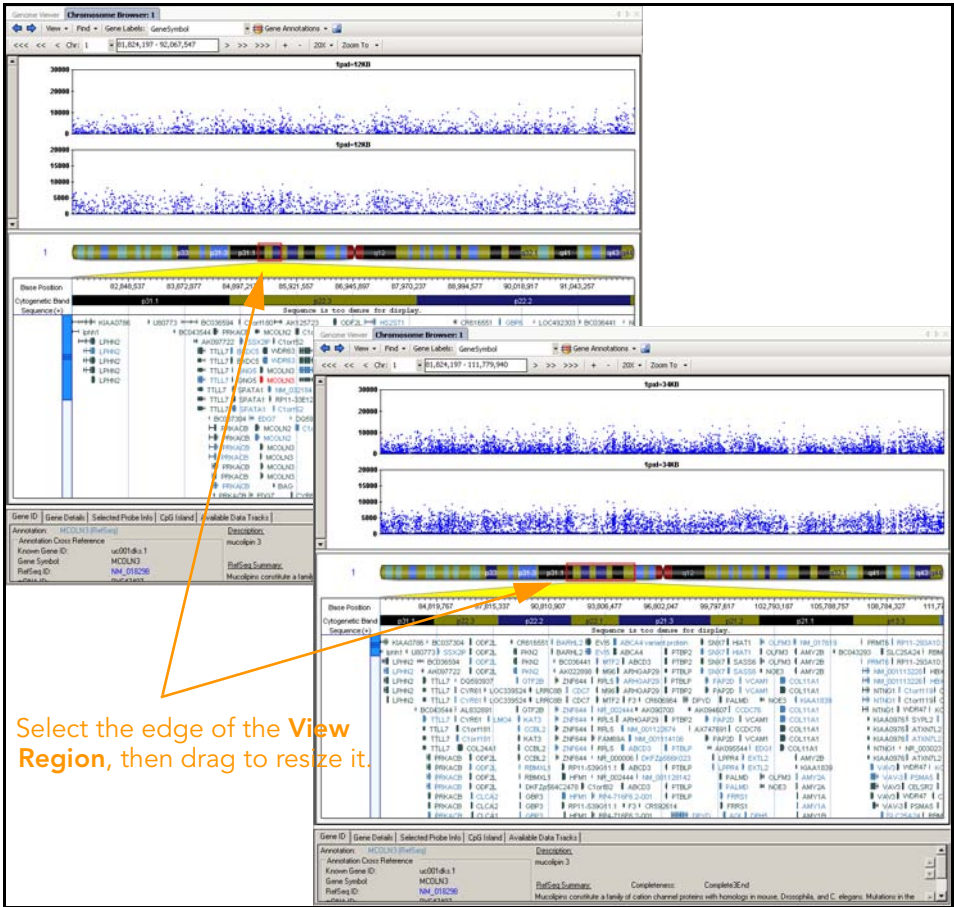
1. Position the cursor inside the **Base Position** axis row.
2. Click and hold the left mouse button and drag the **Base Position** axis, or scroll using the mouse wheel.

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 (Figure 117).



Select the edge of the **View Region**, then drag to resize it.

Figure 117 Resizing the View Region

Viewing Gene Information

The gene you select appears in red in the ICB (Figure 118). 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.

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 118 shows the view zoomed in to an exon 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.

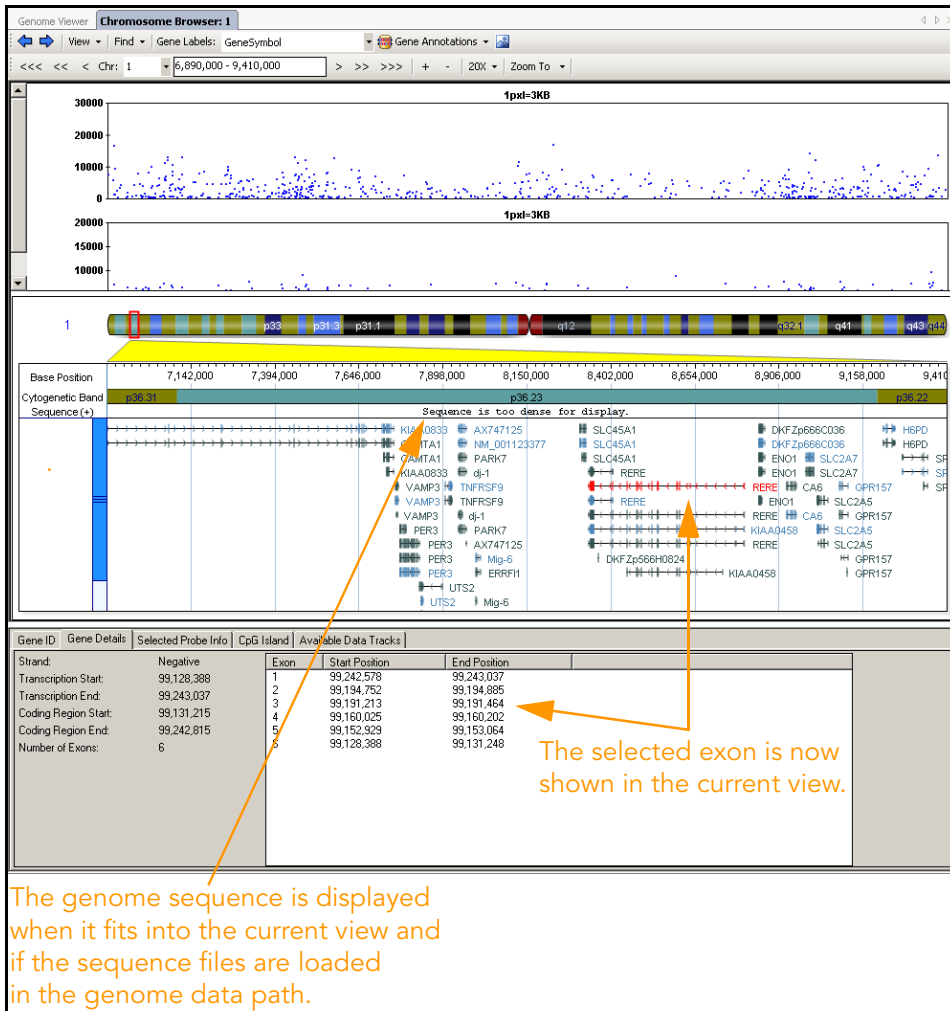


Figure 118 Viewing Gene Exons in Detail

Viewing Project Manifest SNPs

To display SNPs in the current project manifest:

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

The SNPs in the current manifest are displayed (Figure 119).



