Axiom[™]Analysis Suite 3.1 USER GUIDE

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Contents

CHAPTER 1 Introduction	. 9
Overview	. 9
Software and Hardware Requirements	. 9
Sample Data Size Estimates and Required Disk Space	10
Installation Instructions	11
Starting Axiom Analysis Suite	12
Using the Preferences Window Tab	14
Changing the Default Library Folder/Path	15
Setting Up Proxy Server Access	15
Updating NetAffx Library/Annotations	16
Enabling/Disabling Check for Library File Updates at Start Up	17
Installing Custom Array Library Files	17
Uninstalling	18
Windows 7	18
Windows 10	18
CHADTED 2 Derforming on Analysis	10
	19
Setting Up an Analysis	19
Selecting a Mode (Workflow)	19
Selecting an Array Type	20
Importing CEL Files	20
Importing CEL Files by Text	21
Removing Selected CEL Files	21
Setting Up an Analysis Configuration	22
Selecting an Analysis Configuration	22
Using the Analysis Settings Fields	24
Sample QC Fields	24
Genotyping Fields	24
Saving your Analysis Configuration	26
Modifying an Existing Analysis Configuration	27
Setting Up Threshold Settings	28
Customizing Thresholds	28
Sample QC	29 29
Assigning an Output Folder Path	31
Assigning a New Output Folder Path	31
Adding Sub-Folders	31
Assigning a Batch Name	31

Running your Analysis	32
Using the Dashboard Window Tab	34
Open Selected Result(s)	35
Remove Selected Result(s)	35
Viewing the Results Folder in Windows Explorer	35
Browsing For Existing Analysis Results	36
Browsing for Existing Suitcases	37
Opening an Analysis from Windows Explorer	37

CHAPTER 3 The Viewer: Summary Window and Sample Table . . 38

Viewing Options	39
Split-Screen Options	39
Changing a Tab Window to a Full Screen Windows	41
Adjusting the Window Size	42
Summary Window/Tab	43
Data Analysis Summary	43
Viewing the Plate Barcode Table Details	44
Sample Table	45
Importing Sample Attributes	46
Column Headers	46
Rearranging Columns	47
Sorting Columns	47
Single-Click Sorting Method	47
Hiding the Column	47
Filtering Column Data	48
Adding Filters (Method 1)	48
Text-based Columns	48
Numeric Data Columns	49
Showing Filtered Data Only	50 - ·
Clearing an Individual Filter	51
Clearing All Current Filters	52 52
Adding Filters (Method 2)	52
Copying Column Data	55
Setting User Colors	55
Assigning a Color to a Sample	55
Importing Assigned Colors	56
Viewing User Colors in the Cluster Graph	58
Removing an Assigned User Color	59
Searching Keywords	60
Box Plots	61
Viewing the Default Box Plots	61
Changing the Box Plot's Scale Setting Ranges	62
Adding a New Box Plot	62
Reading Box Plot Percentiles	63
Saving the Current Box Plot View	63

Scatter Plot
Viewing the Default Scatter Plot 64
Changing the Scatter Plot's Setting Ranges 65
Adding a New Scatter Plot and Selecting its X and Y Properties
Customizing Color By Settings 67
Saving the Current Scatter Plot View 68
Plate Views
Viewing the Default Plate Views
Adding a New Plate View Metric
Customizing your Plate View Settings 71
Saving the Current Plate View
Concordance Checks
Running a Concordance Check
Comparing All Combinations
Comparing to Reference
Reanalyzing Samples

CHAPTER 4 The Viewer: SNP Summary Table and Cluster Plot . 76

SNP Summary Table	76
Using the SNP Summary Table	78
Setting your SNP Summary Table View	78
Adding and Removing Table Columns	78
Selecting Annotations	79
Saving your Table Column View	80
Copying Selected Row(s)	80
Copying Selected Cell(s)	80
Changing or Reverting Genotype Calls 8	81
Changing Genotype Calls by Text File 8	81
Reverting Selected SNPs for all Samples 8	81
Reverting all SNPs for all Samples	81
Reanalyzing your SNP Summary Table Data	82
Regenerate SNP Metrics	82
Running OTV Caller	85
Managing your SNP List	86
Saving your Current SNP List	86
Exporting your SNP List	86
Importing a SNP List	87
Using your Saved SNP List	87
Displaying SNPs in a SNP list	87
Displaying SNPs that are not in your SNP List	88

Cluster Plot	. 89
Using the Cluster Plot	. 91
Displaying a SNP Cluster Plot that Corresponds with a SNP	. 91
Setting New Scale Setting Ranges	. 92
Customizing Color By Settings	. 92
Selecting Multiple Samples in a Cluster Plot	. 94
Changing a Sample's Call for a Single SNP	. 95
Reverting a Single Call	. 95
Reverting Multiple Calls	. 95
Displaying Cluster Model Data	. 96
Saving the Current Cluster Plot View	. 96
CHAPTER 5 Allele Translation	97
About Translations	. 97
Performing Allele Translation	. 97
Allele Translation Options	. 99
Translation Reports	101
Comprehensive and Summary Translation Report	102
Summary Translation Report	102
Phenotype Translation Report	103
Phenotype Report	103
Opening Translation Report in MS Excel	104
Available Report Fields and Descriptions	105
Array Tracking	105
Gene-specific	105
Marker-specific	108
Tracking Edited Genotype Calls	109
Uncalled Report	110
CHAPTER 6 Exporting	11
Using the Sample Table Export Options	111
Using the SNP Summary Table Export Options	111
Exporting the Current Table	112
Exporting All Data	112
Exporting Signature SNPs	112
Exporting Genotyping Data	112
Result Output Formats	113
Call Output Formats	114
Exported Data Selections	114
Input and Output Files	114

SNP List Filter (Optional)115Output Location (Required)115Output Name (Required)115Changing the SNP Identifier115Changing the Current Annotation File (Optional)116Adding and Removing Annotation Columns116Exporting Cluster Plots to PDF117
CHAPTER 7 External Tools119
Axiom CNV Tool 1.1
Council on Dairy Cattle Breeding (CDCB) Export Tool 120
Axiom Long Format Export Tool 121
APPENDIX A Predefined Region CN Analysis Variations
Predefined Copy Number Analyses 122
With Predefined CN Genotyping 122
With CN-aware Genotyping 123
Genotyping with CN Analysis 124
Setting Up a Genotyping with CN Analysis 124
Selecting a Mode (Workflow) 124
Importing CEL Files
Analysis Settings with Copy Number Options 127
Sample QC
Genotyping
Threshold Configurations with CN Options 129
Sample QC
CN QC
Assigning an Output Folder Path 129
Assigning a Batch Name 129
Bunning your Genotyping with CN Analysis
Viewing your Genotyping and CN Results
Summary Report
Sample Table
SNP Summary Table
CN Summary Table and CN Region Plot 135
CN Summary Table (Overview)
CN Region Plot (Overview)
Overview and Use of the Best Practices Workflow

	APPENDIX B About Allele Translation
	Overview139Gene Table Layout for Haplotyping139Biological Annotations in Translation Reports141Impact of Phase Ambiguity in Haplotyping142Diplotype to Phenotype Translation144Creating a Custom Metabolizer Library File145Metabolizer Library File Format146Reference Databases Used in Translation Data Curation148
1	APPENDIX C Definitions
	Threshold Names149Sample Table154Concordance Columns158Annotations and Columns159SNP Summary Table Definitions162

Introduction



Overview

Axiom Analysis Suite (AxAS) enables you to perform the following functions:

- Run QC and Genotyping Algorithms.
- View QC Data within tables and graphs at a Sample and/or SNP level.
- View Cluster Graphs with the ability to change calls and/or highlight by attribute.
- Export your Data.

Software and Hardware Requirements

64-bit Operating System	Speed	Memory (RAM)	Available Disk Space ¹	Web Browser
Microsoft Windows [®] 7 (64 bit) Professional with Service Pack 1	2.83 GHz Intel Pentium Quad Core Processor	16 GB	150 GB HD + data storage Refer to table below.	Internet Explorer 8.0 and above
Microsoft Windows 10 (64 bit) Professional	2.83 GHz Intel Pentium Quad Core Processor	16 GB	150 GB HD + data storage Refer to table below.	Internet Explorer 8.0 and above

¹Minimum storage requirements are for a single run. Total storage space should include additional space for data storage of input and output files from current and previously completed analyses. In addition, you must have a minimum of 5GB of free space on your C: drive to run an analysis.

Sample Data Size Estimates and Required Disk Space

Before using AxAS, make sure you have enough disk space. See the table below for size estimates. Note: The estimates shown include the contents of the batch name folder.²

# of Markers	Storage Type ³	50 samples	100 samples	500 samples	1000 samples	5000 samples
50K	Input	1.33 GB	2.66 GB	13.3 GB	26.6 GB	133 GB
	Output	158 MB	286 MB	1.27 GB	2.51 GB	12.4 GB
	Total	1.49 GB	2.95 GB	14.57 GB	29.11 GB	145.4 GB
500K	Input	1.33 GB	2.66 GB	13.3 GB	26.6 GB	133 GB
	Output	1.53 GB	2.77 GB	12.6 GB	25.0 GB	124 GB
	Total	2.86 GB	5.43 GB	25.9 GB	51.6 GB	257 GB
850K	Input	1.33 GB	2.66 GB	13.3 GB	26.6 GB	133 GB
	Output	2.59 GB	4.69 GB	21.4 GB	42.4 GB	209 GB
	Total	3.92 GB	7.35 GB	34.7 GB	69.0 GB	342 GB

²A batch name folder is auto-generated during the analysis process. This folder includes all the necessary files needed to view your analysis results in the Viewer.

³Input is the storage size required for CEL files to be analyzed. Output is the storage size required for analysis results files.

Installation Instructions

1. Go to thermofisher.com, then navigate to the following location:

Applications and Techniques > Life Sciences > Microarray Analysis > Software > Axiom Analysis Suite

Or

Click on this link:

https://www.thermofisher.com/us/en/home/life-science/microarray-analysis/ microarray-analysis-instruments-software-services/microarray-analysissoftware/axiom-analysis-suite.html

- 2. Locate and download the zipped Axiom Analysis Suite software package.
- 3. Unzip the file, then double-click **AxiomAnalysisSuiteSetup.exe**.
- 4. Follow the on-screen instructions to complete the installation.

If your system has a previous version installed, the following message appears: (Figure 1)



Acknowledge the message, click OK, then go to "Uninstalling" on page 18.

Starting Axiom Analysis Suite

 Double-click on the Axiom Analysis Suite Desktop shortcut or click Start > All Programs > Thermo Fisher Scientific > Axiom Analysis Suite. The following window appears: (Figure 2)



- 2. Enter a new profile name or click the down-arrow to select an existing profile name.
- 3. Click OK.

The following window appears: (Figure 3)

Į.

Figure 3 Main window						
appliedbiosystems	Axiom Analysis Suite - Pete	() () _ □ ×				
New Analysis Dashboard Preferences						
Mode: Best Practices Workflow Array Type:	Import CEL Files Import CEL Files by Txt Remove Select	ed Files				
CEL Files: 0	Analysis Settings	Threshold Settings				
File Name	Select Analysis Configurations	Select Threshold Configurations				
Click "Import CEL Files" to import files for analysis.	▼ Restore Save Save As	Restore Save As				
Output Folder: C:\Users\Public\Documents\AxiomAna	lysisSuite\Output	Browse Batch Name: Run Analysis				

Using the Preferences Window Tab

Click the **Preferences** window tab (Figure 4) to setup or change a library path, edit Proxy settings, download or update Library/Annotation files.

Figure 4 Main Preferences window		
e appliedbiosystems New Analysis Dashboard Preferences	Axiom Analysis Suite - Pete	() () - □ ×
Library Folder: C:\Users\Public\Documents\AxiomAnalysisSuite\Library		Browse
Proxy Settings: Edit		
NetAffx Library/Annotations: Update		
Check for Library File Updates at Start Up: 🔽		

Changing the Default Library Folder/Path

IMPORTANT! The library folder contains the library and annotation files required to run the Axiom Analysis Suite software.

Do the following to change the default Library folder/path:

1. Click **Browse** (right of library path field).

The Select Library Folder window appears.

- 2. Navigate to the new location you want the library folder to reside.
- 3. Click New Folder.
- 4. Rename the New Folder (as you normally would), then click Select Folder.

Your newly assigned Library folder is set and reflected in the Library Folder directory/ path field, as shown in Figure 5.



Setting Up Proxy Server Access

If your system has to pass through a Proxy Server before it can access the NetAffx server (Internet), click the **Edit** button. (Figure 6)

Figure 6	Proxy Settings
Proxy Se	ttings: Edit

The following window appears: (Figure 7)

Figure 7 Proxy S	Settings Editor window
Proxy Settings Editor	×
Enable Proxy Server Settings:	
Proxy Server Address:	
Proxy Server Port:	
Proxy User:	
Proxy Password:	
	OK Cancel

- 5. Click the **Enable Proxy Server Settings** check box (Figure 7), then contact your IT department for help with completing the required text fields.
- 6. Click OK.

Updating NetAffx Library/Annotations

1. Click on the **Update** button. (Figure 8)



The following window appears: (Figure 9)

Figure 9	NetAffx Login window	
NetAffx User L	ogin Information	×
User:		
Password:		
	ОК	Cancel

2. Enter your NetAffx account email and password, then click **OK** or go to www.netaffx.com and click **Register** to sign up.

Note: If you are unable to connect to NetAffx, make sure you have an active Internet connection, and/or correct Proxy Server settings.

The NetAffx Update window appears. (Figure 10)

Figure 10	NetAffx U	pdate window		
NetAffx Update				:
Check/Uncheck	All			
Update?	Installed	Name	Аггау Туре	Comments
		Analysis Thresholds	Analysis Thresholds	A
		Axiom 384HT Trout Genotyping Array r1	Axiom_Omy50Kv2	
		Axiom 384HT Wheat Breeders Genotyping Array r3	Axiom_WhtBrd-1	
		Axiom Biobank Genotyping Array r2	Axiom_BioBank1	
		Axiom BOS1 Genotyping Array r3	Axiom_GW_Bos_SNP_1	E
		Axiom Buffalo Genotyping Array r2	Axiom_Buffalo	
		Axiom Chicken Genotyping Array r1	Axiom_GW_GT_Chicken	
		Axiom Cotton Genotyping Array r1	Axiom_Cotton	
		Axiom Equine Genotyping Array r1	Axiom_MNEc670	
		Axiom Exome 1 Array r2	Axiom_Exome_1A	
		Axiom Exome 319 Array r1	Axiom_Exome319	
		Axiom Genome-Wide AFR 1 Array - World Array 3 r3	Axiom_KP_UCSF_AFR	
		Axiom Genome-Wide ASI 1 Array r6	Axiom_GW_ASI_SNP	
V	 Image: A second s	Axiom Genome-Wide CEU 1 Array r6	Axiom_GW_Hu_SNP	Update Available
		Axiom Genome-Wide CHB 1 Array r4	Axiom_GW_Hu-CHB_SNP	
		Axiom Genome-Wide CHB 2 Array r1	Axiom_GW_CHB2	
		Axiom Genome-Wide EAS 1 Array - World Array 2 r7	Axiom_KP_UCSF_EAS	
				OK Cancel

3. You must click the check box(es) that correspond with the type of CEL files you want to analyze.

Click the **Check/Uncheck All** check box to select/deselect all the listed check boxes.

4. Click OK.

An Installing Updates progress bar appears.

Enabling/Disabling Check for Library File Updates at Start Up This check box (Figure 11) is checked by default to enable automatic Library File update alerts each time you launch the Axiom Analysis Suite application. (Recommended)



Installing Custom Array Library Files

IMPORTANT! Library files for custom designs must be manually installed.

- 1. Download the zip package provided to you by Affymetrix Bioinformatics Services.
- 2. Unzip the contents of the analysis library files into a single sub-folder within the library file folder.

For multi-species designs, each species should be in its own sub-folder. There should be no other folders within each sub-folder and all annotation information must be in the same location as the .CDF file.

Uninstalling

IMPORTANT! Upgrade installations are NOT supported. You must uninstall the existing version of Axiom Analysis Suite BEFORE installing this new version. Administrative rights to the computer are required before you can uninstall the Axiom Analysis Suite software. For your convenience, no existing library files or user settings are removed during the uninstall process.

Windows 7	1. Click Start > Control Panel.
	The Control Panel window appears.
	2. Click the View by drop-down menu (upper-right), then click to select Category .
	3. In the Programs category, click Uninstall a program .
	The Programs and Features window appears.
	4. Click to select Axiom Analysis Suite , then click Uninstall .
	5. Follow the on-screen instructions.
	6. After the uninstall process is complete, close the Programs and Features window.
	 Use Windows Explorer as you normally would to navigate to the directory: C:\Program Files\Affymetrix
	8. Verify that the Axiom Analysis Suite folder has been removed.
	9. If the folder is present, double-click on it to open it.
	10. Search for any files you want to keep, then move them to different (easily accessible) location.
	11. Delete the Axiom Analysis Suite folder.
	12. Close all open windows, then install the new version, as described in the "Installation Instructions" on page 11.
Windows 10	1. Click the Windows icon (bottom left corner).
	2. Click All apps > Windows System > Control Panel.
	The Control Panel window appears.
	3. In the Programs category, click Uninstall a program .
	The Programs and Features window appears.
	4. Click to select Axiom Analysis Suite, then click Uninstall.
	5. Follow the on-screen instructions.
	6. After the uninstall process is complete, close all open windows.
	 Use Windows Explorer as you normally would to navigate to the directory: C:\Program Files\Affymetrix
	8. Verify that the Axiom Analysis Suite folder has been removed.
	9. If the folder is present, double-click on it to open it.
	10. Search for any files you want to keep, then move them to different (easily accessible) location.
	11. Delete the Axiom Analysis Suite folder.
	12. Close all open windows, then install the new version, as described in the "Installation Instructions" on page 11.