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

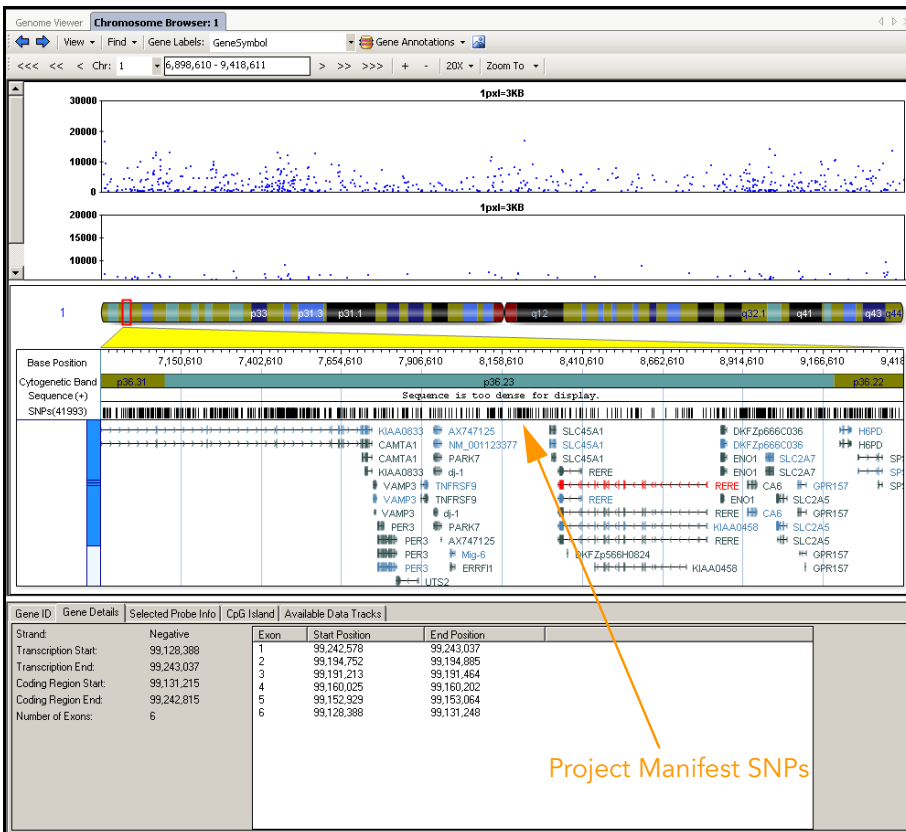


Figure 119 Loading Project Manifest SNPs into the ICB

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

- ▶ To activate the SNP Info tab, click a SNP. The SNP Info tab 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. A vertical blue line overlays the SNP, showing any genes with which the SNP intersects (Figure 120).

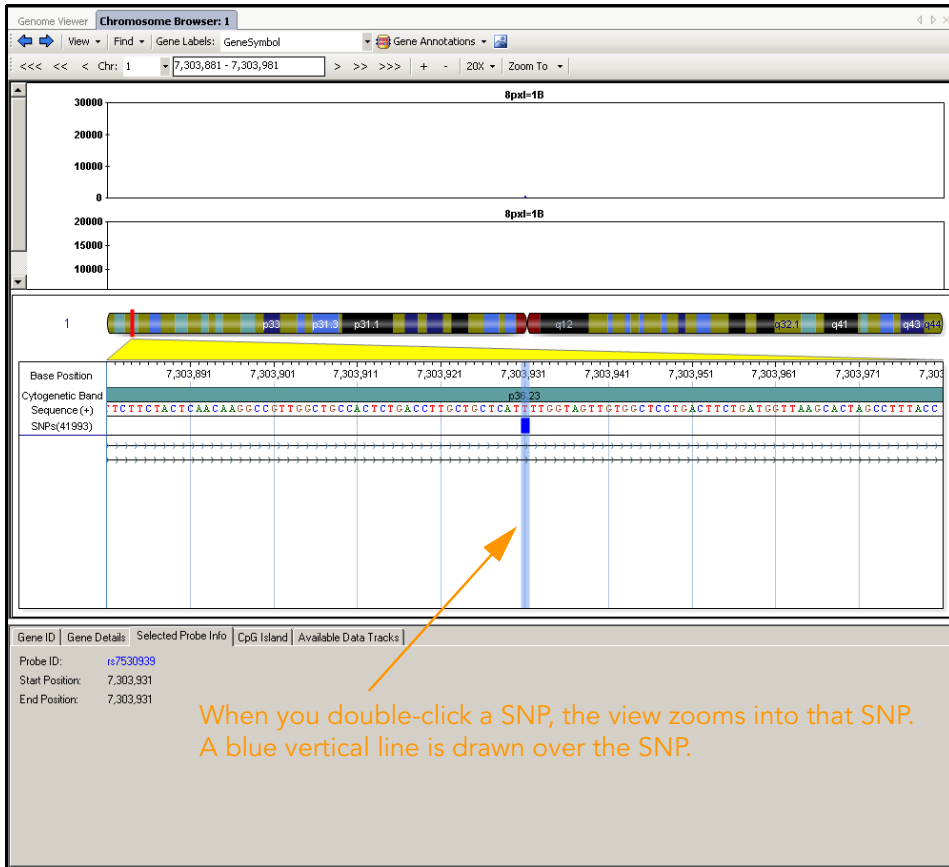


Figure 120 Zoomed View (Double-Clicking a SNP)

Finding Genes or Probes

The ICB provides a powerful search interface for finding and exporting interesting genes and probes.

Genes can be found by using the following types of information:

- ▶ Specified base-pair range
- ▶ Gene name
 - Load from a file with a list of genes
 - Advanced search (exact match, partial match, wild cards, regular expression)
- ▶ Gene identifiers
 - Gene Symbol
 - RefSeq ID
 - Known Gene ID
 - mRNA ID
 - SWISS-PROT ID
 - NCBI Protein Accession

Probes can be found by using the following types of information:

- ▶ Chromosome number
- ▶ Name
- ▶ Specified base-pair range
- ▶ Location within a gene
- ▶ Location within exons
- ▶ Location relative to gene transcription start positions



NOTE

You must load manifest probes before using the Find Probes dialog box.

To load manifest probes:

- ▶ Go to **View | Manifest Probes | SNPs**.

The SNPs appear in the IGV and they are now available for searching.

Gene or probe search results can be copied to the clipboard to use with third-party applications.

The base pair range currently selected in the IGV is used as the default base pair range in the Find Genes or Find Probes dialogs.

Loading Illumina- Provided 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 GenomeStudio.

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

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

The Add/Remove Data Tracks dialog box appears (Figure 121).

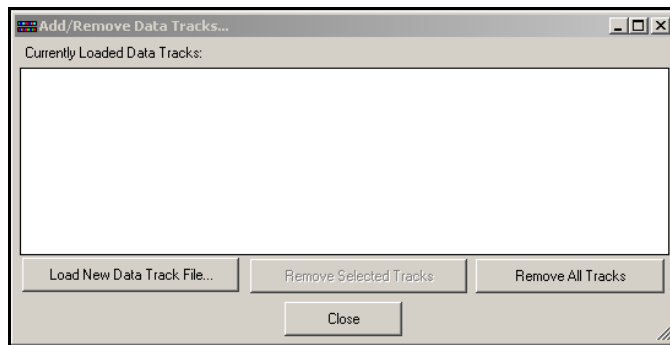


Figure 121 Add/Remove Data Tracks

3. Click **Load New Data Track File**.
4. Browse to the location of the standard linkage disequilibrium data tracks that were automatically installed with GenomeStudio:

```
c:\Program Files\Illumina\GenomeStudio\Modules\GenomeViewer\DataTracks\Smith_LD
```

5. To select one or more of the files that are available to load, do one of the following:
 - a. Double-click one or more of the available files:
 - 10kb.txt
 - 30kb.txt
 - 100kb.txt
 - 500kb.txt
 - 1000kb.txt
 - b. Select one or more of the available files and click **Open**.
- The available data tracks appear in the Add/Remove Data Tracks dialog box (Figure 122).

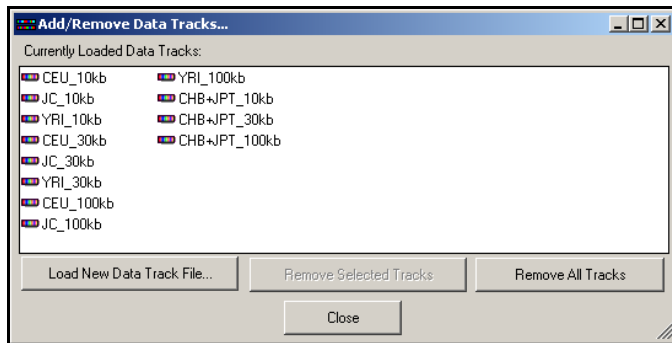


Figure 122 Selected Data Tracks

The loaded data tracks are also listed in the IGV, and graphically displayed in the ICB (Figure 123).

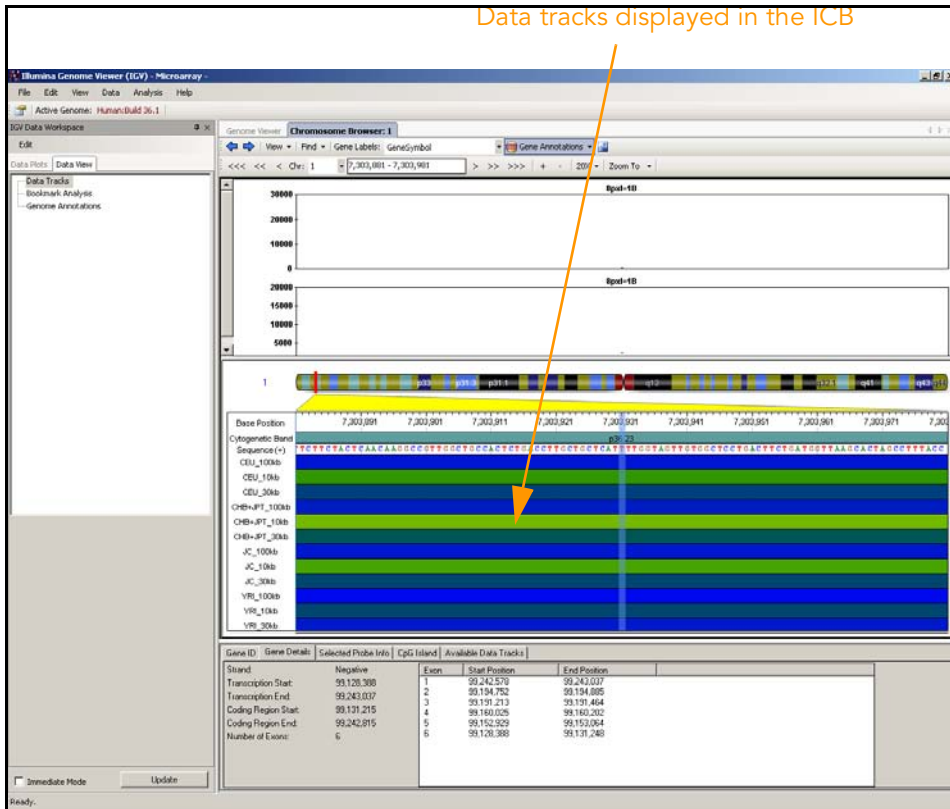


Figure 123 Data Tracks Displayed in the ICB

6. 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:

1. Create a new tab-delimited text file using Excel or another application.
2. 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 7 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 7 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 124).

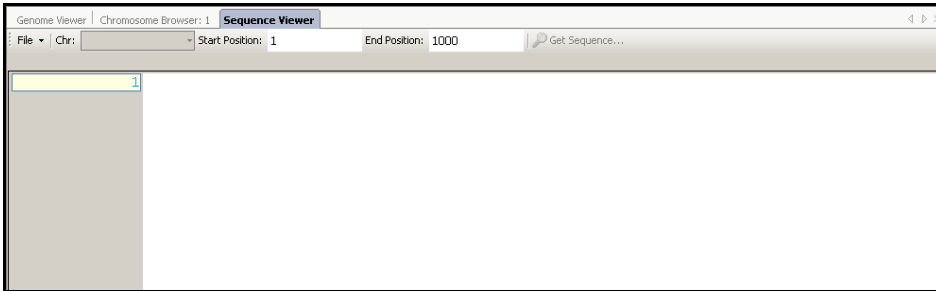


Figure 124 ISV

To view a specific sequence in the ISV:

1. Type the starting base position number of the sequence in the **Start** field.
2. Type the ending base position number of the sequence in the **End** field.
3. Click **Get Sequence**.
The sequence you selected appears in the ISV (Figure 125).



Figure 125 ISV Displaying a Selected Sequence

Using the Chromosome Heat Map

The **Chromosome Heat Map** feature is similar to the heat map feature in the GenomeStudio 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 dialog box appears (Figure 126).

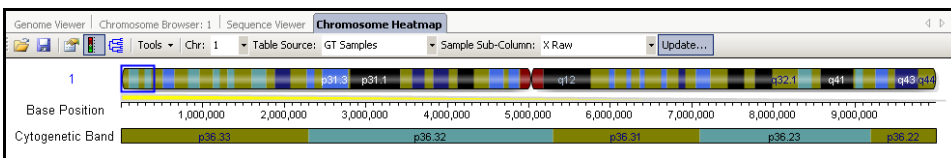


Figure 126 Chromosome Heat Map

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 127).

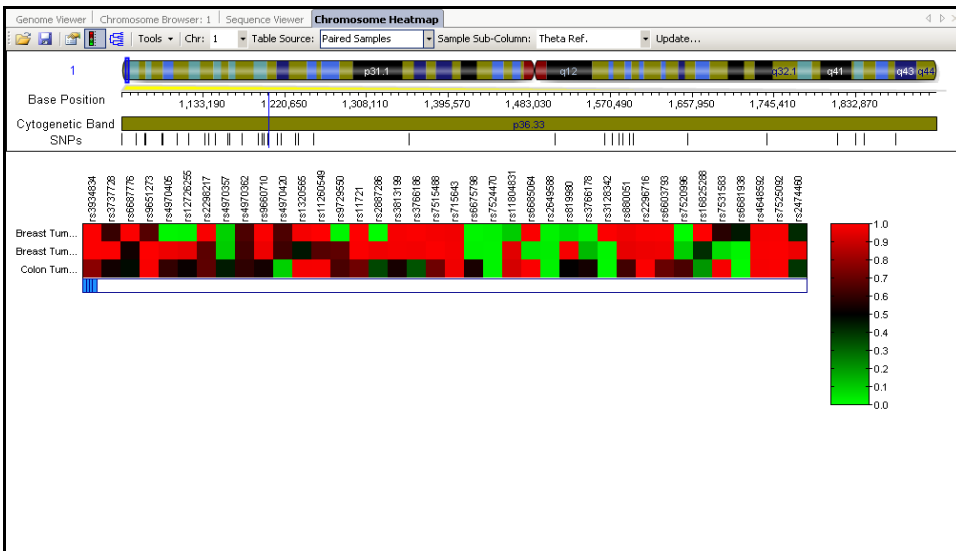



Figure 127 Chromosome Heat Map

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 dialog box, click  **Cluster heat map rows and/or columns**.
The Cluster Options dialog box appears (Figure 128).