Performing an Analysis

After downloading the library and annotation files that match the array type of the CEL files you want to analyze, click the **New Analysis** tab.

The New Analysis window and its three individual panes appear, as shown in Figure 12.

Setting Up an Analysis

40 14 1

.

If you want to run and view CN-aware genotypes, go to page 122. If not, continue to "Selecting a Mode (Workflow)".

Selecting a Mode (Workflow)

—:--

1. From the main Axiom Analysis Suite window tab, click the **Mode** drop-down. (Figure 12)

rigure 12 Main window			
applied biosystems	Axiom Analysis Suite - Pete	() _ □ ×	
New Analysis Dashboard Preferences			
Mode: Best Practices Workflow 🔪 Array Type:	Import CEL Files Import CEL Files by Txt Remove Select	ed Files	
CEL Files: 0	Analysis Settings	Threshold Settings	
File Name	Select Analysis Configurations	Select Threshold Configurations	
	Restore Save Save As	Restore Save Save As	
Olick "Import CEL Eiles" to import files for			
analysis.			
Output Folder: C:\Users\Public\Documents\AxiomAn	LalysisSuite\Output	Browse Batch Name: Run Analysis	

2. Click to select the workflow you want to use.

•	Best Practices Workflow (Default): This workflow performs quality control	
	analysis for samples and plates, genotypes those samples which pass the defined	
	QC thresholds, and then categorizes the probe sets to identify those whose	
	genotypes are recommended for statistical tests in downstream study. Details are	
	available in the Axiom Genotyping Solution Data Analysis Guide (P/N 702961)	

- Sample QC: This workflow performs the quality control analysis for samples and plates. Note this workflow does not produce genotype calls for the passing samples.
- Genotyping: This performs genotyping on the imported CEL files, regardless of the sample and plate QC metrics. Note: Including samples that do not pass defined QC thresholds may reduce the quality of the results for passing samples.
- Summary Only: This workflow produces a summary of the intensities for the probe sets for use in copy number analysis tools. Note: Summary Only does not perform sample QC nor genotyping.
- Selecting an Array 1. Click the **Array Type** drop-down to select the array type you want to use.

Importing CEL Files

Type

1. Click Import CEL Files.

The Add CEL Files window appears.

- 2. Navigate to your CEL file location. Make sure the CEL Files you select coincide with the array type you selected earlier, otherwise a warning message appears.
- 3. Single-click on a CEL file or Ctrl click, Shift click, or press Ctrl A (to select multiple files).
- 4. Click Open.

The CEL Files pane populates and displays your selected CEL files. (Figure 13)

Figure 13 Populated CEL File pane example
CEL Files: 12
File Name
2877_AxiomGWHuSNP1_20101115_Blood_G01
2878_AxiomGWHuSNP1_20101115_Blood_H01
2879_AxiomGWHuSNP1_20101115_Blood_A02
2880_AxiomGWHuSNP1_20101115_Blood_B02
2881_AxiomGWHuSNP1_20101115_Blood_C02
2882_AxiomGWHuSNP1_20101115_Blood_D02



IMPORTANT! The CEL files paths in a TXT file are not recognized (and will not import) if they begin with back slashes (e.g: \path\to\my\file.cel).

Before importing, the two starting back slashes must be replaced with forward slashes (e.g: //path\to\my\file.cel).

Also, your *.txt list file must start with the header cel_files, as shown in Figure 14.

1. Click Import CEL Files by Txt.

The Import CEL Files by Txt window appears.

2. Navigate to the .txt file that contains the list of CEL files you want to process.

Figure 14 Text CEL file list example shown in Notepad		
ſ	CelFileList.txt - Notepad	
L	File Edit Format View Help	
	<u>cel_files</u> //10.0.55.13/r/AGATHA/Raw data/PharmacoScan/5507464290134070917891_v12_DMET_1_cel/NA12043_v12_10uLmPCRspike_ i	

Make sure the CEL Files you select coincide with the array type you selected earlier, otherwise a warning message appears.

- **3.** Single-click on a CEL file or Ctrl click, Shift click, or press Ctrl A (to select multiple files).
- 4. Click Open.

Your CEL Files pane populates and displays each CEL file extracted from your selected text file.

- Removing Selected Use this option to remove unwanted CEL files.
 - 1. Single-click on a CEL file or Ctrl click, Shift click, or press Ctrl A (to select multiple files), then click **Remove Selected Files**.



Setting Up an Analysis Configuration

The Analysis Settings are populated based on the Mode (Workflow) chosen. For example, if Genotyping mode is selected, the Sample QC section of the Analysis Settings is hidden and only the Genotyping section is visible.

Selecting an Analysis Configuration 1. It is highly recommended you click the drop-down menu (Figure 15) and select the option that best matches the number of samples you want to analyze.

Note: The default configuration options displayed in the drop-down menu are based on your array type.

Figure 15	5 Select an analysis configuration drop-down menu	
Analysis Settings		
Select Analys	sis Configurations	
Axiom_GW_H	lu_SNP_96orMore.r6 (Default) ▼	Restore Save Save As
Axiom_GW_H	lu_SNP_96orMore.r6 (Default)	
Axiom_GW_F	lu_SNP_LessThan96.r6 (Default)]

After selecting the appropriate default for the number of your samples, the Analysis Setting pane auto-populates, as shown in Figure 16.

Figure 16 Auto-populated Analysis Setting pane		
example		
Analysis Settings		
Select Analysis Configurations		
Axiom_PMRA_96orMore.r1 (Default)		
Sample OC		
Analysis File:		
Axiom_PMRA_96orMore_Step1.r1.apt-genotype-axiom.AxiomGT1.apt2		
Prior Model File:		
Axiom_PMRA.r1.generic_prior		
SNP List File:		
Axiom_PMRA.r1.step1 🗙		
Gender File (optional):		
Hints/Inbred File (optional):		
💿 Inbred 💿 Hints 🛛 🛄 🔀		
Analysis File:		
Axiom_PMRA.r1.apt-genotype-axiom.AxiomGT1.apt2		
Prior Model File:		
Axiom_PMRA.r1.generic_prior		
Multi-allele Background Prior Model File:		
Axiom_PMRA.r1.generic_prior.mmb		
Multi-allele Pairwise Prior Model File:		
Axiom_PMRA.r1.generic_prior.mmp		
Multi-allele Prior Model File:		
Axiom_PMRA.r1.generic_prior.mm		
SNP List File:		
Axiom_PMRA.r1.step2		
Gender File (optional):		
Hints/Inbred File (optional):		
Dinbred O Hints		
rosterior File Name (optional):		
nr2ran File (recommended)		
Aviom DMRA (1 ns2cnn man		
Axion_rmioAxit.pszsnp_map		

2

Using the Analysis Settings Fields Follow the instructions below to create a new analysis configuration or edit a prepopulated field(s).

Sample QC Fields

- 1. Click the Analysis File drop-down button to select the appropriate XML file.
- 2. Click the **Prior Model File** Browse button.

The Prior Model File window appears.

- **3.** Navigate and select the appropriate file, then click **Open**. Your newly assigned filename is displayed.
- 4. (Optional) Click the **SNP List File** Browse ... button. The SNP List File window appears.
- 5. Navigate and select the appropriate file, then click **Open**. Your newly assigned filename is displayed.
- 6. (Optional) Click the **Gender File** Browse ... button. The Gender File window appears.
- Navigate and select the appropriate file, then click **Open**. Your assigned filename is displayed.
- 8. (Optional) Click the **Hints/Inbred File** Browse ... button. The Hints/Inbred File window appears.
- Navigate and select the appropriate file, then click Open. Your newly assigned path is displayed.
- 10. Click the either the **Inbred** or **Hints** radio button.

Genotyping Fields

- 1. Click the Analysis File drop-down button to select the appropriate XML file.
- 2. Click the **Prior Model File** Browse ... button.

The Prior Model File window appears.

- 3. If multi-allelic probesets are available, the following fields appear:
 - Multi-allele Background Prior Model File
 - Multi-allele Pairwise Prior Model File
 - Multi-allele Prior Model File
 - If you want to change the currently displayed Model file, click the appropriate Multi-allele Browse _____ button to select a different Model file.
- Navigate and select the appropriate file, then click Open. Your newly assigned filename is displayed.
- (Optional) Click the SNP List File Browse ... button. The SNP List File window appears.
- 6. Navigate and select the appropriate file, then click **Open**. Your newly assigned filename is displayed.
- (Optional) Click the Gender File Browse button. The Gender File window appears.

- Navigate and select the appropriate file, then click Open. Your assigned filename is displayed.
- 9. (Optional) Click the **Hints/Inbred File** Browse button. The Hints/Inbred File window appears.
- 10. Navigate and select the appropriate file, then click **Open**. Your assigned filename is displayed.
- 11. Click the either the Inbred or Hints radio button.
- 12. (Optional) Click the **Posterior File Name** Browse button. The Posterior File Name window appears.
- **13**. Navigate to a location where you want to save your posterior file, enter a name, then click **Open**.

Your assigned filename is displayed.

14. Click the **ps2snp File** Browse ... button.

The ps2snp File window appears.

15. Navigate to your ps2snp-file location, then click **Open**.

Your newly assigned filename is displayed.

16. (Optional) If the library package supports it, click the **Genotype Frequency** Browse button.

The Genotype Frequency window appears.

17. Navigate to a location for your genotype frequency file, enter a name, then click **Open**.

Your assigned filename is displayed.

2

Saving your Analysis Configuration

1. After editing your Analysis Configuration settings, click Save (top of Analysis Setting pane). (Figure 17)

Figure 17	Analysis Configuration Save window	
Analysis Setting	s	
Select Analysis Configurations		
Axiom_GW_Hu_S	SNP_96orMore.r6 (Default) Restore Save As	

If the Analysis Configuration that came with the library package already exists, then a **Save Analysis Configuration** window appears. (Figure 18)

Figure 18 Analys Save window	sis Config	juration
Save Analysis Configuration		×
Enter a new or select an existin	ig configuratior	i name
	ОК	Cancel

2. Enter a different configuration name, then click OK.

Your new Analysis Configuration name is saved and available for use in the **Select Analysis Configuration** drop-down menu.

Modifying an Existing Analysis Configuration

1. Click the **Select Analysis Configuration** drop-down, then click to select the saved analysis configuration you want to modify.

Do one or more of the following to modify an existing analysis configuration:

- Click the applicable File field's **Browse** button to navigate to a different location, then click **Open** to reassign its path.
- If needed, click a File field's 🔀 button to delete a displayed path setting.
- Click the **Restore** button to return to the last saved values of the analysis configuration file.
- Click the Save button to overwrite your previously saved configuration
- Click the **Save As** button to save your modified configuration with a different name. [Recommended]



Setting Up Threshold Settings

The settings shown in the **Threshold Setting** pane (Figure 19) are based on the Mode (Workflow) you selected.

For Sample QC and SNP QC name definitions, see "Threshold Names" on page 149.

Figure 19Automated QC Mode ThresholdSettings pane example			
Threshold Settings			
Select Threshold Configura	tions		
Human (Default)	▼ Restore Save Save	ave As	
🔿 Sample QC			
Name	Settings		
DQC	≥ ▼ 0.82	ຄ	
QC call_rate	≥ ▼ 97	5	
Percent of passing sampl	≥ ▼ 95	5	
Average call rate for pass	≥ ▼ 98.5	5	
🔿 SNP QC			
Name	Settings		
species-type	Human	ຄ	
cr-cutoff	≥ ▼ 95	ຄ	
fld-cutoff	≥ ▼ 3.6	ຄ	
het-so-cutoff	≥ ▼ -0.1	ຄ	
het-so-otv-cutoff	≥ ▼ -0.3	5	
hom-ro-1-cutoff	≥ ▼ 0.6	ຄ	
hom-ro-2-cutoff	≥ ▼ 0.3	5	
hom-ro-3-cutoff	≥ ▼ -0.9	5	
hom-ro	true 🔹	5	
hom-het	true 🔹	ຄ	
num-minor-allele-cutoff	≥ ▼ 2	ຄ	
priority-order	Change List Order PolyHighResolution, NoMi	ຄ	
recommended	Checklist PolyHighResolution, NoMinorHom,	5	

Customizing Thresholds

Click the **Select Threshold Configuration** drop-down (Figure 20) to select an appropriate Default Threshold for your starting point. Note: The comparison signs/ operators are preset and cannot be changed.

Figure 20 Select Threshold Configuration	
Select Threshold Configurations	
Diploid (Default)	•
Diploid (Default)	
Human (Default)	
Polyploid (Default)	

Sample QC

All the Sample QC Threshold Settings are populated with default values.

- 1. Click inside each text field to enter a different value, as shown in Figure 21.
 - Click the text field's **I** button to return its value back to its last saved value within the threshold configuration file.

Figure 21 Threshold Name text field example		
DQC	≥ ▼ 0.82	ຄ

SNP QC

- 1. Click the **species-type** drop-down menu to select a different species type.
- 2. Click inside each text field to enter a different value, as shown in Figure 22. Note: General Rule: The het-so-otv-cutoff should be less or equal to het-so-cutoff and het-so-XChr-cutoff.

Figure 22 SNP QC text fields		
cr-cutoff	≥ ▼ 95	ຄ
fld-cutoff	≥ ▼ 3.6	5
het-so-cutoff	≥ ▼ -0.1	5
het-so-XChr-cutoff	≥ ▼ -0.1	5
het-so-otv-cutoff	≥ ▼ -0.3	5

Note: To return a value back to its last saved value, click the text field's souther.

- **3.** Use the **hom-ro and hom-het** drop-down menus to change their True or False values.
- 4. Click inside the **num-minor-allele-cutoff** text field to enter a different value, as shown in Figure 23.

Figure 23 SN	P QC text fields
num-minor-allele-cutoff	2 2

5. The priority-order option enables you to change the order of categories when determining which probesets are selected as the best probeset for a SNP. To change the priority-order of your SNP QC Metric, click Change List Order.

The following window appears: (Figure 24)

Figure 24 Change the Priority Order window
Change the Priority Order
Click on a name, then drag and drop it to its new position. After your list is set, click OK.
Name
PolyHighResolution
NoMinorHom
OTV
MonoHighResolution
CallRateBelowThreshold
OK Cancel

6. Click and hold onto the selection you want to move, then drag and drop it into its new position. After you get the order of priority you want, click **OK**.

Note: To return the list back to its default priority, click the priority-order field's button.

7. Use the **recommended** checklist to choose the PS_Classification conversion types for your analysis. To change the recommended options, click Checklist.

The recommended window appears. (Figure 25)

F	Figure 25 Recommended window	
	recommended	×
	PolyHighResolution	1
	✓ NoMinorHom	
	ΟΤV	
	MonoHighResolution	
	CallRateBelowThreshold	
L		-
	OK Cancel	

8. Click to check/uncheck the available recommended options, then click OK.

Note: If all recommended options are unchecked, the software uses the following default values:

- Human: PolyHighResolution, NoMinorHom, MonoHighResolution
- Diploid: PolyHighResolution, NoMinorHom, MonoHighResolution
- Polyploid: PolyHighResolution

Assigning an Output Folder Path

Assigning a New Output Folder Path

1. Click the **Output Folder** path's **Browse** button. (Figure 26)

Figure 26 Output Folder field	
Output Folder: C:\Users\Public\Documents\AxiomAnalysisSuite\Output\	Browse

An Explorer window appears.

2. Navigate to the recommended path C:\Users\Public\Documents\AxiomAnalysisSuite\Output, then click **Select Folder**.

Your selected output folder path is now displayed.

Adding Sub-Folders

Note: To better organize your output results, you can add sub-folders to your newly assigned output result path's folder.

To add sub-folders to your newly assigned result path's folder:

- 1. Click the Output Folder's **Browse** button to return to your assigned output path and/or folder.
- 2. In the Explorer window, click New Folder.
- 3. Enter a sub-folder name.
- 4. Click Select Folder.

The newly created sub-folder now appears in the output result information window.

5. Repeat the above steps 1-4 to add more sub-folders, then click Select Folder.

Assigning a Batch	The batch file is produced while your analysis is running and includes all the necessary
Name	files needed to view your analysis in the Axiom Analysis Suite Viewer.

1. Enter a name in the **Batch Name** field. (Figure 27)

IMPORTANT! Each Batch Name you create must be unique for the set of batches listed in the Dashboard window tab and unique within the same destination folder.