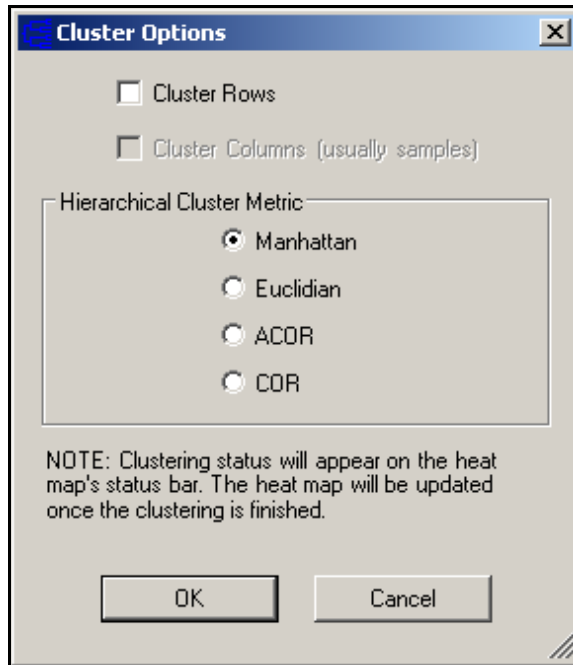


Figure 128 Cluster Options

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 129).

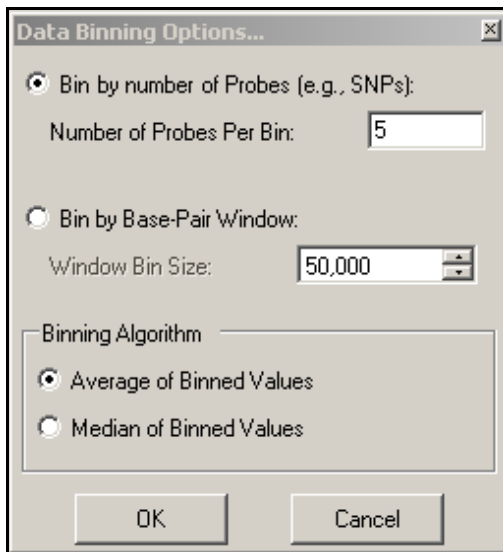


Figure 129 Data Binning Options

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 130).

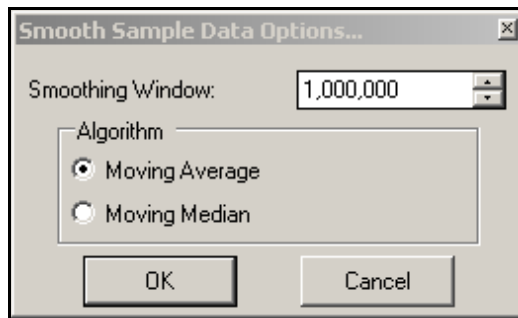


Figure 130 Smooth Sample Data Options

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.

Note:

Bookmarking Tool

Topics

- 148 Introduction
- 148 Using the Bookmarking Tool
 - 149 Viewing Bookmarks in the ICB
 - 151 Using the Bookmark Viewer
 - 155 Manually Creating Bookmarks
 - 157 Editing Bookmarks
 - 158 Saving Bookmarks
 - 161 Creating a Bookmark List
 - 162 Editing Bookmark Types

Introduction

This chapter describes how to use the bookmarking tool.

Using the Bookmarking Tool

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

The bookmarking tool shows bookmarks created by applying autobookmarking algorithms to your data, or by converting CNV analyses to bookmarks. In addition to viewing bookmarks generated by external tools, you can use the bookmarking tool to manually create your own bookmarks.

Bookmarks 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 GenomeStudio 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.

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 dialog box.

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.

The bookmark information you entered appears in a popup window (Figure 131).

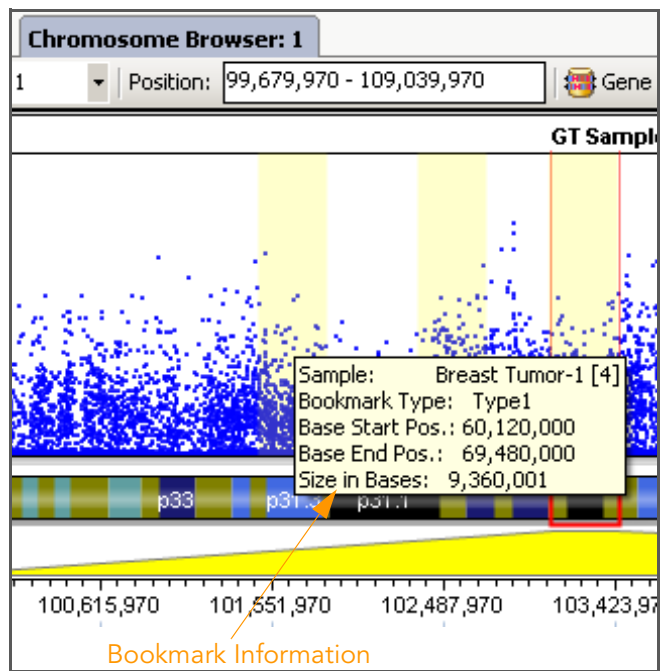


Figure 131 Bookmark Information Popup Window

Locking Bookmarks

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

To lock bookmarks:

1. Open the Bookmark dialog box and select the bookmark you want to edit.
2. Click **Edit**.
The Edit Bookmark dialog box appears (Figure 132).
3. Clear the **Enable Manual Placement** checkbox.
4. This bookmark is now fixed in place and cannot be moved in the ICB.

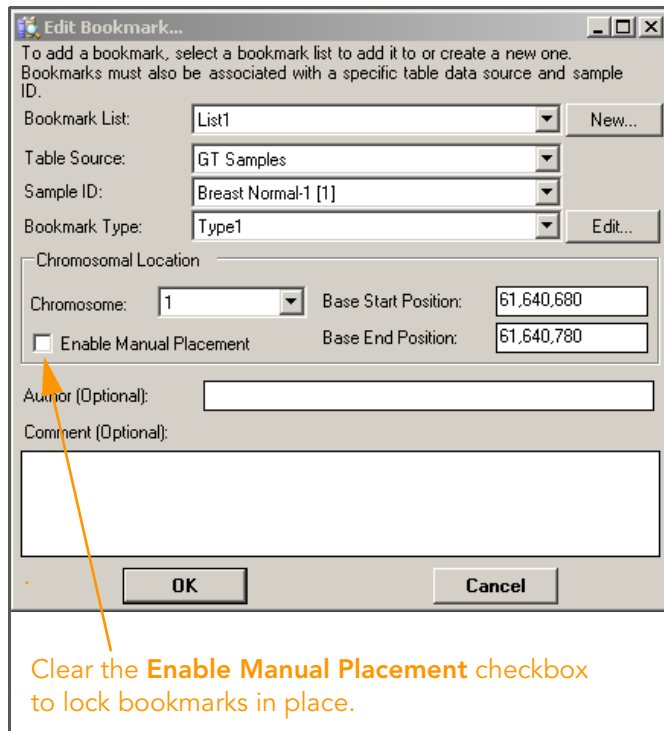


Figure 132 Enable Manual Placement Checkbox Cleared

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.

Using the Bookmark Viewer

The Bookmark Viewer is a dialog box that allows you to manage, view, and export your bookmarks.