Figure 27 Name	Enter a Batch	
Batch Name:	Run Analysis	

Note: A folder (with the same name as your entered batch name) is auto-generated during the analysis process. This folder includes all the necessary files needed to view your analysis results in the Viewer.

Running your Analysis

- 1. Click Run Analysis.
- If you have not saved any changes to your configured Analysis Settings, a Save Analysis Configuration window appears. (Figure 28) Click **Yes**.

Figure 28 Save Analysis Configuration prompt window		
Save Analysis Configuration	×	
The current analysis configuration has not been saved. Do you want to save the current analysis configuration before proceeding?		
Yes	lo	

• Enter a new analysis name or use the drop-down to select a previously saved name, then click **OK**. (Figure 29)

Figure 29 Save Analysis Configuration window	
Save Analysis Configuration	×
Enter a new or select an existing configuration name	•
OK Car	icel

• If you have not saved any changes to your configured Threshold Settings, a Save Threshold Configuration window appears. (Figure 30) Click **Yes**.

Figure 30 Save Threshold Settings prompt window					
Save Threshold Configuration					
The current threshold configuration has not been saved. Do you want to save the current threshold configuration before proceeding?					
Yes No					

2

• Enter a new threshold name or use the drop-down to select a previously saved name, then click **OK**.

Figure 31 window	Save Threshol	d Settings				
Save Threshold Co	onfiguration	×				
Enter a new or select an existing configuration name						
		•				
	ОК	Cancel				

The Dashboard window/tab appears and shows the status of your running analysis. (Figure 32) Click Stop to cancel an analysis in progress.

Figure	Figure 32 Dashboard window/tab - Status bar and Stop button example									
New Analysis	New Analysis Dashboard Preferences									
Workflows	Workflows									
Batch Name	Date Created	Workfl	Array Type	Status	Elapsed	Status Message	Warning	Action		
Test_4	1/15/2015 12:40:10 PM	Best Practices Workflow	Axiom_GW	19 %	2 minutes	GenotypeNodeGenotyping::doRun() start	-	Stop		

Using the Dashboard Window Tab

v Analysis Dashboard Pref orkflows atch Name Date Created Wor Testl 9/24/2014 3:44:51 PM Best PH Wor Wor Test_9 9/8/2014 10:14:40 AM Best PH Wor Wor Test_7 9/4/2014 2:42:41 PM Best PH Wor Wor Test_6 9/4/2014 2:26:08 PM Best PH Wor	Dashboard window				
orkflows Unit Order	Dashboard Preferences				
atch Name Date Created Wor Testl 9/24/2014 Best P. Test_9 9/8/2014 Best P. Test_7 9/4/2014 Best P. Test_6 9/4/2014 Best P. Test_6 9/4/2014 Best P. Word 2:26:08 PM Word	▼				
Test_9 9/8/2014 10:14:40 AM Best Pr Wor Test_7 9/4/2014 2:42:41 PM Best Pr Wor Test_6 9/4/2014 2:26:08 PM Best Pr Wor	Ite Created Workfl Array Type S 9/24/2014 Best Practices Axiom_GW Image: Compare the second s	O % Elapsed 0 % 25 seconds	Status Message Opening cdf file: C:/Users/ppavic/Desktop/ Axiom_Console_Library_Files/Axiom_GW_Hu_SNP/ Axiom_GW_Hu_SNP.i6.cdf	Warning Error in running Signature SNP Analysis.	Action
9/4/2014 2:42:41 PM Best PI Worl Test_6 9/4/2014 2:26:08 PM Best PI Worl	9/8/2014 Best Practices Axiom_GW S	uccess		Plate(s) 5500944105295011411127 . Plate pass rate < 95	Oper
Test.6 9/4/2014 2:26:08 PM Work	9/4/2014 Best Practices Axiom_GW S	uccess		Plate(s) 5500944105295011411127 . Plate pass rate < 95	Oper
	9/4/2014 Best Practices Axiom_GW I	Failed	Could not find file 'C:\Users\Public\Documents \Axiom_Analysis_Suite_Beta\Output\Test_6 \CELFileList_win.txt'.	Could not find file 'C:\Users\Public\Documents \Axiom_Analysis_Suite_Beta\Output\Test_6 \CELFileList_win.txt'.	Start

The Dashboard tab window displays existing results. (Figure 33)

2

Open Selected	Do one of the following to open a selected result:
Result(s)	 Click on the Action column's Open button to open a study.
	Note: The Open button is relabeled Start if the Dashboard's Status column displays Failed. A workflow can fail if there is a processing error (for example, needed files are unavailable) or if no samples pass QC. If the Warning column indicates that no samples passed QC, yet you still want to open the study to review its QC results, use one of the alternate methods below to open a selected study.
	 Single-click on a study, then click Open Selected Result(s).
	Double-click on a study to open it.
	 Right-click on a study, then click Open. Note: Click on any of the Workflow header columns to sort your listed results.
	After a few moments, the Viewer opens and displays your study (as you last left it).
Remove Selected	Do one of the following to remove a selected result:
Result(s)	• Single-click to highlight the analysis you want to remove, then click Remove Selected Result(s) .
	• Right-click on the highlighted analysis, then click Remove from List .
Viewing the Results Folder in Windows Explorer	 Right-click on the highlighted analysis, then click Open in Windows Explorer. Your Analysis Results folder now appears in the Explorer window.

Browsing For Existing Analysis Results The Axiom Analysis Suite Dashboard displays/stores your results for each user profile. Note: Navigate to the Workflows sub-folder to locate other User Profile folders (that may contain more recent results).

Do the following if a study is not listed on the Dashboard:

1. Click Browse for Existing Analysis Result.

A Select Analysis Result File window appears.

2. Click on a recent analysis, then click Select Folder.

After a few moments, your analysis opens as you last left it.

3. After your analysis has successfully completed. click Open (Figure 34). Alternatively, click to highlight the completed analysis, then click Open Selected Result(s).

Figu	Figure 34 Dashboard window/tab									
New Ar	New Analysis Dashboard Preferences									
Workt	Workflows									
Batch	n Name	Date Created	Workfl	Array Type	Status	Elapsed	Status Message	Warning	Action	
Te	est_3	1/5/2015 1:17:19 PM	Best Practices Workflow	Axiom_GW	Success	7 minutes		+	Open	
Te	est_2	1/5/2015 11:01:35 AM	Genotyping	Axiom_GW	Success	2 minutes			Open	

The Axiom Analysis Suite Viewer appears.

For instructions on how to use the Viewer, continue to Chapter 3.
Browsing for Existing Suitcases

In AxAS v1.0, a suitcase file was auto-generated during an analysis and included all the necessary files needed to view your analysis in the Viewer.

For AxAs v1.1 and newer, the suitcase has been replaced with a higher performing batch name folder. Your suitcase file must be converted to this new folder format before it can be opened in the newly installed Viewer.

Do the following to convert your suitcase file to a batch name folder:

1. Click Browse for Existing Suitcase.

A Select Analysis Result File window appears.

2. Click to highlight a suitcase file, then click **Open**.

An **Axiom Analysis Suite Suitcase Conversion** message window appears. (Figure 35)

Figure 35 Convert suitcase file to batch name folder message
Axiom Analysis Suite Suitcase Conversion
A v1.0 Suitcase file is detected. To use the Suitcase, it must be first converted to a v1.1 Batch folder layout. Select OK to extract to a v1.1 Batch folder. Select Cancel to exit.
Delete suitcase file after successful conversion.
OK Cancel

- 3. If you want to retain your v1.0 suitcase file for archiving purposes, leave the **Delete suitcase file after successful conversion** check box unchecked. Click on this check box if you want your suitcase file to be auto-deleted after it is converted.
- 4. Click OK.

Allow a few moments for your suitcase file to convert to the v1.1 batch name folder format.

The Axiom Analysis Suite Viewer appears.

For instructions on how to use the Viewer, continue to Chapter 3.

 Opening an Analysis from Windows Explorer, instead of through the application's Dashboard widow tab. To do this:
 1. Right-click on an analysis batch folder and select Open in Axiom Analysis Suite Viewer. A Select Profile window appears.
 2. At the Select Profile window, enter a new or select an existing profile name. If your selected folder is a valid analysis folder, the application's Viewer opens as it normally would.



The Viewer: Summary Window and Sample Table

After setting up and successfully running an analysis, as described in Chapter 2, the Axiom Analysis Suite Viewer opens. (Figure 36)

Figure 36 Main Viewer window	V										
Summary Sample Table SNP Summary Table E	tternal Tools		✓BP:	D 0	BP: QC	🛛 SP: QC_	CR vs D 🛙	PV: D 🛙	PV: QC (Cluster P	
Export to File			Scale	Settin	gs						•
<u>Analysis Summary</u> Batch Name: Test_21 Array Package Name: Axiom_GW_Hu_SNP.r6 Array Type Name: Axiom_GW_Hu_SNP 		*					C by affyme	etrix-plate-b	earcode		
Array Display Name: Axiom Genome-Wide CEU 1 Workflow Type: Best Practices Workflow Date Created: 7/13/2015 3:08:20 PM	Array r6						I				
Sample Summary				0.75							
 Number of input samples, 221 Samples passing DQC: 205 out of 221 Samples passing DQC and QC CR: 194 out of 221 Samples passing DQC, QC CR and Plate QC: 124 out of samples did not pass: 97 Number of input samples without QC information: 0 Number of Samples Genotyped: 124 Average QC CR for the passing samples: 99.205 Gender Calls Counts: female=108 male=95 unknown 	at of 221 (56.109%) =2	E	рóс	0.5							
Plate Barcode Result Number of Nur files in a batch f d	mber of Number of Rumber of Files Samples of ailing failing QC that sish QC Call rate passed	Percent / passing c amples		0.35							
5500944096756081910675 PASSED 5 5500944099025082810502 PASSED 7	0 0 5 0 0 7	100 <u>\$</u> 100		0.25							
5500944099030090110310 PASSED 24 5500944105292111210211 FAILED 96	0 1 23 9 16 10 70 7	95.833 · 2.917 ·									
5500944105295011411127 PASSED 81 5500944105295011411131 PASSED 6	0 0 81 0 0 6	100 9 100 9									
5300944105295011411135 PASSED 2	0 0 2	100 9									
SNP Metrics Summary Number of SNPs: 587352				0	81910675	82810502	90110310	11210211	11411127	11411131	11411135
ConversionType Count Percentage PolyHighResolution 460121 78.338 NoMinorHom 94176 16.034 MonoHighResolution 12063 2.054 Other 11189 1.905		*			55009440967560	55009440990250	55009440990300	55009441052921	55009441052950	55009441052950	55009441052950

Viewing Options

As shown in Figure 36 on page 38, the Viewer (by default) displays a **side-by-side** split-screen configuration.

To change side by side split-screen to a top and bottom configuration:

1. Click the Horizontal Split icon. (Figure 37)

ummary	Sample Table SNP S	Summary Table External	Fools			
xport to File	e					1
<u>Analys</u>	is Summary					1
• Ba • An	ttch Name: Test_21 ray Package Name: Axion	n_GW_Hu_SNP.r6				
• An • An	ray Type Name: Axiom_G ray Display Name: Axiom	W_Hu_SNP Genome-Wide CEU 1 Array re	5			
• Wo • Da	orkilow Type: Best Practice ate Created: 7/13/2015 3:0	es Workflow 8:20 PM				
Sample	Summary					
• Nu	mber of input samples: 221	t of 221				
 Sat Sat 	mples passing DQC and QC mples passing DQC and QC mples passing DOC. OC CF	CR: 194 out of 221 and Plate OC: 124 out of 221	(56,109%)			
	mples did not pass: 97	· · · · · · · · · · · · · · · · · · ·	(
 Sat Nu 	umber of input samples with	out QC information: 0				
• Sa • Nu • Nu	unber of input samples with unber of Samples Genotype wrane OC CR for the passin	out QC information: 0 d: 124 n commise: 00 205				,
• Sat • Nu • Nu • Δτι • Δτι	amber of input samples with mber of Samples Genotype erage OC CR for the passin BP: QC_CR ⊠ SP: QC	out QC information: 0 d: 124 σ camplec: 00 205 .CR vs DQC Σ PV: DQC Σ	PV: QC_CR S Cluster Pl	ot		•
• Sat • Nu • Nu • Au • Au	Imber of input samples with Imber of Samples Genotype arane OC CR for the passin BP: QC_CR SP: QC B	out QC information: 0 d: 124 a complee: 00 705 CR vs DQC 20 PV: DQC 20	PV: QC_CR 3 Cluster Pi	ot		Þ
 Satistical Satistical Structure Nu Nu Avv P: DQC Satistical Structure Satistical Structure	Innber of input samples with imber of Samples Genotype errane OC CR for the passin B PP: QC CR I SP: QC 3	cR vs DQC E PV: DQC E	PV: QC_CR S Cluster PI DQC by	at affymetrix-plate-barcode		,
 Satisfield Nu Nu Aux P: DQC [8] ale Settings 1 0.75	Imber of input samples with imber of Samples Genotype erane OC CR for the passin B BP: QC CR II SP: QC 3	ou QC information: 0 d: 124 a cammler: 00 705 CR vs DQC S PV: DQC S	PV: QC.CR 🖾 Cluster Pl DQC by	affymetrix-plate-barcode		
P: DQC	Imber of Imput samples with mber of Samples Genotype erane OC CR for the nassin BP: QC_CR 0 SP: QC 8	out QC information: 0 d: 124 a cammler: 00 705 CR vs DQC © PV: DQC ©	PV: QC CR © Cluster Pl DQC b	at	 	,
• Sat • Nu • Nu • Nu • Au	Imber of Imput samples with mber of Samples Genotype erane OC CR for the nassin BP: QC_CR 00 SP: QC 3	out QC Information: 0 d 124 e cannela: 00 7NS CR vs DQC 2 PV: DQC 2	PV: QC CR © Cluster PI	at]		
Sat Nu Nu Nu Nu Avv	Imber of input samples with mber of Samples Genotype arane OC CB for the rossin B BP: QC CR D SP: QC 3	out QC Information: 0 d: 124 e envirolat: 00 7NS CR vs DQC 8 PV: DQC 8	PV: QC CR © Cluster Pl DQC by	v affymetrix-plate-barcode		
Sat Nu Nu Nu Nu O	mber of input samples with mber of Samples Genotype strand Of CB for the recein BB: QC CR D SP: QC 3	ou QC Information: 0 d: 124 e envirole: 00 7NS CR vs DQC 8 PV: DQC 8	PV: QC CR © Cluster Pl DQC by	at vaffymetrix-plate-barcode	- R	
Sat Nu Nu Nu Nu O	Amber of input samples with mber of Samples Genotype arane Of CB for the assent BB: QC CR D SP: QC 3	ou QC Information: 0 4:124 e cannola: 00 705 CR vs DQC S PV: DQC S 	PV: QC CR © Cluster Pl DQC b:	at affymetrix-plate-barcode		,
Sat Nu Nu Nu Nu O	Imber of input samples with mber of Samples Genotype arane Of CB for the resent BB: QC CR D SP: QC 3	CR vs DQC 8 PV: DQC 8	PV: QC CR © Cluster Pl DQC b:	at y affymetrix-plate-barcode	 IE TITALISE CO	,

To disable the split-screen:

1. Click the **Disable Split-Screen** icon. (Figure 38)

Figure 38	Disable Split-
Screen ico	n

The split-screen becomes 1 window. (Figure 39)