To launch the bookmark viewer, do one of the following:

- ▶ In the main menu of the IGV, go to **View | Bookmark Viewer**.
- ▶ In the ICB toolbar, go to **View | Bookmark Viewer**.

The Bookmark Viewer appears (Figure 133).

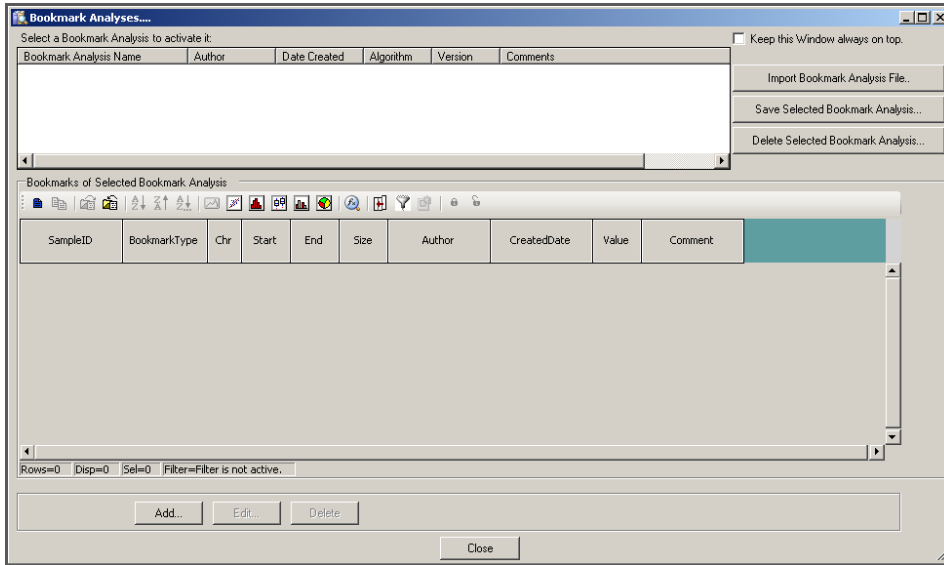


Figure 133 Bookmark Viewer

Select a bookmark analysis to see the corresponding bookmarks in the lower table (Figure 134).

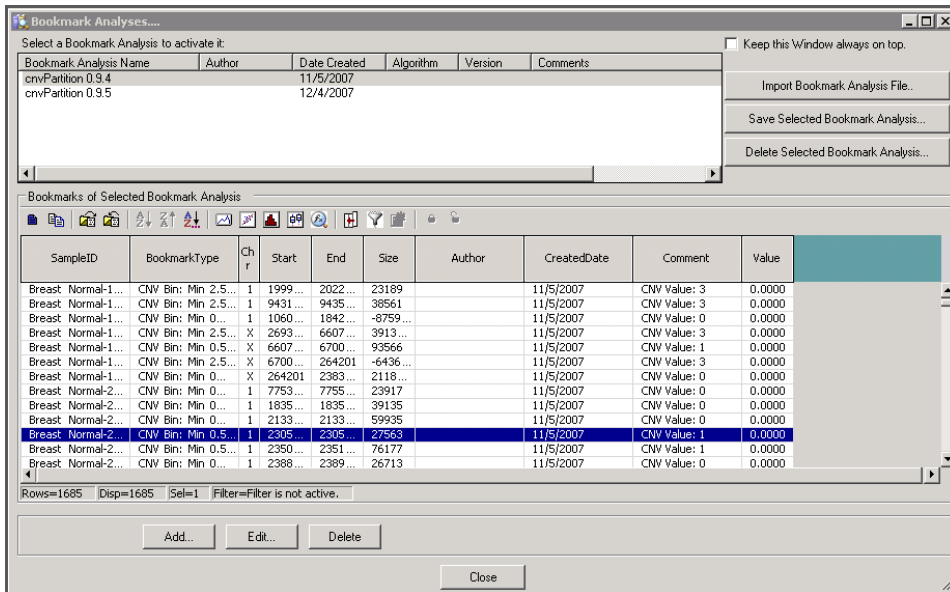


Figure 134 Bookmark Viewer with Bookmark Analysis Selected

Using the Bookmark Viewer, you can perform many of the same actions that you can perform in other GenomeStudio tables; for example: sorting, marking, filtering, and plotting.

Saving a Selected Bookmark Analysis

Saving a bookmark analysis is an easy way to share results with other GenomeStudio users.

To save a bookmark analysis:

1. Select a bookmark analysis.
2. Click **Save Selected Bookmark Analysis**.
The Save As dialog box appears, and a name for your bookmark file is automatically generated.
3. **[Optional]** Edit the name of the bookmark analysis.
4. Browse to the location where you want to save your bookmark analysis.
5. Click **Save**.
The bookmark analysis is saved as an *.xml file in the location you specified.

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

Importing Bookmarks

By importing and exporting bookmark files, you can share bookmark files with other GenomeStudio users, and save bookmark files to be used again later.



Bookmarks from one GenomeStudio project can be imported into any other GenomeStudio project as long as the sample names are the same.

To import a bookmark file:

1. In the IGV, go to **View | Bookmark Viewer**.
The Bookmark Viewer opens.
2. Click **Import Bookmark Analysis File**.

3. Browse to the location of the bookmark file you want to import.
4. Click **Open**.
The bookmarks are loaded in the Bookmark Viewer and overlaid on the data displayed in the ICB.

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**.
The Bookmark Viewer dialog box appears.
2. In the Bookmark Viewer dialog box, click once on the descriptive information for the bookmark you want to delete.
3. Click **Delete**.
The Delete Selected Bookmarks dialog box appears (Figure 135).

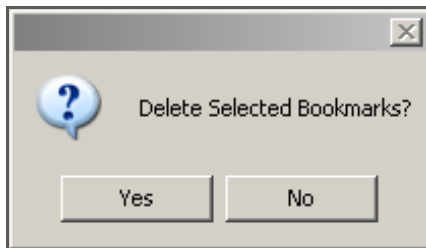


Figure 135 Delete Selected Bookmarks

4. Click **Yes**.
The bookmark is removed from the Bookmark Viewer dialog box and the ICB.

Deleting Multiple Bookmarks

To delete multiple bookmarks:

1. In the ICB, select **View | Bookmark Viewer**.
2. In the Bookmark Viewer dialog box, 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 136).

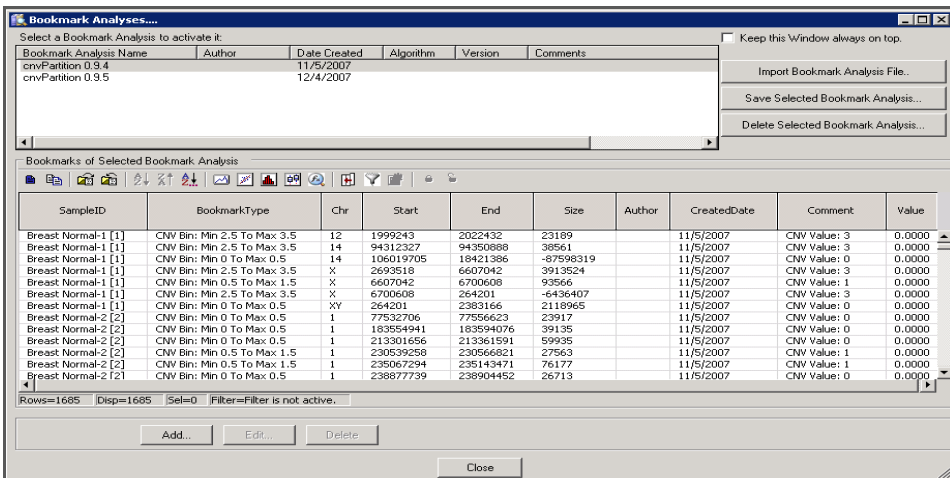


Figure 136 Deleting Multiple Bookmarks

5. Click **Delete**.
The Delete Selected Bookmarks dialog box appears.
6. Click **Yes**.
The selected bookmarks are deleted from the Bookmark Viewer dialog box and the ICB.

Manually Creating Bookmarks

To manually create bookmarks using the GenomeStudio bookmarking tool:

1. In the Bookmark Viewer, click **Add**.
The Add Bookmark dialog box appears (Figure 137).

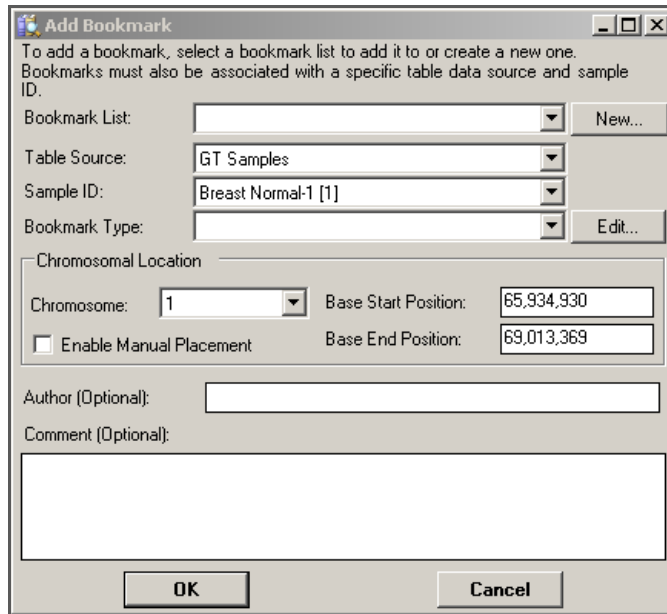


Figure 137 Add Bookmark

2. Using the dropdown lists, 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**.
3. [Optional] Create a new bookmark. See *Saving Bookmarks* on page 158.
4. [Optional] Create a new bookmark list. See *Creating a Bookmark List* on page 161.
5. [Optional] Edit bookmark characteristics. See *Editing Bookmark Types* on page 162.
6. 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 138), and saved with your project. This allows you to refer to the bookmarked region again later.

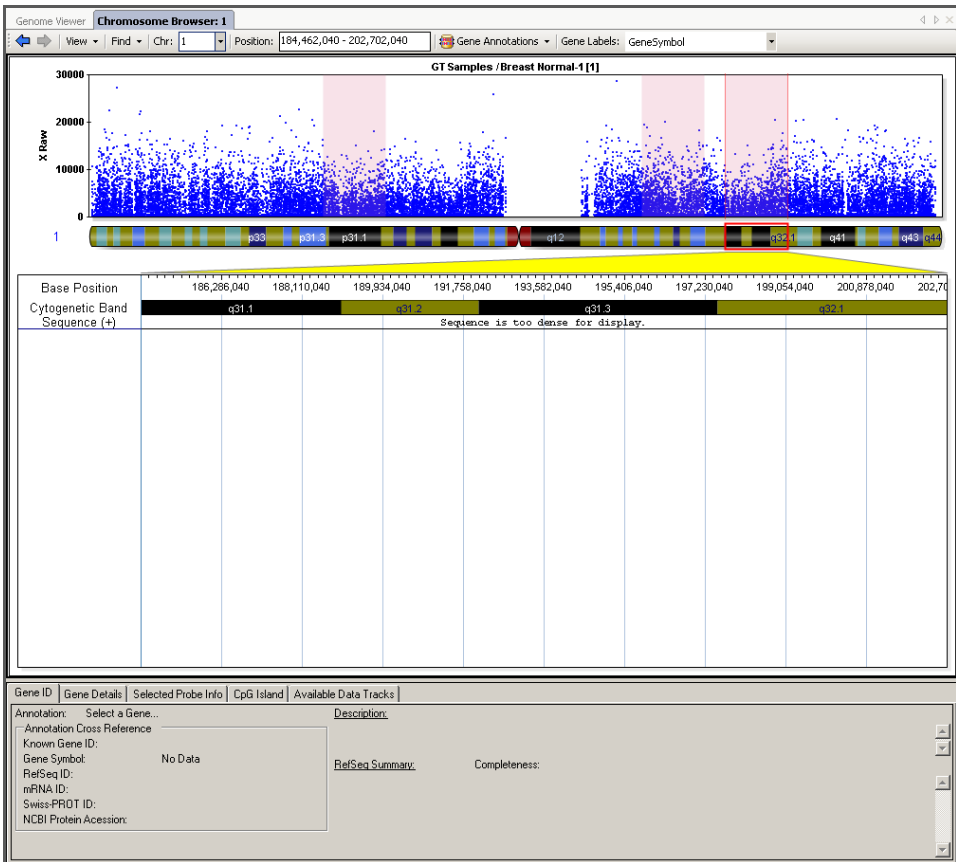


Figure 138 Bookmarks Displayed in the ICB

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

Editing Bookmarks

Bookmarks are flexible and customizable. You can move bookmarks or resize them according to your preferences. To do this, you need to make changes to the View Region, which is defined by the red rectangle shown in the ICB.

Moving a Bookmark

Perform the following steps to move a bookmark:

1. In the ICB, position the cursor over the center of the View Region you want to move.
2. Drag the View Region to the left or right.
 The bookmark is moved to the new location (Figure 139).

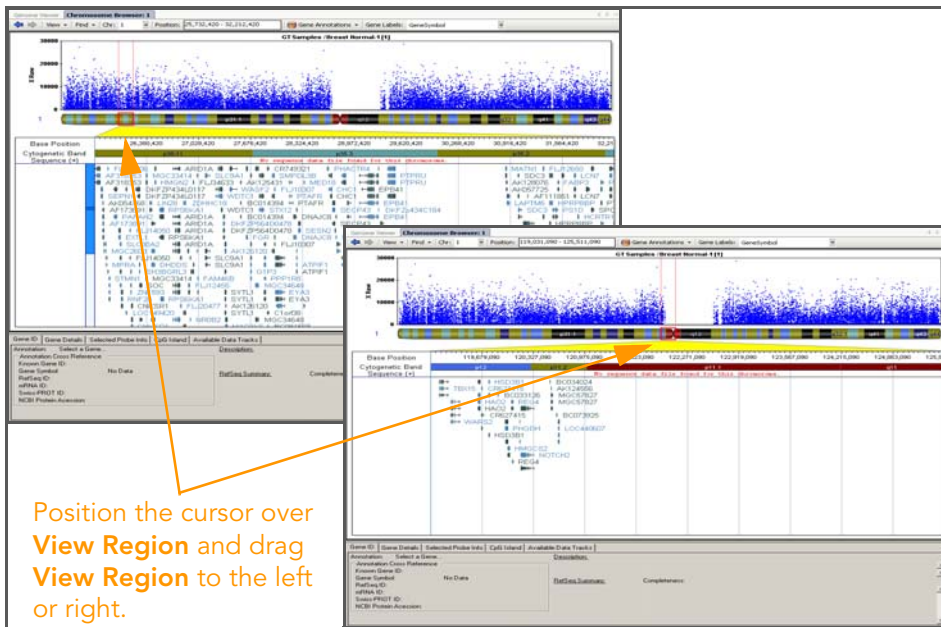


Figure 139 Moving View Region

Saving Bookmarks

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. In the ICB Context Menu, right-click and select **Add Bookmark**.