Split-Screen Options

	Full	wi	ndo	w ۷	viev	v e>	kamp	le				
Sample Table	SNP Su	immary	Table	Externa	al Tools	BP: D	ାପ୍ର 🖾 🛛 BP: ପ୍	QC_CR ା SP: QC_CR vs DQC @	PV: DQC 🖾 🖡	V: QC_CR 🖾 Clı	uster Plot	
File												
sis Summary												
atch Name: Test_2 rray Package Nam rray Type Name: A rray Display Name 'orkflow Type: Bes ate Created: 7/13/	1 ae: Axiom_GW a: Axiom C t Practices 2015 3:08:	GW_Hu '_Hu_SN Genome-' Workflo 20 PM	SNP.r6 P Wide CEI w	U 1 Array	r6							
<u>Summary</u>												
Samples passing DQC Samples passing DQC	and QC (CR: 194	out of 22	1 4 out of 2	21 (56.1	09%)						
Samples did not pass: Number of input sam Number of Samples G Average QC CR for ti Gender Calls Counts:	2, QC CR : 97 ples witho Genotyped: he passing female=10	ut QC in 124 samples 8 male= Number of files	formation 99.205 95 unkno	n: 0 own=2	Number	Percent	Average call rate					
les did not pass: ber of input sam ber of Samples O uge QC CR for ti er Calls Counts: se Barcode	C, QC CK : 97 ples witho Genotyped: he passing female=10 Result	ut QC in 124 samples 8 male= Number of files in a batch	formation 99.205 95 unknot of files failing dish QC	n: 0 own=2 Number of files failing QC Call rate	Number of samples that passed	Percent of passing samples	Average call rate for passing samples					
did not pass: of input sam of Samples Q QC CR for ti calls Counts: rcode 6603250778	C, QC CK : 97 ples witho Genotyped: he passing female=10 Result	ut QC in 124 samples 8 male= Number of files in a batch 96	formation 99.205 95 unkno of files failing dish QC	n: 0 own=2 Number of files failing QC Call rate 0	Number of samples that passed 96	Percent of passing samples 100	Average call rate for passing samples 99.977					
did not pass: of input sam of Samples Q QC CR for the alls Counts: recode 5603250778 5820237750	C, QC CK : 97 ples witho Genotyped: he passing female=10 Result PASSED PASSED	nd Plate ut QC in 124 samples 8 male= of files in a batch 96 96	formation 99.205 95 unkno of files failing dish QC 0	n: 0 wn=2 Number of files failing QC Call rate 0 0	Number of samples that passed 96 96	Percent of passing samples 100 100	Average call rate for passing samples 99.977 99.976					
kid not pass: f input sam f Samples O (C CR for ti alls Counts: code 6603250778 820237750 1142456642	C, QC CK = 97 97 ples witho Genotyped: he passing female=10 Result PASSED PASSED PASSED	Number of files in a batch 96 96 96	formation 99.205 95 unkno of files failing dish QC 0 0	n: 0 wn=2 Number of files failing QC Call rate 0 0 0	Number of samples that passed 96 96 96 96	Percent of passing samples 100 100	Average call rate for passing samples 99.977 99.976 99.976					
did not pass: of input sam of Samples G QC CR for th Calls Counts: 0012250778 0012250778 00142456642 0215303673	C, QC CK : 97 ples witho fenotyped: he passing female=10 PASSED PASSED PASSED PASSED PASSED	And Plate ut QC in: 124 samples: 8 male= in a batch 96 96 96 96 96	formation 99.205 95 unkno of files failing dish QC 0 0 0	n: 0 wn=2 Number of files failing QC Call rate 0 0 0 0 0 0	Number of samples that passed 96 96 96 96	Percent of passing samples 100 100 100	Average for passing samples 99.977 99.976 99.976 99.976					
tid not pass: of input samples C QC CR for ti alls Counts: code 5603250778 5620237750 1142456642 3221580667 1063333673 1184043350	C, QC CK : 97 ples witho Genotyped: he passing female=10 PASSED PASSED PASSED PASSED PASSED PASSED	nd Plate ut QC in: 124 samples: 8 male= of files in a batch 96 96 96 96 96 96	formation 99.205 95 unkno of files failing dish QC 0 0 0 0 0 0 0	n: 0 wn=2 Number of files failing QC Call rate 0 0 0 0 0 0 0 0 0	Number of samples that 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100	Average call rate for passing pamples 99.977 99.976 99.977 99.976					
did not pass: of input sam of Samples C QC CR for ti Calls Counts: arcode 16603250778 306820237750 30142456642 56221580667 37063333673 4184043350	C QC CK : 97 ples witho Genotyped: he passing female=10 PASSED PASSED PASSED PASSED PASSED PASSED PASSED	nd Plate ut QC in: 124 samples: 8 male= 8 male= in a batch 96 96 96 96 96 96 96	formation 99.205 95 unkno 95 unkno 0 0 0 0 0 0 0 0 0 0 0 0 0	n: 0 wn=2 Number of files failing QC Call rate 0 0 0 0 0 0 0 0 0 0 0 0 0	Number of samples that passed 96 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100 100	Average call rate for passing samples 99.977 99.976 99.976 99.977 99.977 99.977					
idi not pass: f input sam f Samples C C CR for ti ills Counts: code 603250778 820237750 1142456642 221580667 05333673 184043330 16404330	C, QC CK : 97 ples witho Genotyped: he passing female=10 PASSED PASSED PASSED PASSED PASSED PASSED PASSED PASSED	Number of files in a batch 96 96 96 96 96 96 96 96 96	formation 99.205 95 unkno of files failing dish QC 0 0 0 0 0 0	n: 0 wn=2 Number of files failing QC call rate 0 0 0 0 0 0 0 0 0 0 0 0 0	Number of samples that passed 96 96 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100 100	Average call rate for passing passing 99.977 99.976 99.976 99.977 99.977 99.977 99.977					
ples did not pass: ber of limput sam ber of Samples C age QC CR for th der Calls Counts: te Barcode 27446603250778 43106620237780 1976014256642 11605221500657 116256642 11656563820115 118545664221451 11854564221451 11854564221451 11854564221451 11854564221451 1185456422461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 11855642461 118556426642461 118556426642461 118556426642461 118556426642461 118556426642461 118556426642664 11855642664 11855642664 11855664 11855664 11855664 11855666 118556666 11855666666 118556666666666666666666666666666666666	, QC CK : 97 ples witho Genotyped the passing female=10 PASSED PASSED PASSED PASSED PASSED PASSED PASSED	Number of files in a batch 96 96 96 96 96 96 96 96 96 96 96 96 96	formation 99.205 95 unkno of files failing dish QC 0 0 0 0 0 0 0 0 0	n: 0 wwn=2 Number of files failing QC Call rate 0 0 0 0 0 0 0 0 0 0 0 0 0	Number of samples that passed 96 96 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100 100 100	Average call rate for passing 3amples 99.977 99.976 99.976 99.977 99.977 99.977 99.977 99.977					
oles di ano pass bes of inpui sam bes of Samples G ange QC RE for di ler Calls Counts: a Barcode 1746602210778 1306020217706 1306020217060 1306020217060 1306020217060 1306020217060 1306020217060 1306020217060 1306020217060 1306020210 13060202178 1306020218 13060202178 13060202178 1306020218 1306020 13060000	7352	and Plate 124 sampless 8 male= Number of files 96 96 96 96 96 96 96 96 96	formation 99.205 95 unkno of files failing dish QC 0 0 0 0 0 0 0	n: 0 wm=2 Number of files failing QC Call rate 0 0 0 0 0 0 0 0 0 0 0 0 0	Number of samples passed 96 96 96 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100 100	Average call rate for passing anaples 99.376 99.376 99.376 99.377 99.377 99.377 99.377 99.577					
ples dd nor pass ber of ingut sam ber of Samples G ber of Samples G reads and the samples of the samples of reads of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the samples of the reads of the samples of the samples of the samples of the samples of the reads of the samples of the	7352 Count	and Plate tat QC im 124 sampless & male= 8 male= 8 male= 96 96 96 96 96 96 96 96 96 96 96	rcentage	n: 0 wn=2 Number of files failing QC Call rate 0 0 0 0 0 0 0 0 0 0 0 0 0	Number of 96 96 96 96 96 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100 100 100	Average call rate to for passing ramples 99.377 99.376 99.377 99.377 99.377 99.377 99.377 99.377 99.377					
mples did not pass: umber of input sam umber of Samples O Verage QC 200 K for th ender Calls Counts: Valte Barcode 02744660325776 23310620227750 23310620227750 2331062022750 2331062022750 2331062022750 2331062022750 2331062022750 2331062022750 2331062022750 2331062022750 233106202750 233016202750 233106202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 233016202750 2330162022750 2330162022750 2330162022750 2330162022750 233016202750 23301000000000000000000000000000000000	7352 Count C	and Plate tt QC im 124 8 samples 8 male= 96 96 96 96 96 96 96 96 96 96	contraction contracti	n: 0 wwn=2 Number of files QC Call o 0 0 0 0 0 0 0 0 0 0 0 0 0	Number of passed 96 96 96 96 96 96 96 96 96	Percent passing samples 100 100 100 100 100 100 100	Average call rate for passing and the second second and the second second second and the second second second and the second second second second and the second second second second and the second second second second second and the second second second second second second and the second second second second second second second and the second second second second second second second and the second secon					
amples did not pass: umber of nyu sam umber of Samples CR for th everage QC CR for th everage QC CR for th everage QC CR for the output of the second second second second output of the second second second second second second second second secon	7352	and Plate triangle of the second se	Contraction contr	n: 0 wwn=2 wwn	Number of samples that paced 96 96 96 96 96 96 96	Percent of passing samples 100 100 100 100 100 100 100	Average call rate program rample 99.577 99.576 99.576 99.577 99.577 99.577 99.577 99.577 99.577 99.577					

2. Click on any window tab (Figure 39) to view it in full window mode.

To return to the default side by side split-screen configuration:

1. Click the Vertical Split icon. (Figure 40)



Figure 41 Split Vertical View example													
Summary Sample Table SNP Summary Table External Tools	⁶ BP: D G BP: QC G SP: QC.CR vs D G PV: D G PV: QC G Cluster P U □ Coate Stringer												
Export to File	E DOC hu afferentelle slote horsende												
Analysis Summary Batch Name: Test_21 Array Package Name: Axiom_GW_Hu_SNP16 Array Package Name: Axiom Genome-Wide CEU 1 Array 16 Array Display Name: Axiom Genome-Wide CEU 1 Array 16 Workflow Type: Bate Packiest Workflow Date Created: 7/13/2015 308/20 PM Samples Samples nameles: 221 Samples passing DQC 205 out of 221 Samples passing DQC 205 out of 221 Samples passing DQC 205 CR And Pate QC: 124 out of 221 (56.109%) Samples date of pass samples without QC information: 0 Named or fingut samples without QC information: 0 Named or fingut samples without QC information: 0 Named C fingutes passing DQC 205 Gender Calls County Created and Page 205 Samples date of pass samples: 98.205 Gender Calls County Created Page 205 Samples Calls County Created Page 205													
Plate Barcole Result Number of files in a batch Number of files in addition Number of files failing control Number of files failing control Number of files files control Number of files control Number of control Number of contro Numbero	0.25												
Stot0944.0523901.1411135 PASSED 2 0 2 100 5 View details SNP Metrics Summary	C.001019995.49801460055 11211114 TUGESCOIL460055 2050182392.516634460055 2050182392.516634460055												

Changing a Tab Window to a Full Screen Windows

To toggle a tab window to full screen:

- 1. Locate the tab you want to make full screen.
- 2. Click on a tab's white triangle graphic. (Figure 42)

Figure 42 Tab to window toggle icon
Sample Table

The window tab is now a window.

3. Double-click anywhere along the top of the window to change it to full screen.

To toggle a full screen window to its default tab window:

- 1. Double-click anywhere along the top of the window.
- 2. Click the **X** button (top right) to close the window.

The window returns to its default tab window and position.

Adjusting the Window Size

To change the size of a window pane:

1. Click, hold, then drag the edge of the window pane (Figure 43) to resize it.

Figure 43 Split Vertical View example





Summary Window/Tab

The Summary window/tab (Figure 44) displays a summary snapshot of your analysis, including detailed threshold values, and tables based on your analysis.

Data Analysis Summary Note: Each workflow type reports different information within the Analysis Summary window. Figure 44 is an example of a Best Practices workflow.

Figure 44 Summ	ary wi	ndow t	ab							
Export to File							 Export to File: Click this button to export the Summary report as a PDF file. 			
Analysis Summary Batch Name: Test_21 Array Package Name: A Array Type Name: Aion Array Display Name: Aion Workflow Type: Best Pra Date Created: 7/13/2015	xiom_GW_ m_GW_Hu_ xiom Genon actices Wor 5 3:08:20 PN	_Hu_SNP.r6 _SNP ne-Wide CEV kflow M	Analysis Summary: Contains informa- tion about the array type, the workflow run and the date processed							
Sample Summary Number of input samples: Samples passing DQC: 20 Samples passing DQC and Samples passing DQC, Q Samples did not pass: 97 Number of input samples Number of Samples Geno Average QC CR for the p Gender Calls Counts: fem	221 5 out of 22 d QC CR: 1 C CR and P without QC typed: 124 assing samp ale=108 ma	1 94 out of 22 late QC: 124 c information ples: 99.205 le=95 unkno	1 4 out of 22 n: 0 wn=2	1 (56.109%	5)		Sample Summary: Breaks down the sample QC for your analysis run and displays the number that pass each of your QC Thresholds. In addition, it provides the average QC Call Rate (CR) and breakdown of the genders found within your batch of samples.			
Plate OC Summary							Plate QC Summary: Contains sample QC			
Plate Barcode 55009440996756081910675 5500944099025082810502 5500944099030090110310 5500944105292111210211 5500944105295011411127 5500944105295011411125 Views dotails	Result PASSED PASSED PASSED FAILED PASSED PASSED PASSED	Number of files in a batch 5 7 24 96 81 6 2	Number of files failing dish QC 0 0 16 0 0 0 0	Number of files failing QC Call rate 0 0 1 1 0 0 0 0 0 0	Number of samples that passed 5 7 23 7 0 81 6 2	Percent of passing samples 100 100 95.833 72.917 100 100 100	information for each plate including the number samples failing DQC, QC Call Rate, the Percent of passing samples. and the average Call Rate for your passing samples.			
SNP Metrics Summary							SNP Metrics Summary: This section con-			
Number of SNPs: 587352 ConversionType PolyHighResolution MonorHom MonorHom Other CallRateBelowThreshold Hemizygous	Count 460121 94176 12063 111189 7568 1971	Percentage 78.338 16.034 2.054 1.905 1.288 0.336					the SNPs in the analysis by PS_Classification. For more information on these categories <i>see "Regenerate SNP Metrics" on page 82</i> .			
otv Sample OC Thresholds • DQC: ≥ 0.81 • QC call_rate: ≥ 97 • Percent of passing sample • Average call rate for passi	264 es: ≥ 88 ng samples:	0.045 =≥ 98.5					Sample QC Thresholds: Displays the Sample QC Thresholds used for your analysis run and their associated SNP QC Metrics.			
SNP OC Thresholds • species-type: Diploid • cr-cutoff ≥ 94 • fid-cutoff: ≥ 3.6 • het-so-cutoff: ≥ -0.1							SNP QC Thresholds: Displays the Thresholds used for your analysis run and their associated SNP QC Metrics.			

Viewing the Plate Barcode Table Details

- 1. In the Summary window tab, click View Details . (Figure 44)
 - A window opens and displays a text file version of your Sample QC information (by plate). (Figure 45)

Figure 45 Notepad window		
Plate QC Details		<u> </u>
File Edit Format View Help Thresholds used by Different QC steps for passi	ng samples:	
Dish QC threshold >= 0.81 QC Call rate threshold >= 97 Plate QC PercentSamplesPassed >= 88% Plate QC AverageCallRate >= 98.5		
######################################	########### 6081910675	
Result: PASSED		
Number of files in a batch 5 plate_num_of_files_failed_dish_qc 0 plate_num_of_files_failed_qc_call_rate Percent of passing samples 100% Average call rate for passing samples	0 99.144	
######################################	########## 5082810502	
Result: PASSED		
Number of files in a batch 7 plate_num_of_files_failed_dish_gc 0 plate_num_of_files_failed_gc_call_rate Percent of passing samples 100% Average call rate for passing samples	0 98.93	
######################################	<i>#########</i> 0090110310	
Result: PASSED		
Number of files in a batch 24 plate_num_of_files_failed_dish_gc 0 plate_num_of_files_failed_gc_call_rate Percent of passing samples 95.833% Average call path for passing complex	1	
Following files failed QC Call rate criteria NA19160_AxiomGWHUSNP1_20100906_InputMassTitr-20	0ng_Kit-ODevLot-1a_B04.CEL QC Call rate value 95.822 < QC Call rate threshold 97	
×	F	•

Sample Table

Note: Depending on the Threshold values you set (prior to running your analysis), color-coded Pass or Fail cells may appear in the table, as shown in Figure 46.