The Add Bookmark dialog box appears (Figure 140).

Figure 140 Add Bookmark

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
 - Base Start Position
 - Base End Position
 - Enable Manual Placement
 - Author
 - Comment
3. Click **OK**.
A bookmark is created in the current position of the View Region (Figure 141).

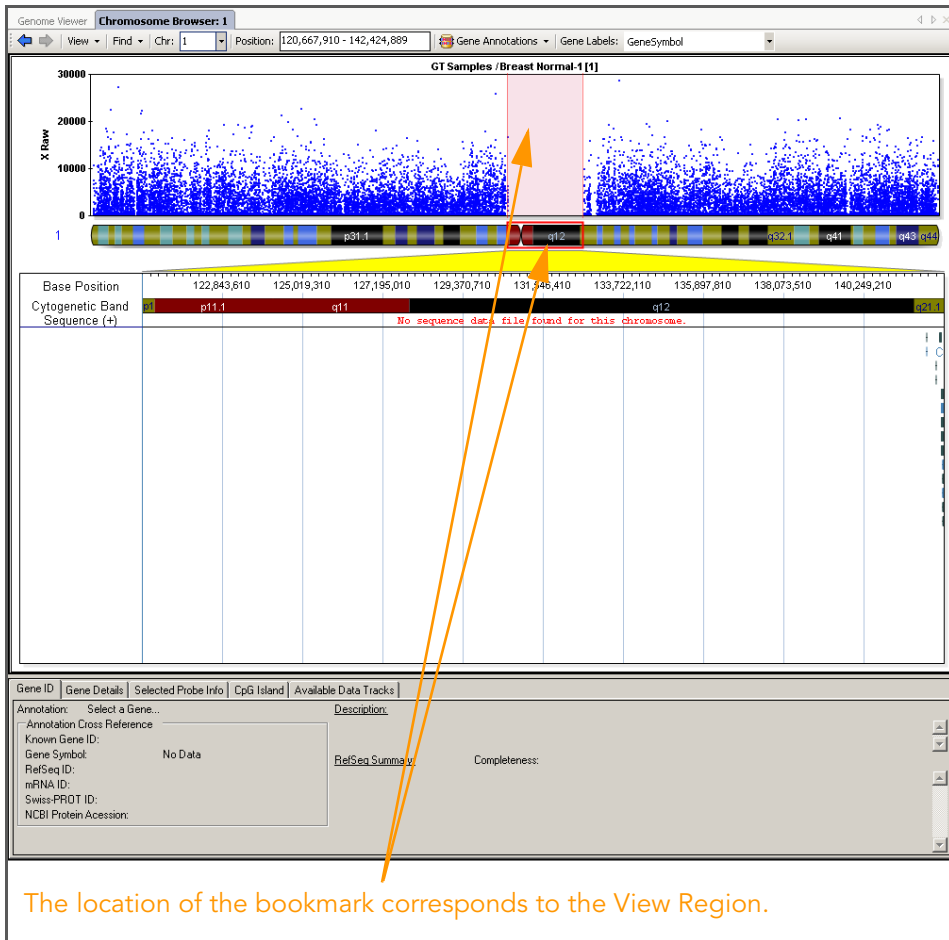


Figure 141 Bookmark Created at View Region

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 dialog box, select **New**.
The New Bookmark List dialog box appears (Figure 142).

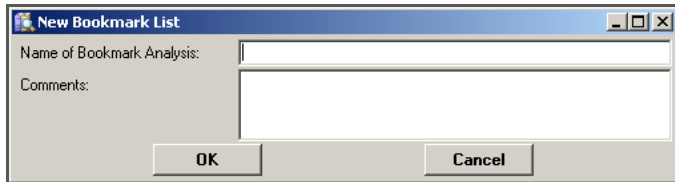


Figure 142 New Bookmark List

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

The new bookmark list is added to the selections in the **Bookmark List** dropdown list in the Add Bookmark dialog box (Figure 143).

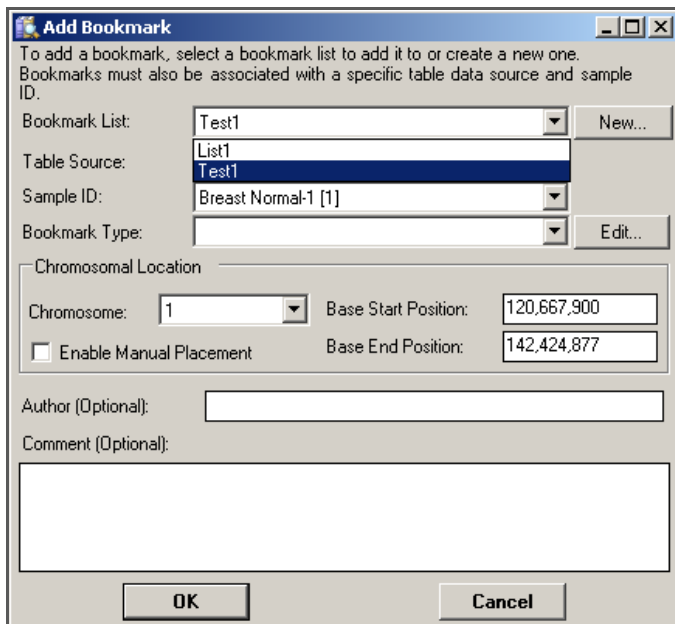


Figure 143 Add Bookmark, New Bookmark Visible

Editing Bookmark Types

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

To choose or edit 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 144).

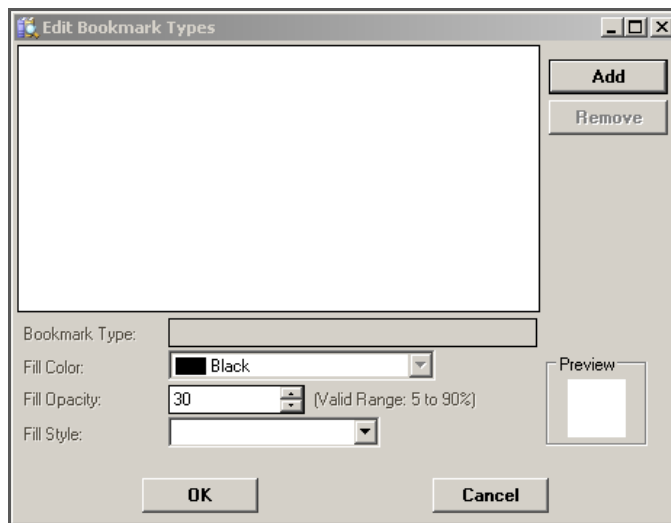


Figure 144 Edit Bookmark Types

3. Define a bookmark type for the region you want to bookmark.
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 dialog box, enter your comments in the **Comment** field (Figure 145).

Add Bookmark

To add a bookmark, select a bookmark list to add it to or create a new one. Bookmarks must also be associated with a specific table data source and sample ID.