Summary Sample Table	SNP Su	mmary	Table				
Scatter Plot Box Plot Plate Vi	ew Concord	ance R	eanalyze S	elected Sa	mples In	port Sam	ple Attributes 🔻
Revert Calls 🔻							
Annhy View View Star	haw/Hida Cal	lumana 1	Evenent	Tilta			
Apply view + [Save view] Si	now/ Hide Col	lumns	Export	• Fille	5 -		
			00		00		00
Sample Filename	Pass/Fail	DQC	call rate	call_rate	het rate	het_rate	computed gen
			con_race				compared_gen
2 (1).CEL	Fail	0.985	98.406		25.887		female
2 (2).CEL	Fail	0.973	97.225		25.526		male
2 (3).CEL	Fail	0.974	99.685		25.284		female
2 (4).CEL	Fail	0.984	93.444		24.464		male
2 (5).CEL	Fail	0.972	96.704		25.046		male
NA07034_a550094-4096756-0	Pass	0.977	99.103	98.727	27.79	27.144	male
NA10857_a550094-4096756-0	Pass	0.985	98.891	98.604	27.976	27.215	male
VA12057_a550094-4096756-0	Pass	0.992	100.598	100.325	27.79	27.649	female
VA12752_a550094-4096756-0	Pass	0.99	98.349	98.046	27.383	26.989	male
VA12814_a550094-4096756-0	Pass	0.947	98.525	98.074	27.852	27.175	male
VA18504_a550094-4099025-0	Pass	0.983	98.086	97.719	25.366	24.717	male
VA18517_a550094-4099025-0	Pass	0.983	101.79	101.44	26.362	25.679	female
VA18863_a550094-4099025-0	Pass	0.966	98.221	97.755	26.202	25.478	male
VA19101_a550094-4099025-0	Pass	0.951	98.468	98.012	26.078	25.44	male
VA19130_a550094-4099025-0	Pass	0.983	98.7	98.208	25.248	25.075	male
VA19238 a550094-4099025-0	Pass	0.948	101.016	100.599	26.97	26.397	female
	Pass	0.937	101.191	100.792	26.511	25.981	female
A19098 AxiomGWHuSNP1	Pass	0.929	98.711	98.308	25.64	25.207	male
VA19099 AxiomGWHuSNP1	Pass	0.951	101.398	101.077	26.124	25.746	female
NA19116 AxiomGWHuSNP1	Pass	0.942	101.955	101.42	26.073	25.538	female
VA19119 AxiomGWHuSNP1	Pass	0.974	99.283	98,949	25,294	24.917	male
VA19127 AxiomGWHuSNP1	Pass	0.962	101.945	101.581	26.455	25,901	female
VA19128 AxiomGWHuSNP1	Pass	0.959	99.123	98.771	25.825	25.035	male
VA19130 AxiomGWHuSNP1	Pass	0.938	98,757	98,391	25.459	25,192	male
VA19131_AxiomGWHuSNP1	Pass	0.945	101.857	101.232	26.975	26.044	female
VA19140 AxiomGWHuSNP1	Pass	0.944	101.491	101.106	26.547	25.851	female
VA19141 AxiomGWHuSNP1	Pass	0.96	98,963	98.688	25.036	24.87	male
VA19143 AviomGWHuSNP1	Pacc	0.92	101 573	100 991	26 795	26 118	female
VA19144 AviomGWHuSNP1	Pass	0.972	99.17	98 91 3	25 903	24 898	male
VA19152 AxiomGWHuSNP1	Pass	0.948	101 934	101 545	26 228	25 799	female
VA19152_AxiomGWHuSNP1	Dace	0.940	08 001	98.602	25.062	2/ 882	male
A19159_AxiomGWHuSNP1	Dace	0.000	101 /12	101 020	25.002	25.952	female
VA19159_AxiomGWHuSNP1	Fail	0.929	06 202	101.029	20.377	20.002	male
VA10102 AviomGWHuSNP1	Dace	0.020	08 592	00 27	25.026	25.067	male
VA15152_AXIONGWHUSNP1	Dace	0.959	101 500	101 204	25.020	25.007	female
VA19195_AXIOMGWHUSNP1	Pass	0.933	101.599	101.290	20.009	25.705	female
VA19200_AXIOMGWHUSINP1	Pass	0.944	101.383	101.100	20.119	25./14	remaie
NA19207_AXIOMGWHUSNP1	Pass	0.934	98.41/	98.143	23,830	25.05	male
VA19222_AXIOMGWHUSNP1	Pass	0.8/4	100.2/3	99.811	27.177	20.541	Temale
VA19223_AxiomGWHuSNP1	Pass	0.935	98./47	98.458	25.851	24.967	male
NA19238_AxiomGWHuSNP1	Pass	0.944	101.578	101.235	26.578	26.008	temale

Importing Sample Attributes

To import sample attributes into your Sample Table:

- 1. Click the Import Sample Attributes drop-down.
- 2. Click to select either **Import from ARR Files** or **Import from CSV/Tab-Delimited Text File**.

An Explorer window appears.

IMPORTANT! Your text-based CEL file must start with the header *Sample Filename* and include the full CEL file name, as shown in Figure 47.

Figure 47 Tab-delimited text CEL file example shown in Excel

	А	В	С
1	Sample Filename	String Name	Number Name
2	NA18961_AxiomGWHuSNP1_20101007_TrainingMix_Rtstab-DL1a-Omo_E09.CEL	A1	1
3	NA12760_AxiomGWHuSNP1_20101007_TrainingMix_Rtstab-DL1a-Omo_B06.CEL	A2	1
4	NA19160_AxiomGWHuSNP1_20100906_InputMassTitr-200ng_Kit-ODevLot-1a_B04.CEL	A3	1
5	NA18960_AxiomGWHuSNP1_20101007_TrainingMix_Rtstab-DL1a-Omo_E08.CEL	A4	1
6	NA12814_AxiomGWHuSNP1_20101007_TrainingMix_Rtstab-DL1a-Omo_B09.CEL	A5	1
7	NA19210_AxiomGWHuSNP1_20101007_TrainingMix_Rtstab-DL1a-Omo_F12.CEL	A6	1

3. Navigate to the applicable file location, then click **Open**.

Column Headers The default Sample Table column view is as shown. (Figure 48)

Figure 48 Defau	Figure 48 Default Sample Table Columns														
Sample Filename	Pass/Fail	DQC	QC call_rate	call_rate	QC het_rate	het_rate	QC computed_gender	affymetrix- plate- barcode	affymetrix- plate-peg- wellposition	Average call rate for passi	Percent of passing				

To show or hide table columns:

- 1. Click the **Show/Hide Columns** drop-down menu.
- 2. Click each available column name's check box to show it or remove it from the table. See "Annotations and Columns" on page 159 for their definitions.
- 3. Click outside the Show/Hide Columns drop-down menu to close it.

To save your customized Sample Table column view:

1. Click Save View.

The Save Current View window appears. (Figure 49)

Figure 49 View	Save	Current
Save Current Vie	w	×
Custom View N	lame:	
I		•
	ОК	Cancel

Enter a name for your custom table view, then click OK.
 Your newly saved name is now added to the Apply View drop-down menu.

To show ALL available columns within the Sample Table:

1. Click the Apply View drop-down menu, then select All Columns View.

Rearranging Columns

- 1. Click on a column you want to move.
- 2. Drag it (left or right) to its new location.
- 3. Release the mouse button.

The column is now in its new position.

Sorting Columns

 Select a column, then right-click on it. A right-click menu appears. (Figure 50)

Figure 50 Menu	Right-	click Co	olumn
Sample File	name	affymetrix- plate-	affymetrix- plate-peg-
2550465-41	Sort By Asce	nding	
a550465-41	Sort By Desc	ending	
a550465-41 🍸	Filter		
a550465-41 a550465-41	Copy Colum	ın	
a550465-41	Clear Curren	t Column Filt	er
a550465-41 🏾 🍅			-
a550465-41	Hide Colum	n	

2. Click to select either Sort By Ascending (A-Z) or Sort By Descending (Z-A).

Single-Click Sorting Method

1. Single-click on a column header to sort its data in an ascending order. Single-click on the same column header to sort its data in a descending order

Hiding the Column

- Select the column you want to hide from the table, then right-click on it. A right-click menu appears. (Figure 50)
- 2. Click the Hide Column check box to remove it from the table.

Filtering Column Data

Note: All Sample Table columns are filterable.

Adding Filters (Method 1)

1. Select a column, then right-click on it. The following window appears: (Figure 51)



2. Click Filter.

Text-based Columns

If the column you want to filter contains text-based data, the Contains drop-down menu appears. (Figure 52)



To apply a filter to a text-based column:

1. Click the **Contains** drop-down menu to select a filtering property. (Figure 53)

Figure 53	Drop-do	own Menu
Sample Filename		×
Contains Contains Equals Not Contai Not Equal	n	
	ОК	Cancel

- 2. Click inside the text entry box to enter a value. (Figure 53)
- 3. OPTIONAL: Click 🧝 to add additional filters.

Figure 54 Or or And Relationship Logic	
Sample Filename	×
Contains Contai	
Relationship: Or And 	
OK Cancel	

- 4. Click the **Or** or **And** radio button to choose **Or** or **AND** relationship logic. (Figure 54)
- 5. Repeat steps 1-4 as needed.
- 6. To remove a filter(s), click 🔽.

Numeric Data Columns

If the column you want to filter contains numeric data, a symbol drop-down menu appears. (Figure 55)

Figure 55	Filter Pr	roperties
DQC		×
<		3
	ОК	Cancel

To apply a filter to a value-based column:

1. Click the **Symbol Value** drop-down menu to select the filtering symbol you want. (Figure 56)

Figure 56	Drop-do	wn Menu
DQC	• 	×
	ОК	Cancel

2. Click inside the text entry box

to enter the value(s). (Figure 56)



3. OPTIONAL: Click 😨 to add filter(s).



- 4. Click the **Or** or **And** radio button to choose **Or** or **AND** relationship logic. (Figure 57)
- 5. If needed, repeat steps 1-4.
- 6. Click OK.

To remove a filter(s), click 🔞.

Showing Filtered Data Only

• Click the **Show Filtered Only** check box Show Filtered Only to show only the data that passes the filters.

Uncheck this box to show all data, including data that did not pass your filter criteria setting(s). In this mode, data that passes the filter appears in light gray, as shown in Figure 58 on page 51.

Sample Filename	Pass/Fail	DQC	QC call_rate	call_r 🍸	QC het_rate	het_r 🍸	QC computed_gen	ł
NA11840_AxiomGWHuSNP1	Pass	0.871	100.371	99.964	29.57	29.226	female	1
NA12875_AxiomGWHuSNP1	Pass	0.833	99.479	98.943	30.292	29.594	female	
NA18577_AxiomGWHuSNP1	Pass	0.847	99.381	99.02	26.831	26.318	female	
NA18593_AxiomGWHuSNP1	Pass	0.902	99.665	99.318	26.403	25.716	female	
NA18942_AxiomGWHuSNP1	Pass	0.869	100.289	99.987	26.599	25.908	female	
NA18949_AxiomGWHuSNP1	Pass	0.925	99.175	98.832	25.913	24.988	female	
NA19099_AxiomGWHuSNP1	Pass	0.851	100.397	99.973	27.079	26.676	female	
NA19222_AxiomGWHuSNP1	Pass	0.874	100.273	99.811	27.177	26.541	female	
2877_AxiomGWHuSNP1_2010		0.982	102.424	102.115	26.847	25.092	female	
2878_AxiomGWHuSNP1_2010		0.972	102.357	101.935	25.49	24.78	female	
2881_AxiomGWHuSNP1_2010		0.951	101.96	101.588	28.585	27.786	female	
2884_AxiomGWHuSNP1_2010		0.955	102.089	101.636	26.083	25.184	female	
2885_AxiomGWHuSNP1_2010		0.969	102.213	101.81	27.749	27.9	female	
2888_AxiomGWHuSNP1_2010		0.945	101.676	101.279	28.471	28.032	female	
2889_AxiomGWHuSNP1_2010		0.986	102.563	102.292	28.678	27.89	female	
2890_AxiomGWHuSNP1_2010		0.97	102.352	101.956	26.393	25.224	female	
2893_AxiomGWHuSNP1_2010		0.965	102.357	101.93	28.781	28.135	female	
2894_AxiomGWHuSNP1_2010		0.967	102.233	101.855	25.897	25.244	female	
2895_AxiomGWHuSNP1_2010		0.978	102.527	102.255	25.676	24.955	female	
2897_AxiomGWHuSNP1_2010		0.965	102.553	102.1	26.248	25.142	female	
2899_AxiomGWHuSNP1_2010		0.969	102.373	101.972	28.796	28.016	female	
2900_AxiomGWHuSNP1_2010		0.953	102.094	101.667	28.177	28.08	female	
2901_AxiomGWHuSNP1_2010		0.979	102.357	101.977	28.409	27.583	female	
2902_AxiomGWHuSNP1_2010		0.979	102.486	102.163	26.955	25.39	female	
2903_AxiomGWHuSNP1_2010		0.975	102.507	102.082	28.961	28.181	female	
2905_AxiomGWHuSNP1_2010		0.965	102.285	101.844	28.09	27.9	female	
2907_AxiomGWHuSNP1_2010		0.95	102.104	101.603	26.119	25.442	female	
2908_AxiomGWHuSNP1_2010		0.951	102.146	101.701	28.93	27.792	female	
2909_AxiomGWHuSNP1_2010		0.963	102.383	102.008	25.418	24.772	female	
2915_AxiomGWHuSNP1_2010		0.961	102.151	101.827	28.817	28.035	female	
2916_AxiomGWHuSNP1_2010		0.985	102.481	102.216	26.063	24.87	female	-
2917_AxiomGWHuSNP1_2010		0.956	102.12	101.717	28.894	28.33	female	-
2943_AxiomGWHuSNP1_2010		0.955	102.058	101.634	25.619	25.129	female	-
2944_AxiomGWHuSNP1_2010		0.966	102.393	101.848	25.774	24.926	female	
2947_AxiomGWHuSNP1_2010		0.961	102.192	101.853	29.013	28.086	female	
2948_AxiomGWHuSNP1_2010	Pass	0.955	102.316	101.84	25.634	24.856	female	-
NA11882_AxiomGWHuSNP1		0.915	101.553	101.186	28.796	27.976	female	
NA12057_a550094-4096756-0		0.992	100.598	100.325	27.79	27.649	female	
NA12239_AxiomGWHuSNP1		0.88	100.634	100.195	29.26	28.962	female	-
NA12249_AxiomGWHuSNP1		0.924	101.656	101.173	28.095	27.622	female	-

Figure 58	Sample Table window tab - Show Filter Only
unchecked	l example

Clearing an Individual Filter

 Right-click on the filtered column you want to clear. The following window appears: (Figure 59)

Figure Menu	59 Right-click Column
QC comput <u>ed aen</u>	QC QC
—	Sort By Ascending
	Sort By Descending
Y	Filter
لأكا	Copy Column
	Clear Current Column Filter
	Hide Column

2. Click **Clear Current Column Filter**. The filter is removed.

Clearing All Current Filters

• Click the Filters drop-down, then select Clear Current Filters. (Figure 60)

Figure 60	Filters Menu
Filters 🔻	
Manage Filters	
Clear Current I	Filter(s)

Adding Filters (Method 2)

Use this method if you want to change more than one of your Sample Table column filters at the same time.

1. Click the Filters drop-down menu, then click Manage Filters.

The Manage Filters window appears. (Figure 61)

Figure 6 ⁻	1 Manage Filters v	vindow	
Manage Filters			×
Ac	ld Column Filter	Clear All	
Column	Filter		
\mathbf{X}	Select a Column	ı	
		ОК	Cancel

Note: If the column you want to filter contains text-based data, the Contains dropdown menu appears. If the column you want to filter contains numeric data, a symbol drop-down menu appears.



2. Click the **Column** drop-down, then click to select the Column name you want to create a filter for. (Figure 62)

nanage riters			
Add Co	lumn Filter	Clear All	
Column	Filter		
\boxtimes	 Select a Column 		
Pass/Fail DQC QC call_rate call_rate QC het_rate <u>het_rate</u> QC computed affymetrix-plat Average call ra Percent of pas	gender e-barcode e-peg-wellposition te for passing samples sing samples	_	

3. Click the **Symbol Value** drop-down menu to select the filtering symbol you want. (Figure 63)

Figure 63 Manag	ge Filters window - Select a	l Symbol
Manage Filters	1 1 1 1 0 75 1 1	×
Add Column Filter	er Clear All	
Column	Filter	
🗙 het_rate 🔹		~~
	× ≤	
	2	
	ОК	Cancel

- 4. Click inside the text entry box to enter new value(s). (Figure 63)
- 5. OPTIONAL: If you want to add an additional filter to a column, click 😨.

6. Click the **Or** or **And** radio button to choose **Or** or **AND** relationship logic. (Figure 64)

Figure 64 Manage Filters window - OR or AN Relationship	ID
Manage Filters	×
Add Column Filter Clear All	
Column Filter	
🗙 het_rate 🔹 😵 🤜	3
> •	
Relationship: Or And	
OK 0	ancel

7. If needed, click Add Column Filter, then repeat the above steps. (Figure 65)

Figure 65 Manag Column Filter	e Fi	Iters window - Adding another
Manage Filters		×
Add Column Filter		Clear All
Column	Filter	
K het_rate ▼	7	< • •
		Relationship: 💿 Or 💿 And
🗙 Sample Filename 🔹	7	Contains -
		Contains Equals Not Contain Not Equal
		OK Cancel
		· · · · · · · · · · · · · · · · · · ·

8. Click OK.

To remove a filter(s), click 🔽.

Click Clear All to remove ALL filters in the Manage Filters window.

Copying Column Data

To copy column data to your clipboard:

 Click to select a column you want to copy to a clipboard, then right-click on it. The following window appears: (Figure 66)



2. Click Copy Column.

The column data is now ready for pasting (Ctrl v).

Setting User Colors Use this feature to more easily identify different sets between the Sample Table and Cluster Graph.

Assigning a Color to a Sample

1. Right-click on the sample you want to assign a color to.

A menu appears. (Figure 67)

Figure 67 Right-	click m	enu -	Set U	se	r C	olor		
IL (JICEL	i un	0.572	50.704			20.040		
NA07034_a550094-4096756-0	Pass	0 977	99 103	Q	8.727	27.79	27.144	male
NA10857_a550094-4096756-0	Copy Sele	ected Row	/(s)		3.604	27.976	27.215	male
NA12057_a550094-4096756-0	Copy Sele	ected Cell	(s)).325	27.79	27.649	female
NA12752_a550094-4096756-0	Cat Hear (Celer				Import File		-
NA12814_a550094-4096756-0	Set User (.0101		•	_	Import File		-
NA18504_a550094-4099025-0	Remove l	Jser Colo	r			Red		2
NA18517_a550094-4099025-0	Pass	0.983	101./9	1		Gold		ale
NA18863_a550094-4099025-0	Pass	0.966	98.221	2		Plue		2
NA19101_a550094-4099025-0	Pass	0.951	98.468	2		Diue		2
NA19130_a550094-4099025-0	Pass	0.983	98.7	9		Aquamarin	e	2
NA19238_a550094-4099025-0	Pass	0.948	101.016	10		Gray		ale
NA19238_a550094-4099025-0	Pass	0.937	101.191	10	_	LightGray	_	ale
NA19098_AxiomGWHuSNP1	Pass	0.929	98.711	9	_	Lightolay		2
NA19099_AxiomGWHuSNP1	Pass	0.951	101.398	10		Green		ale
NA19116_AxiomGWHuSNP1	Pass	0.942	101.955	1		RoyalBlue		ale
NA19119_AxiomGWHuSNP1	Pass	0.974	99.283	5		0		-
NA19127_AxiomGWHuSNP1	Pass	0.962	101.945	10		Orange		ale
NA19128_AxiomGWHuSNP1	Pass	0.959	99.123	5		LawnGreen	1	2
NA19130_AxiomGWHuSNP1	Pass	0.938	98.757	5		Magenta		
NA19131_AxiomGWHuSNP1	Pass	0.945	101.857	10		Proven		ale
NA19140_AxiomGWHuSNP1	Pass	0.944	101.491	10		Brown		ale
NA19141_AxiomGWHuSNP1	Pass	0.96	98.963	9		Yellow		2
NA19143_AxiomGWHuSNP1	Pass	0.92	101.573	10		Purple		ale
NA19144_AxiomGWHuSNP1	Pass	0.972	99.17	9		Plack		2
NA19152_AxiomGWHuSNP1	Pass	0.948	101.934	10		BIACK		ale
NIA101ED A. Sear CMULLICHIDI	D	0.050	00 001	0	0 60 1	25.062	14 001	

- 2. Mouse over Set User Color.
 - A color pallet appears.

З

3. Click on the color you want.

A **user_color** column is automatically added to your Sample Table and contains your sample's newly assigned color, as shown in Figure 68.

	Figur	e 68	Right	-click menu	- Set U	ser Colo	r			
ĺ	Summa	ry <mark>🗖 Sa</mark> r	mple Tab	le SNP Summa	ry Table	External Too	ols			
	Scatter P	lot Box Pl	ot Plate	view Concordance	Reanalyze Se	elected Sample	s Import	Sample A	ttributes 🔻	
	Revert C	alls 🔻					_			
	Apply Vi	ew 🔻 Sa	ve View	Show/Hide Columns	Export	▼ Filters ▼				_
	call_rate	QC het_rate	het_rate	QC computed_gender	affymet plate- 🔶 barcode	affymetrix- plate-peg- wellposition	Average call rate for passi	Percent of passing	user_color	
		25.887		female	550094408	E10	98.439	60		*
		25.526		male	550094408	G05	98.439	60		
		25.284		female	550094408	E05	98.439	60		
		24.464		male	550094408	A12	98.439	60		E
		25.046		male	550094408	C01	98.439	60		
	98.727	27.79	27.144	male	550094409	A05	99.093	100		
	98.604	27.976	27.215	male	550094409	B07	99.093	100		
	100.325	27.79	27.649	female	550094409	F09	99.093	100		
	00 046	20 20	26.000	male	550004400	L10	00 002	100		

Importing Assigned Colors

Use this feature if you want to assign colors to a large number of samples or if your Sample Table contains a vast amount of samples and you want to assign a color to only a few samples.

1. Use MS Excel or MS Notepad (as you normally would) to create a two column table. (Figure 69)



IMPORTANT! Your **user_color** entries must match the color pallet naming conventions shown in Figure 70. Example: **RoyalBlue** not **Royal Blue**.

- 2. Save your two column table as a tab-delimited text file to an easily accessible location.
- 3. At the Sample Table, right-click on any sample.

A menu appears. (Figure 70)

- (2)	1.00	0.512	50.704			22.070		mans
NA07034_a550094-4096756-0_	Pass	0 977	99 103	q	8.727	27.79	27.144	male
NA10857_a550094-4096756-0	Copy Sele	ected Rov	v(s)		3.604	27.976	27.215	male
NA12057_a550094-4096756-0	Copy Sele	cted Cel	(s)).325	27.79	27.64	finale
NA12752_a550094-4096756-0	Set User (olor				Import File		-
NA12814_a550094-4096756-0	Set oser e	.0101		-	_	Importine		ł
NA18504_a550094-4099025-0	Remove l	Jser Colo	r			Red		
NA18517_a550094-4099025-0	Pass	0.983	101./9	1		Gold		ale
NA18863_a550094-4099025-0	Pass	0.966	98.221	9		Plue		E .
NA19101_a550094-4099025-0	Pass	0.951	98.468	9		Diue		2
NA19130_a550094-4099025-0	Pass	0.983	98.7	9		Aquamarin	e	2
NA19238_a550094-4099025-0	Pass	0.948	101.016	10		Gray		ale
NA19238_a550094-4099025-0	Pass	0.937	101.191	10	_	LinktConv	_	ale
NA19098_AxiomGWHuSNP1	. Pass	0.929	98.711	9	_	Lightoray		ŧ
NA19099_AxiomGWHuSNP1	. Pass	0.951	101.398	10		Green		ale
NA19116_AxiomGWHuSNP1	. Pass	0.942	101.955	1		RovalBlue		ale
NA19119_AxiomGWHuSNP1	. Pass	0.974	99.283	9		, 		-
NA19127_AxiomGWHuSNP1	. Pass	0.962	101.945	10		Orange		ale
NA19128_AxiomGWHuSNP1	Pass	0.959	99.123	9		LawnGreen		-
NA19130_AxiomGWHuSNP1	Pass	0.938	98.757	9		Magenta		
NA19131_AxiomGWHuSNP1	Pass	0.945	101.857	10		0		ale
NA19140_AxiomGWHuSNP1	Pass	0.944	101.491	10		Brown		ale
NA19141_AxiomGWHuSNP1	Pass	0.96	98.963	9		Yellow		e
NA19143_AxiomGWHuSNP1	Pass	0.92	101.573	10		Purple		ale
NA19144_AxiomGWHuSNP1	Pass	0.972	99.17	9		DI L		2
NA19152_AxiomGWHuSNP1	. Pass	0.948	101.934	10		віаск		ale

- 4. Mouse over Set User Color.
- 5. Click on **Import File...**

An Import User Colors Explorer window appears.

6. Locate your saved TXT file, click to highlight it, then click **Open**.

Your TXT file is now incorporated into the Sample Table.

7. Scroll the Sample Table right to see the added **user_color** column and assigned sample colors.

Viewing User Colors in the Cluster Graph

1. From the Cluster Graph, click the **Color By** drop-down menu. (Figure 71)

BP: D 🖾 BP: QC 🖾 SP: QC_CR vs D 🖄 PV: D 🖾 PV: QC 🖾 🔽 Cluster P	
Scale Settings Color By: user_color Shape By: Genotype Type: Contrast Revert Calls	· 🔛
12 QC allele_mad_residuals_mean QC allele_mad_residuals_stdev QC allele_summarization_mean QC calles_summarization_stdev QC call_rate QC cluster_distance_mean QC cluster_distance_stdev QC computed gender QC cotal_call_rate QC total_het_rate QC total_het_rate QC total_hom_rate reagent_discrimination_value reagent_version saturation_AT saturation_GC T_signal_mean total_call_rate total_het_rate user_color △ △ 11.5 - Genotype A AA △ 11.5 - Ellipses:	r (2) Blue (2) Green (2) marine (2) (1) Gray (1) (2) ge (2) (100) thers) (7) e (73) (43) (8)

Figure 71 Color By menu - user_color

2. From the Sample Table, single-click on a color-coded sample file or Ctrl click, Shift click, or press Ctrl A (to select multiple color-coded sample files).



Each highlighted sample and its assigned color are now displayed in the Cluster Graph, as show in Figure 72.



Removing an Assigned User Color

1. From the Sample Table, right-click on the sample containing the color you want to remove.

A menu appears.

2. Click Remove User Color.

Your previously assigned sample color is now removed.

 Searching Keywords
 Note: The Find in Table tool can locate exact (case insensitive) matches. It also accepts wild-card (*) characters to aid in your search. Example: ABC*

 1. Click inside the
 P Find in Table

 2. Enter a keyword or number.

3. Click the **Up** or **Down** button.

When a match is found, the appropriate table entry is highlighted. If a graph is displayed, the appropriate graph point is also highlighted.

Box Plots

Note: By default, the Viewer generates two Box Plots. (Figure 73)

Viewing the Default Box Plots

Figure 73 Ta	able ar	nd E	Box I	Plot	1												
Summary Sample Table	SNP Sun	nmary 1	Table					F :	3P: D	BP: QC	🛛 SP:	QC_CR vs D	🛛 PV:	D 🛛 PV:	: QC 🛛	Cluster P	
Scatter Plot Box Plot Plate Vie	w Concorda	nce Re	analvze S	elected Sar	nples In	port Same	le Attributes 🔻	Sc	ale Sati	nas							
Revert Calls V			,		<u> </u>					-							
Annha Vinna T	au /I lida Cali		C	Tilter							<u> </u>	DQC by af	fymetrix-p	late-barco	de		
Apply view + Save view Sh	iow/ Hide Colu	imns 🔹	Export	• Fliter	5 -				-							<u> </u>	
Sample Filename	Pass/Fail	DQC	QC call_rate	call_rate	QC het_rate	het_rate	QC computed_gen 1				I		Ħ		T		
2 (1).CEL	Fail	0.985	98.406		25.887		female	A									
2 (3).CEL	Fail	0.974	99.685		25.284		female										
NA12057_a550094-4096756-0	Pass	0.992	100.598	100.325	27.79	27.649	female						'				
NA18517_a550094-4099025-0	Pass	0.983	101.79	101.44	26.362	25.679	female										
NA19238_a550094-4099025-0	Pass	0.948	101.016	100.599	26.97	26.397	female		0.75								
NA19238_a550094-4099025-0	Pass	0.937	101.191	100./92	26.511	25.981	female		0.75								
NA19099_AxiomGWHuSNP1	Pass	0.951	101.398	101.0//	26.124	25./46	temale										
NA19110_AXIOMGWHUSNP1	Pass	0.942	101.955	101.42	20.073	25.038	female										
NA19127_AxiomGWHuSNP1	Pass	0.962	101.945	101.381	20,455	25.901	female										
NA19140 AxiomGWHuSNP1	Pass	0.945	101.007	101.252	26.547	25.851	female										
NA19143 AxiomGWHuSNP1	Pass	0.92	101.573	100.991	26.795	26.118	female										
NA19152 AxiomGWHuSNP1	Pass	0.948	101.934	101.545	26.228	25,799	female										
NA19159 AxiomGWHuSNP1	Pass	0.929	101.413	101.029	26.377	25.852	female		2.								
NA19193 AxiomGWHuSNP1	Pass	0.953	101.599	101.296	26.609	25,705	female	2	۲ ₀₅								
NA19206_AxiomGWHuSNP1	Pass	0.944	101.583	101.166	26.119	25.714	female							'			
NA19222_AxiomGWHuSNP1	Pass	0.874	100.273	99.811	27.177	26.541	female										
NA19238_AxiomGWHuSNP1	Pass	0.944	101.578	101.235	26.578	26.008	female										
NA11840_AxiomGWHuSNP1	Pass	0.871	100.371	99.964	29.57	29.226	female										
NA11882_AxiomGWHuSNP1	Pass	0.915	101.553	101.186	28.796	27.976	female										
NA12239_AxiomGWHuSNP1	Pass	0.88	100.634	100.195	29.26	28.962	female										
NA12249_AxiomGWHuSNP1	Pass	0.924	101.656	101.173	28.095	27.622	female										
NA12717_AxiomGWHuSNP1	Pass	0.878	100.769	100.273	29.993	28.917	female										
NA12751_AxiomGWHuSNP1	Pass	0.939	101.754	101.414	28.394	28.079	female		0.25								
NA12/61_AxiomGWHuSNP1	Pass	0.909	101.088	100./61	28.218	28.118	female										
NA12/63_AxiomGWHuSNP1	Pass	0.931	101.645	101.123	29.276	28.001	female										
NAL2813_AxiomGWHuSNP1	Pass	0.9	101.32	100.658	29.183	28.275	temale										
NA12872 AviomGWHuSNP1	Pass	0.904	101.0/3	101.524	29,415	20.48/	female										
NA12875 AviomGWHuSNP1	Dace	0.833	00.470	08 0/12	20.002	27.038	female										
NA12892 AxiomGWHuSNP1	Pass	0.942	102 037	101 587	28 755	29.094	female										
NA18526 AxiomGWHuSNP1	Pass	0.907	101.516	101.072	26,217	25.485	female										
NA18529 AxiomGWHuSNP1	Pass	0.921	101.465	101.159	25.598	25,231	female										
NA18532 AxiomGWHuSNP1	Pass	0.937	101.764	101.349	25,939	25,218	female		0								
NA18537 AxiomGWHuSNP1	Pass	0.896	101.191	100.756	26,284	26.08	female			002	675	502	310	211	127	131	135
NA18540_AxiomGWHuSNP1	Pass	0.952	101.589	101.301	24.577	23.496	female			710	910	810	110	210	411	411	411
NA18545_AxiomGWHuSNP1	Pass	0.963	102.182	101.894	25.774	24.88	female			020	180	082	060	111	110	011	110
NA18566_AxiomGWHuSNP1	Pass	0.943	102.022	101.542	25.903	25.236	female			380	5756	025	020	5292	5295	295	5295
NA18570_AxiomGWHuSNP1	Pass	0.955	102.094	101.739	25.542	25.146	female			1081	960t	660t	660t	1105	1105	1105	1105
NA18573_AxiomGWHuSNP1	Pass	0.92	101.558	101.163	26.439	25.634	female	-		0944	9944	0944	0944	0944	0944	0947	0944
4	n	0.017	00 201		25.024	26.240				5500	2200	5500	2200	5500	2200	5500	2200
	C 1.000	<u></u>															
Find in Table NV Rov	w Count: 226	Selected	1:0				Show Filtered On	У									

IMPORTANT! You cannot change a plot's axis values after it has been created. However, you can change its scale and coloring properties. See "Changing the Box Plot's Scale Setting Ranges" on page 62.

To change a Box Plot's axis properties, you must create a new Box Plot. See "Adding a New Box Plot" on page 62.

Changing the Box Plot's Scale Setting Ranges 1. Click Scale Settings .

The following window appears. (Figure 74)

Figure 74 window	Scale Settings
Scale Settings	×
Auto Scale Min X: 0.8 Min Y: 95	Max X: 1 Max Y: 100
	Default
	OK Cancel

By default, the window displays your current range values.

- 2. Uncheck the **Auto Scale** check box to enter different ranges in the provided text fields.
- 3. Click OK.

Your new settings are now reflected within the Box Plot. Modified Set Scale values are auto-saved.

If needed, click Default to return all values back to their factory settings.

Adding a New Box Plot

1. Click the **Box Plot** button.

The following window appears: (Figure 75)

Figu menu	r e 75 JS	Box F	Plot Ve	ersus
New Bl Group Y-	o By: affy axis: DQ	/metrix-plat C	e-barcode	× •
			ОК	Cancel

- 2. Click the **Group By** drop-down menu to select the X-axis for your new Box Plot. Your X-axis selection determines your new Box Plot's boxes and whiskers, based on the data group of values that are compiled.
- Click the Y-axis drop-down menu to select the Y-axis you want. For Group By and Y-axis definitions, see "Sample Table" on page 154.
- 4. Click OK.

A new Box Plot window tab is created.

Reading Box Plot Percentiles

See Figure 76.



At any time, click **X** to remove a window/tab, as shown in Figure 77.

Figure 77 New Window/Tab	
BP: 🗵 BP: Q 🖾 SP: QC_CR vs 🖄 PV: 🖄 PV: Q 🖾 Cluster	
Scale Settings	Remove Tab

Saving the Current Box Plot View

- 1. Click the **Save Image button**.
 - An Explorer window appears.
- 2. Navigate to where you want to save the .PNG file, enter a filename, then click **Save**.



Scatter Plot

By default, the Viewer generates 1 Scatter Plot of **QC call_rate** vs. **DQC**. The data displayed in the plot are colored and shaped by **QC computed_gender**, as shown in Figure 78.