Bookmark List:

Table Source:

Sample ID:

Bookmark Type:

Chromosomal Location

Chromosome: Base Start Position:

Enable Manual Placement Base End Position:

Author (Optional):

Comment (Optional):

Figure 145 Adding Comments to a Bookmark

2. Click **OK**.

Your comments are saved with the bookmark.



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

Index

A

- ACOR clustering method 76
- adding comments to bookmarks 163
- adjusting bookmark size and position 151
- applications
 - microarray 2
 - sequencing 2
- available genomes, IGV 112

B

- binning data, chromosome heat map 143
- bookmark analyses, IGV 112
- bookmark list, creating 161
- bookmark types, editing 162
- Bookmark Viewer 151
- bookmarking tool 148
- bookmarks 148
 - adding comments to 163
 - adjusting size and position 151
 - creating manually 155
 - deleting 154
 - displaying information 149
 - editing 157
 - importing 153
 - locking 150
 - moving 158
 - saving 153, 158
- box plots 60

C

- chromosome heat map 140
 - binning data 143
 - clustering data in 141
 - data binning in 140
 - data clustering in 140
 - data smoothing 140
 - properties 141
- chromosome slide show mode (IGV) 96, 97, 98
- chromosome heat map
 - smoothing data 144
- clearing a filter 42
- clearing marks 28
- closing GenomeStudio 10
- cluster analysis 76
- clustering data in the chromosome heat map 141
- clustering heat map data 75
- clustering methods 141
 - COR 142
 - Euclidian 142
 - Manhattan 142
- CNV analyses 148
- column chooser 43
- complex filter, creating 40
- context menu, ICB 127
- COR clustering method 76, 142
- creating a bookmark list 161
- creating a filter
 - complex 40
 - simple 39
- creating a new column 32, 33, 34
- creating a presentation image 87

D

- data
 - creating a new column 32
 - exporting to a file 29
 - importing from a file 29
 - sorting by column 31
- data binning, chromosome heat map 140
- data clustering, chromosome heat map 140
- data plots tab, IGV 106
- data properties tab, IGV 109
- data smoothing, chromosome heat map 140
- data tracks 108, 112, 134, 137
 - loading Illumina data tracks 134
 - loading your own data tracks 137
- data view tab, IGV 108
- data, viewing in the IGV 111, 113
- deleting a user-defined subcolumn 36
- deleting bookmarks 154
- dendrograms 77
- detachable docking windows 12
- display properties tab, IGV 110
- displaying bookmark information 149
- docking icons 13
- domain 7

E

- editing a user-defined subcolumn 35
- editing bookmark types 162
- editing bookmarks 157
- Euclidian clustering method 76, 142
- exons, viewing in detail 130
- exporting data to a file 29

F

- filtering table rows 38
- filters
 - clearing 42
 - loading 42
 - notes 42
 - saving 41
- finding genes or probes in the ICB 133
- Framework 2, 4
- frequency plots 61

G

- Gene Details tab, ICB 128
- Gene ID tab, ICB 128
- GenomeStudio
 - closing 10
 - domain 7
 - Framework version number 9
 - installing 3
 - main window 8
 - module version numbers 9
 - registration 7
 - serial numbers 4
 - starting 7
 - user name 7
- getting data files to view in the IGV 113
- Goldenpath (UCSC database) 113

H

- heat maps 72
 - data clustering in 75
 - populating with data 73
- hierarchical cluster metric options 76
 - ACOR 76
 - COR 76
 - Euclidian 76
 - Manhattan 76
- histograms 64
 - autoscale 66
 - clearing a filter 70
 - customizing 66
 - data range of 66
 - filtering table data in 69
 - marking a selection in 70
 - resolution of 66
 - selection area 66, 67
 - statistics area 68
 - view area 66

I

- ICB 123
 - context menu 127
 - finding genes or probes 133
 - Gene Details tab 128

- Gene ID tab 128
 - project manifest SNPs 131
 - toolbar 124
 - view region 127
 - view region, resizing 127
- I**
- IGV 90
 - available genomes 112
 - bookmark analyses 112
 - chromosome slide show mode 96, 97, 98
 - data plot type 99
 - display properties 99
 - getting data files 113
 - launching 90
 - loaded data tracks in 112
 - main window 91
 - menus 102
 - plotting data 103
 - selecting options, data plots tab 106
 - selecting options, data properties tab 109
 - selecting options, data view tab 108
 - selecting options, display properties tab 110
 - toolbar 100
 - viewing data in 111
 - whole genome view mode 96, 97
 - IGV Data Workspace 103
 - Illumina Chromosome Browser 123
 - Illumina Genome Viewer 90
 - Illumina Sequence Viewer 138
 - immediate mode (IGV) 96
 - importing bookmarks 153
 - importing data from a file 29
 - installing GenomeStudio 3
 - ISV 138
 - viewing sequences in 138
- L**
- license, GenomeStudio 4, 5
 - line plots 49
 - changing plot settings 55
 - changing plot type 54
 - changing X-axis label 52
 - plotting a single column 49
 - plotting two columns 51
 - loading a filter 42
 - locking bookmarks 150
 - locking table columns 43
- M**
- main window, GenomeStudio 8
 - main window, IGV 91
 - Manhattan clustering method 76, 142
 - manually creating bookmarks 155
 - mark color, selecting 27, 59, 71
 - mark name, selecting 27, 59, 70
 - marking samples in a plot 58
 - marking table rows 26
 - marks, clearing 28
 - menus, IGV 102
 - moving bookmarks 158
- P**
- Pearson correlation 76
 - pie charts 63
 - plots and graphs
 - bar graphs 56
 - box plots 60
 - frequency plots 61
 - heat maps 72
 - histograms 64
 - line plots 49
 - pie charts 63
 - scatter plots 57
 - plotting data in the IGV 103
 - populating heat maps with data 73
 - presentation image, creating 87
 - project manifest SNPs, viewing in the ICB 131
- R**
- registration 7
 - resizing the view region, ICB 127
- S**
- samples, marking in a plot 58
 - saving a bookmark analysis 153

- saving a filter 41
- saving bookmarks 158
- scatter plots 57
- searching in tables 24
- selecting a mark color 27, 59, 71
- selecting a mark name 27, 59, 70
- selecting options, data plots tab, IGV 106
- selecting options, data properties tab, IGV 109
- selecting options, data view tab, IGV 108

- selecting options, display properties tab, IGV 110
- sequence files 117
- serial numbers, GenomeStudio 4
- similarities and distances 76
- simple filter, creating 39
- smoothing data, chromosome heat map 144
- software license agreement 4, 5
- software version numbers 9
- sorting data by column 31
- starting GenomeStudio 7
- subcolumns
 - deleting 36
 - editing 35

T

- table columns
 - locking 43
 - selecting 22
 - showing and hiding 23
 - unlocking 45
- table rows
 - filtering 38
 - marking 26
 - selecting 21
- tables 20
 - searching in 24
 - with subcolumns 21
 - without subcolumns 20
- toolbars
 - ICB 124
 - IGV 100

U

- UCSC Goldenpath database 113
- unlocking table columns 45
- user name 7

V

- view region, ICB 127
- viewing data in the IGV 111
- viewing gene exons in detail 130
- viewing project manifest SNPs in the ICB 131

W

- whole genome view mode (IGV) 96, 97
- window configurations
 - default 17
 - saved 17