Viewing the Default Scatter Plot

 Click to highlight a table entry to view its location within the Scatter Plot or click on a data point to highlight its corresponding entry in the Sample Table. (Figure 78)

Figure 78 Table and Scatter Plot ary Sample Table SNP Summary Table BP: D... 🛛 BP: QC ... 🖾 SP: QC_CR vs D... 🖾 PV: D... 🖾 PV: QC ... 🖾 Cluster P.. Scatter Plot Box Plot Plate View Concordance Reanalyze Selected Samples Import Sample Attributes 🔻 Scale Settings Color By: QC computed_gender Shape By: QC computed_gender Revert Calls 🔻 QC call rate vs DQC 100 Apply View 🔻 Save View Show/Hide Columns 🔻 Export 🔻 Filters 🔻 QC computed female male unknown (109 (95 DQC QC call_rate QC computed_gen.. Sample Filename Pass/Fail call_rate het_rate het_rate QC computed gender NA19141_AxiomGWHuSNP1_... 0.96 98 963 98 688 25.036 24.87 male ∆ female ○ male ⊽ unknown (109)NA19144 AxiomGWHuSNP1 Pass 0.972 99.17 98.913 25.903 24.898 male (95) NA19153_AxiomGWHuSNP1_... 0.953 98.901 98.602 25.062 24.882 male ass NA19160_AxiomGWHuSNP1_... 27.135 0.826 96,302 male NA19192_AxiomGWHuSNP1_ Pass 0.939 98,582 98.37 25.026 25.067 male NA19207 AxiomGWHuSNP1 25.05 male Pass 0.934 98,417 98.143 25.836 24.967 male NA19223_AxiomGWHuSNP1_... 0.935 98.747 98.458 25.851 ass 98.25 98,732 25.153 male NA19239_AxiomGWHuSNP1_... 99.066 25.567 0.961 NA11839 AxiomGWHuSNP1 0.965 98.695 98.382 27.842 27.104 male Pass NA11881 AxiomGWHuSNP1 .. 98.68 98.331 28.146 27.098 male Pass 0.919 NA12146_AxiomGWHuSNP1_... 0.927 98.576 98.259 27.749 27.524 male ass NA12248_AxiomGWHuSNP1_... Pass 0.956 99.262 98,793 27.646 26.944 male NA12264 AxiomGWHuSNP1_.. 0.933 98,984 98,551 28.353 27.332 male Pass NA12716_AxiomGWHuSNP1_... 0.883 97,709 27.687 27.463 male 98.241 Pass 0 NA12750_AxiomGWHuSNP1_.. 0.931 98.927 98.521 27.95 27.314 male Pass NA12760 AxiomGWHuSNP1 Fail 0.823 96.044 29,296 male 27.419 male 97,929 NA12812 AxiomGWHuSNP1 ... Pass 0.905 98.288 27.12 0 call_rate NA12814_AxiomGWHuSNP1_... 97.437 Pass 0.832 96.911 28.062 male 96 NA12872_AxiomGWHuSNP1_... 😽.678 male 0.899 98.055 97,706 28.131 NA12874_AxiomGWHuSNP1_... Pass 0.926 99.051 98,549 26.975 26.307 male g 0 27.181 male NA12891_AxiomGWHuSNP1_.. Pass 0.943 98.927 98.714 27.445 NA18605_AxiomGWHuSNP1_... 0.914 98.494 98.19 25.825 24.715 male ass 25.093 male NA18608_AxiomGWHuSNP1_... 0.873 98,169 97,492 25.304 Pass NA18944_AxiomGWHuSNP1_.. 0.951 99.082 98,762 24.716 24.198 male Pass NA18945_AxiomGWHuSNP1_... 0.92 98.602 98.473 25.346 24.471 male Pass NA18953_AxiomGWHuSNP1_... Pass 0.909 98.375 98.036 25.418 24.612 male NA18959_AxiomGWHuSNP1_... 0.897 98,422 98.021 25.5 25.02 male Pass NA18960 AxiomGWHuSNP1 .. 25.351 Pass 0.887 97.22 96.966 25.092 male NA18961_AxiomGWHuSNP1_... 0.883 95,771 26.387 Fail male NA18967_AxiomGWHuSNP1_... 0.908 98.12 24.512 male 98.447 24.959 94.75 NA18970_AxiomGWHuSNP1_ Pass 0.921 98.473 98.086 25.82 24.784 male 97.876 NA18971 AxiomGWHuSNP1 ... Pass 0.891 98.205 25.077 24.628 male 25.176 male NA19119_AxiomGWHuSNP1_... 0.929 98.747 98.402 25.521 Pass 25.375 male NA19128_AxiomGWHuSNP1_... 0.915 98.726 26.016 98.25 NA19141 AxiomGWHuSNP1 0.912 98.68 98.142 25.273 25.116 male Pass NA19144 AxiomGWHuSNP1 ... 99.216 25.031 male 0.956 98.829 26.093 Pass NA19160_AxiomGWHuSNP1_... 0.916 98.876 98.346 25.83 25.236 male ass NA19192_AxiomGWHuSNP1_... Pass 0.957 99.268 98,884 25.139 24.995 male 0 NA19210 AxiomGWHuSNP1 Pass 0.9 97.844 97.646 27,486 26.837 male NA19239_AxiomGWHuSNP1 98.313 97.951 25.836 25.514 male 0.9)ass 93 ⊾ 0.8 0.85 0.9 0.95 𝒫 Find in Table Row Count: 226 Selected: 1 👔 👎 Show Filtered Only DQC

IMPORTANT! You cannot change the default Scatter Plot's pre-defined X and Y definitions, however you can change its Scale Settings and Color By and Shape By configuration.

To change a Scatter Plot's axis properties, you must create a new Scatter Plot. See "Adding a New Scatter Plot and Selecting its X and Y Properties" on page 65.

Changing the Scatter Plot's Setting Ranges 1. Click Scale Settings.

The following window appears. (Figure 79)

Scale Set	tinas		
	cings		
Min Vi	Scale	Max Vi (1
Min Vi	0.0	Max Vi -	100
	55	Ividx 1;	100
L			Defaul

By default, the window displays your current range values.

- 2. Enter your new ranges in the appropriate text fields.
- 3. Click OK.

Your new settings are now reflected within the Scatter Plot. Modified Set scale values are auto-saved.

If needed, click Default to return all values back to their factory settings.

1. Click the **Scatter Plot** button.

The following window appears: (Figure 80)

Figure menus	80	Sca	itter	Plot	Ve	rsus
New SP						
X-axis:	DQC					•
Y-axis:	QC ca	ll_rate				•
				ОК		Cancel

- 2. Use the drop-down menus to select your Plot's versus scenario (X and Y axis). For definitions, see "Sample Table" on page 154.
- 3. Click OK.

A new Scatter Plot window tab is created.

At any time, click **X** to remove a window/tab, as shown in Figure 81.

BP: 🛛 BP: Q 🖾 SP: QC_CR vs 🖾 PV: 🖾 PV: Q 🖾 Cluster 🔽 🔲 🗖	Figure 81 New Window/Tab	
Scale Settinge	BP: 🕄 BP: Q 🗟 SP: QC_CR vs 🖄 PV: 🗟 PV: Q 🗟 Cluster	
Sale Settings	Scale Settings	Remove Tab

Adding a New Scatter Plot and Selecting its X and Y Properties 4. Click the Color By and Shape By drop-down menus to select the combination view you want. See "Sample Table" on page 154 for Color By and Shape By definitions. Note: Your imported sample attributes are also available for use within the Color By and Shape By drop-down menus. For information on importing sample attributes, see "Importing Sample Attributes" on page 46. A legend appears within the plot. (Figure 82)



The graph can display up to 10 different colors and up to 10 different shapes. If the attributes selected for display have more than 10 categories, categories 1 through 9 are displayed normally, but categories 10 and higher get grouped together.

If your study has more than 10 values:

- If the value is text, the software takes the first nine values and assigns each a color or shape. The remaining values are put into a bin labeled **Other**. All values in the Other bin have the same color or shape.
- If the value is a date or number, the software divides the range of data into 10 equal bins and assigns a color or shape to each bin. If the data includes one or more outliers, it is possible to have one value in a particular bin and all other values in another bin.
- 1. Click Configure

The Color Scale Configuration window appears. (Figure 83)

Figure 8	3 Color By option	າຣ
Color Scale Con	figuration	×
28.941	Auto Scale Cutoff Type: Above Cutoff Failing Max: 28.941 Cutoff: 26.125	Above Cutoff Failing Above Cutoff Failing Below Cutoff Failing No Cutoff
23.309	23.309	5
	OK Cancel	

- 2. Use the provided text fields and color drop-down menus to customize your Color By selection.
- Auto Scale check box (when checked) uses the actual minimum (lower bound) and maximum (upper bound) as your min/max scale. Uncheck the Auto Scale check box to enter your min and max number scales in the provided fields.
- Click the **Cutoff Type** drop-down menu to select your cutoff preference.
 - Above Cutoff Failing This presents a hard visual cutoff graph of all values that fail ABOVE the Cutoff value entered. The Above Cutoff data is represented by the color defined for Max. (Green in Figure 83)

Customizing Color By Settings

Axiom[™] Analysis Suite 3.1 User Guide

 Below Cutoff Failing - This presents a hard visual cutoff graph of all values that fail BELOW the Cutoff value entered. The Below Cutoff data is represented by the color defined for Min. (Red in Figure 84)

Figure 84	4 Below Cutoff
Color Scale Cont	figuration 🗙
28.941	✓ Auto Scale Cutoff Type: Below Cutoff Failing
26.125	Max: 28.941 ■ ▼ Cutoff: 26.125 ■ ▼
23.309	Min: 23.309
	Default
	Default OK Cancel

 No Cutoff - This presents a smooth 3-point gradient of your defined Max, Min, and colors. (Figure 85)

Figure 8	5 No Cutoff	
Color Scale Cor	nfiguration	×
28.941	📝 Auto Scale	
	Cutoff Type:	
	No Cutoff 🔹	
26 1 25	Max:	
20,123	Cutoff:	
	26.125	
	Min:	
23.309	23.309	
	Defau	ult
	OK Cance	

3. Click OK.

Your Cutoff preference, entered values, and color selections are now displayed on the graph and saved for future use. If needed, click Default to revert all values back to their factory settings.

Saving the Current Scatter Plot View

1. Click the **Save Image La** button.

An Explorer window appears.

2. Navigate to where you want to save the .PNG file, enter a filename, then click **OK**.

Plate Views

By default, the Viewer generates two Plate Views. (Figure 86) Each have a factory set metric. Plate View 1 is **DQC by Plate** and Plate View 2 is **QC call_rate by Plate**.

To display a different metric you must create a new Plate View. For more details, see "Adding a New Plate View Metric" on page 70.

The Plate Views display the currently selected (highlighted) metric from the Sample Table and are a graphic representation of the plate used. For example, 96 count plate layouts are shown in Figure 86.

Viewing the Default Plate Views

1. Click to highlight a table entry to view its location within the Plate View or click on a plate position to highlight its corresponding table entry, as shown in Figure 86.



IMPORTANT! You cannot change a default Plate View, however you can change its Scale Settings, gradient, and coloring. See "Customizing your Plate View Settings" on page 71.

З

Adding a New Plate View Metric

The default Plate Views cannot be altered, therefore you must click the **Plate View** button to create a new Plate View to reflect your Metric change.

1. Click the **Plate View** button.

The following window appears: (Figure 87)

Figure 87 Plate View Metric setting	
New PV Metric: Pass/Fail	×
OK Cancel	5

- 2. Use the drop-down menus to select your Plate View's Metric setting. See the tables in "Sample Table" on page 154 for Metric definitions.
- 3. Click OK.

The new Plate View window tab appears.

At any time, click X to remove a window/tab, as shown in Figure 88.

Figure 88 New Window/Tab		
BP: 🛛 BP: Q 🖾 SP: QC_CR vs 🖾 PV: 🖾 PV: Q 🖾 Cluster	V .,	
Scale Settings		Remove Tab



Customizing your Plate View Settings

1. Click Configure .

The Color Scale Configuration window appears. (Figure 89)

Figure 89 options	9 Color Scale	
Color Scale Cor	nfiguration	>
28.941	📝 Auto Scale	
	Cutoff Type:	
	Above Cutoff Failing 🔹	
25.125	Max:	
20.125	28.941	
	26.125	
	Min:	
23.309	23.309	
	Default	:
	OK Cancel	

- 2. Use the provided text fields and color drop-down menus to customize your Color By selection.
- Auto Scale check box (when checked) uses the actual minimum (lower bound) and maximum (upper bound) as your min/max scale. Uncheck the Auto Scale check box to enter your min and max number scales in the provided fields. Note: If the Auto Scale check box remains unchecked, you must enter new scale limits for each subsequent analysis.
- Click the **Cutoff Type** drop-down menu to select the appropriate cutoff (based on the custom Cutoff value you entered).
- 3. Click OK.

Your new preferences are now displayed and saved for future use.

At any time, click the **Default** button to revert all the Color Scale Configuration window values back to their factory setting.

- Saving the Current Plate View
- Click the Save Image button. An Explorer window appears.
- 2. Navigate to where you want to save the .PNG file, enter a filename, then click OK.



Concordance Checks

- **Compare all combinations** enables you to compare the SNP calls for all samples. The concordance between all pairwise comparisons for the samples in the dataset/ suitcase are reported.
- **Compare to reference** enables you to compare every sample to a single reference file.

Running a Concordance Check 1. Click the **Concordance** button.

The following window appears: (Figure 90)

Figure 90 Concordance window
Concordance X
 Compare all combinations Compare to reference
Browse for reference file:
 Compare all SNPs Compare SNPs within SNP Summary Table Compare signature SNPs
OK Cancel

Comparing All Combinations

- 1. Make sure the **Compare all combinations** radio button is selected.
- 2. By default, the **Compare all SNPs** button is selected. If needed, click the **Compare signature SNPs within the SNP Summary Table**, or **Compare signature SNPs** radio button.
- 3. Click OK.

After a few moments, the following Concordance window tab appears: (Figure 91)

IMPORTANT! The amount of time to calculate concordance is proportional to the number of samples squared and the number of SNPs. It is highly recommended you use <1000 SNPs for an All versus All concordance check.
	~			
12	- 25	ь.		
82.	-24	σ.	50	
	r			
		ь.	100	
		-	- 25	
18.		σ.		
	h	-		

Figure	91 De	fault Co	ncordanc	e Table example
	γ			
Summary	Sample 1	Table SN	P Summary Ta	ble External Tools Concordance
Apply View	 Save Vie 	w Show/Hi	de Columns 🔻	Export Filters
			%	
Index	Base	Reference	Concordance	
1	28//_Axio	28/8_Axio	64.294	
2	28/7_Axio	28/9_Axio	57.144	
5	2877_Axio	2880_AXIO	59.274	
4	2877_Axio	2881_AXIO	57.157	
5	2877 Avio	2883 Avio	57.13/	
7	2877 Avio	2005_AXIO	64.069	
/ 8	2877 Axio	2885 Avic	56,922	
9	2877 Avio	2887 Avio	6/ 122	
10	2877 Avic	2888 Avic	58 59/	
11	2877 Avic	2889 Avic	57.007	
12	2877 Axie	2890 Axie	63,819	
13	2877 Avio	2891 Avio	56.931	
14	2877_Axio	2892 Axio	57.061	
15	2877 Avio	2893 Avio	57.12	
16	2877 Axio	2894 Axio	64,313	
17	2877 Axio	2895 Axio	64.35	
18	2877 Axio	2896 Axio	57.142	
19	2877 Axio	2897 Axio	64.04	
20	2877 Axio	2898 Axio	58.86	
21	2877 Axio	2899 Axio	57.049	
22	2877 Axio	2900 Axio	56.827	
23	2877 Axio	2901 Axio	57.112	
24	2877 Axio	2902 Axio	64.178	
25	2877_Axio	2903_Axio	58.895	
26	2877_Axio	2904_Axio	56.902	
27	2877_Axio	2905_Axio	57.054	
28	2877_Axio	2907_Axio	64.122	
29	2877_Axio	2908_Axio	56.957	
30	2877_Axio	2909_Axio	64.429	
31	2877_Axio	2910_Axio	56.887	
32	2877_Axio	2915_Axio	56.952	
33	2877_Axio	2916_Axio	64.49	
34	2877_Axio	2917_Axio	57.753	
35	2877_Axio	2918_Axio	57.146	
36	2877_Axio	2943_Axio	64.28	
37	2877_Axio	2944_Axio	64.369	
38	2877_Axio	2946_Axio	57.067	
39	2877_Axio	2947_Axio	58.427	
40	2877_Axio	2948_Axio	64.345	
₽ Find in	Table 🔨	Row Court	t: 7626 Selected	t: 0 👔 📕 🔍 Show Filtered On

For definitions of the Concordance columns, see Table 13 on page 158.

Comparing to Reference

1. Click the **Compare to reference** button.

The Browse for the reference file field is now activated.

2. Click the **Browse** button.

A Windows Explorer window appears.

3. Navigate to the appropriate reference file location, then click **Open**.

Your Reference file is displayed.

IMPORTANT! The reference file you select can have more than two columns. However, only the first two columns are used during the *Compare to Reference* concordance check. Also, your reference genotypes must be reported using letter call codes (e.g. AA, AB, BB, NoCall).

- By default, the Compare all SNPs button is selected. If needed, click the Compare signature SNPs within the SNP Summary Table, or Compare signature SNPs radio button.
- 5. Click OK.

After a few moments, a Concordance window tab appears.

Reanalyzing Samples

To reanalyze sample(s) displayed in the Sample Table:

- 1. Single-click on a CEL file or Ctrl click, Shift click, or press Ctrl A (to select multiple files).
- 2. Click the **Reanalyze Selected Samples** button.

The following message appears. (Figure 92)



3. Click OK.

The Viewer closes. Your selected samples are displayed and ready for re-analysis in a New Analysis window tab. (Figure 93)

Figure 93 New Analysis	window tab		
New Analysis Dashboard Preferences			
Mode: Genotyping Array Type: Axiom	n Genome-Wide CEU 1 Array r6 🔹 Import CEL Files Import CEL Files by Txt Re	move Selected Files	
CEL Files: 18	Analysis Settings	Threshold Settings	
File Name	Select Analysis Configurations	Select Threshold Configura	tions
NA19119_AxiomGWHuSNP1_20100906_InputMassTitr	Batch Configuration Restore Save Save As	Batch Configuration	
NA19127_AxiomGWHuSNP1_20100906_InputMassTitr	© Cepetyning		
NA19128_AxiomGWHuSNP1_20100906_InputMassTitr NA19130_AxiomGWHuSNP1_20100906_InputMassTitr	Anabreis Eiler	SINF QC	6
NA19131_AxiomGWHuSNP1_20100906_InputMassTitr	Aviom GW Hu SND LessThan96 Sten2 of ant-proheset-genotype AviomGT1 a	Name	Settings
NA19140_AxiomGWHuSNP1_20100906_InputMassTitr	Prior Model File:	species-type	Diploid •
NA19141_AxiomGWHuSNP1_20100906_InputMassTitr	Axiom GW Hu SNP.r6.AxiomGT1	cr-cutoff	≥ ▼ 94
NA19143_AxiomGWHuSNP1_20100906_InputMassTitr	SNP List File:	fld-cutoff	2 • 3.6
NA19152_AxiomGWHuSNP1_20100906_InputMassTitr	Axiom_GW_Hu_SNP.r6.AxiomGWAS_HuSNP_1_step2	het-so-sutoff	
NA19153_AxiomGWHuSNP1_20100906_InputMassTitr	Gender File (optional):	net-so-cuton	
NA19159_AxiomGWHuSNP1_20100906_InputMassTitr		het-so-otv-cutoff	≥ - 0.3
NA19160_AxiomGWHuSNP1_20100906_InputMassTitr NA19192_AxiomGWHuSNP1_20100906_InputMassTitr-	Hints/Inbred File (optional):	hom-ro-1-cutoff	≥ ▼ 0.6
NA19193 AxiomGWHuSNP1 20100906 InputMassTitr	🔘 Inbred 🖲 Hints 🛛 🗙	hom-ro-2-cutoff	2 • 0.3
NA19206_AxiomGWHuSNP1_20100906_InputMassTitr	Posterior File Name (optional):	have as 2 substit	
NA19207_AxiomGWHuSNP1_20100906_InputMassTitr		nom-ro-s-cutori	2 -0.9
NA19222_AxiomGWHuSNP1_20100906_InputMassTitr	ps2snp File (recommended):	hom-ro	true 🔹 🔹
	Axiom_GW_Hu_SNP.r6.ps2snp_map	hom-het	true 🔹 🔹
		num-minor-allele-cutoff	2 2
		priority-order	Change List Order PolyHighResolution, NoMi 🔊
		recommended	Checklist PolyHighResolution, NoMinorHom,
Output Folder: C:\Users\Public\Documents\AxiomAnalys	isSuite\Output	Brov	wse Batch Name: Test_21 Run Analysis

4. See Chapter 2, "Performing an Analysis" on page 19 for instructions on setting up an analysis.



The Viewer: SNP Summary Table and Cluster Plot

SNP Summary Table

Figure 94 is an example of a standard SNP Summary Table

Figure 94	SNP Su	mma	ary Table win	dow													
SNP Summary Tab	le																*
Select Annotation	Manage SNP Lis	t 🔻 🛛	Change/Revert Calls 🔻	Reanal	vze 🔻												
Apply View V	we View Show/	Hide Co	lumps V Export V	Filterr	v												
Apply view 1 3	ave view Show/	Hide CC	numins • Export •	Fillers													
probeset_id	ConversionType	CR	MinorAlleleFrequency	H.W.p- Value	FLD	HomFLD	HetSO	HomRO	Nclus	n_AA	n_AB	n_BB	n_NC	hemizygous	HomHet	gender_metrics	Call Modified
AFFX-KIT-000001	PolyHighResol	100	0.464	0.306	20.691	44.561	0.325	2.754	3	39	76	51	0	0	0	all	False
AFFX-KIT-000002	PolyHighResol	99.398	0.33	0.482	8.127	18.035	0.191	0.846	3	76	69	20	1	0	0	all	False
AFFX-KIT-000003	PolyHighResol	100	0.22	0.007	6.492	13.807	0.146	0.738	3	107	45	14	0	0	0	all	False
AFFX-KIT-000004	PolyHighResol	100	0.319	0.145	15.227	33.186	0.318	2.124	3	81	64	21	0	0	0	all	False
AFFX-KIT-000005	PolyHighResol	100	0.081	1	5.139	11.779	0.094	1.069	3	140	25	1	0	0	0	all	False
AFFX-KIT-000008	PolyHighResol	100	0.322	0.532	5.412	11.248	0.133	1.127	3	19	69	78	0	0	0	all	False
AFFX-KIT-000009	NoMinorHom	100	0.075	0.602	21.081		0.247	4.47	2	0	25	141	0	0	1	all	False
AFFX-KIT-000012	PolyHighResol	100	0.368	0.388	20.114	42.061	0.238	2.784	3	69	72	25	0	0	0	all	False
AFFX-KIT-000013	PolyHighResol	99.398	0.136	0.19	5.134	10.814	0.169	0.813	3	125	35	5	1	0	0	all	False
AFFX-KIT-000014	PolyHighResol	100	0.099	0.663	18.947	38.956	0.567	2.803	3	2	29	135	0	0	0	all	False
AFFX-KIT-000015	PolyHighResol	100	0.181	1	30.972	67.77	0.439	4.658	3	5	50	111	0	0	0	all	False
AFFX-KIT-000016	PolyHighResol	100	0.135	1	23.086	48.374	0.579	2.409	3	3	39	124	0	0	0	all	False
AFFX-KIT-000017	PolyHighResol	100	0.078	1	20.903	44.214	0.42	3.888	3	141	24	1	0	0	0	all	False
AFFX-KIT-000018	PolyHighResol	99.398	0.23	1	16.629	34.399	0.254	2.451	3	9	58	98	1	0	0	all	False
AFFX-KIT-000019	PolyHighResol	99.398	0.373	0.043	6.281	15.899	0.235	1.536	3	29	65	71	1	0	0	all	False
AFFX-KIT-000021	PolyHighResol	99.398	0.179	0.79	8.169	17.256	0.331	1.384	3	112	47	6	1	0	0	all	False
AFFX-KIT-000022	PolyHighResol	100	0.434	0.381	18.57	38.557	0.397	2.523	3	56	76	34	0	0	0	all	False
AFFX-KIT-000023	PolyHighResol	99.398	0.309	0.238	6.384	14.553	0.193	1.015	3	82	64	19	1	0	0	all	False
AFFX-KIT-000025	PolyHighResol	100	0.494	0.439	17.222	36.978	0.348	2.333	3	43	78	45	0	0	0	all	False
AFFX-KIT-000026	PolyHighResol	99.398	0.436	0.147	13.408	33.126	0.509	1.785	3	36	72	57	1	0	0	all	False
AFFX-KIT-000027	PolyHighResol	100	0.41	0	23.078	47.688	0.238	2.894	3	41	54	71	0	0	0	all	False
AFFX-KIT-000029	PolyHighResol	99.398	0.397	0	11.659	24.178	0.135	1.097	3	75	49	41	1	0	0	all	False
AFFX-KIT-000031	PolyHighResol	100	0.238	0.671	15.932	33.638	0.379	2.353	3	95	63	8	0	0	0	all	False
AFFX-KIT-000032	PolyHighResol	100	0.377	0.071	20.768	43.819	0.607	2.467	3	29	67	70	0	0	0	all	False
AFFX-KIT-000033	PolyHighResol	99.398	0.258	0	13.454	27.084	0.505	2.629	3	102	41	22	1	0	0	all	False
AFFX-KIT-000049	PolyHighResol	100	0.386	0.583	12.018	25.089	0.511	2.074	3	61	82	23	0	0	0	all	False
AFFX-KIT-000050	PolyHighResol	100	0.473	0.227	6.032	12.798	0.147	0.776	3	50	75	41	0	0	0	all	False
AFFX-KIT-000121	PolyHighResol	100	0.283	0.15	13.623	28.574	0.309	2.419	3	10	31	49	0	0	0	female	False
AFFX-KIT-000198	PolyHighResol	100	0.356	0.228	17.117	36.866	0.365	2.704	3	40	36	14	0	0	0	female	False
AFFX-KIT-000209	PolyHighResol	100	0.333	0.018	4.766	10.896	0.129	0.697	3	15	30	45	0	0	0	female	False
AFFX-KIT-000226	PolyHighResol	98.889	0.371	0.002	12.302	27.039	0.556	2.075	3	42	28	19	1	0	0	female	False
AFFX-KIT-000408	PolyHighResol	100	0.217	1	19.59	41.485	0.12	3.567	3	4	31	55	0	0	0	female	False
AX-162003418	MonoHighRes	100							1							NA	False
AX-162005580	PolyHighResol	100							2							NA	False
AX-162007660	CallRateBelow	77.711							3							NA	False
AX-162011899	PolyHighResol	100							3							NA	False
AX-162020167	Other	100							1							NA	False
NA	∧ ∨ Row Co	ount: 37	Selected: 0 👔 🤳													🗸 Show	Filtered Only



Multi-allele probesets are probesets that can report calls from more than two alleles. Multi allele probesets can be identified by selecting the annotation field **Allele_Count**. Additional columns can be shown that report metrics for multi-allele probesets.

Multi-allele probesets are reported differently than the more common biallele probesets.

- Multi-allele probesets will not report NoMinorHom or OTV ConversionTypes.
- Multi-allele probesets do not report values for many of the default SNP Summary Table columns. Click **Show/Hide Columns** to select additional Columns with multi-allele probeset metrics.

Note: If an analysis batch does not include samples needed to evaluate a SNP's performance, then no metrics (including ConversionType) will be reported. For example, if an analysis batch does not contain male samples, no metrics are reported for Y chromosome SNPs and none of these SNPs are added to the Recommended SNP list.

Figure 95 represents a SNP Summary Table with CN-aware genotyping and Allele Translation.

Note: The Perform Allele Translation, button only appears if supported arrays are available. See Chapter 5, "Allele Translation" on page 97 for more information.

A SNP Summary Table with CN-aware genotyping also displays a count of calls for hemizygous genotypes (n_A, n_B) and a count of ZeroCN calls (n_CN0), as shown in Figure 95. These additional columns appear for arrays that support copy number-aware genotyping. For more information, see Appendix A, "Predefined Region CN Analysis Variations" on page 122.

Fig	gure 95	5 SNP S	umn	nary Table wi	th CN	V-av	vare g	enot	yping	and	a All	ele T	rans	slati	on						
Sum Select Appl	mary Sa t Annotation y View 🔻 🖇	mple Table / Manage SNP Lis Save View Show/	SNP Su it 🔻 (Hide Co	Immary Table CN Change/Revert Calls olumns Export	Summary Reanalyz Filters 🔻	y Table ie 🔻 🖡	e Cluste Perform Alle	er Plot le Trans	CN Rec	jion Pl	ot		F								
pri	obeset_id	ConversionType	CR	MinorAlleleFrequency	H.W.p- Value	FLD	HomFLD	HetSO	HomRO	Nclus	n_AA	n_AB	n_88	n_A	n_B	n_CN0	n_NC	hemizygous	HomHet	Call Modified	
AFFX	-KIT-000001	PolyHighResol	100	0.464	0.306	20.649	44.464	0.322	2.754	3	39	76	51	0	0	0	0	0	0	False	
AFFX	-KIT-000002	PolyHighResol	99.398	0.33	0.482	8.127	17.968	0.199	0.854	3	76	69	20	0	0	0	1	0	0	False	
AFFX	-KIT-000003	PolyHighResol	100	0.22	0.007	6.331	13.592	0.153	0.76	3	107	45	14	0	0	0	0	0	0	False	
AFFX	-KIT-000004	PolyHighResol	100	0.319	0.145	14.997	32.701	0.315	2.126	3	81	64	21	0	0	0	0	0	0	False	
AFFX	-KIT-000005	PolyHighResol	99.398	0.079	1	5.144	11.911	0.128	1.072	3	140	24	1	0	0	0	1	0	0	False	
AFFX	-KIT-000008	PolyHighResol	100	0.322	0.532	5.368	11.118	0.132	1.123	3	19	69	78	0	0	0	0	0	0	False	
AFFX	-KIT-000009	NoMinorHom	100	0.075	0.602	20.912		0.247	4.47	2	0	25	141	0	0	0	0	0	1	False	
AFFX	-KIT-000012	PolyHighResol	100	0.368	0.388	19.026	39.889	0.24	2.763	3	69	72	25	0	0	0	0	0	0	False	
AFFX	-KIT-000013	PolyHighResol	99.398	0.136	0.19	5.259	10.952	0.193	0.84	3	125	35	5	0	0	0	1	0	0	False	
AFFX	-KIT-000014	PolyHighResol	100	0.099	0.663	19.334	39.101	0.554	2.898	3	2	29	135	0	0	0	0	0	0	False	-
APPY	LTT 000045		400	0 404		20.240	C 4 000	0 400	4.620		r			0	•	م	<u>ہ</u>	<u>م</u>		- I	_
<u>م</u>	^r ind in Table	A Row Co	ount: 129	9544 Selected: 1 👔 🤳															📝 Sh	ow Filtered C)nly

Using the SNP Summary Table

Setting your SNP Summary Table View 1. Click the Apply View drop-down. (Figure 96)

Figure 96 Apply View drop-down
Apply View 🔻
Default View
All Columns View

- The **Default View** is the initial table view and includes a preset number of columns.
- The All Columns View displays the maximum available columns.

Adding and Removing Table Columns

1. Click the Show/Hide Columns drop-down. (Figure 97)

A list of available columns appear.

Fig dro	gure 97 Show/Hide Columns op-down							
Show	//Hide Columns 🔻							
\checkmark	probeset_id							
	affy_snp_id							
\checkmark	CR							
\checkmark	FLD							
\checkmark	HomFLD							
\checkmark	HetSO							
\checkmark	HomRO							
	nMinorAllele							
\checkmark	Nclus							
	n AA							

 Click the check box next to the column(s) you want to add (Show) on the table. Click to uncheck a column you want to remove (Hide) from the table. Mouse over the menu's down arrow to reveal more available column choices. See Table 15 on page 162 for column definitions.



Selecting Annotations

1. Click the **Select Annotation** button.

The following window appears. (Figure 98)

Select Annotations	*
Annotation File:	
PharmacoScan.na35.annot.db	▼ Browse
Select Apportation Column(s) to Add	
✓ affy_snp_id_annot	
Chromosome	
Chromosome Start	
Chromosome Stop	
Strand	
Strand Versus dbSNP	
Probe Count	
Cytoband	
ChrX pseudo-autosomal region	
Flank	
Allele A	=
Allele B	
Ref Allele	
Alt Allele	
Associated Gene	
Genetic Map	
Microsatellite	
Heterozygous Allele Frequencies	
INUMBER OF INDIVIDUAIS	
Miner Allele	
Minor Allele Frequency	
Biomedical	
Annotation Notes	
Ordered Alleles	
Allele Count	-

- 2. Select the appropriate Annotation File from the drop-down menu list.
- Click the check box next to the Annotation Column(s) you want to add to the table or click the Check/Uncheck All check box (Figure 98) to add or remove ALL available annotations. See "Annotations and Columns" on page 159 for each Annotation's definition.
- 4. Click OK.

Your selected annotation columns are now added to the right side of the SNP Summary Table.

Saving your Table Column View

Row(s)

1. After you have your preferred SNP Summary Table columns set, click Save View.

The following window appears: (Figure 99)

Figure 99 Save New Custom View
Save Current View
Custom View Name:
OK Cancel

2. Enter a name, then click OK.

Your custom table view is now saved and stored inside the Apply View menu for future use.

Copying Selected 1. Right-click on a row you want to copy. Optional: Single-click on a row or Ctrl click, Shift click, or press Ctrl A (to select multiple rows).

A menu appears. (Figure 100)

Figure 10	00 Righ	t-clic	k me	enu				
AX-11086538	100 59.70	L 31.343	8.955	0.246	0.202	PolyHi	ighResolution	
AX-11086545	100 35.82	L 50.746	13 433	0 388	0 575	PolyHi	abResolution	
AX-11086572	100 32.83	5 28.358	3	Copy Select	ed Row	(s)		
AX-11086574	100 8.95	5 17.91	7.	Copy Select	ed Cell((s)		1
AX-11086577	100 44.77	5 41.791	1		LOND		10.1.1.	Ī
AX-11086578	100 22.38	3 25.373	5	Add Selecte	d SINP(s	s) to SN	IP List	1
AX-11086580	100 8.95	5 32.836	5	Remove Sele	ected S	NP(s) f	rom SNP List	1
AV 11096501	100 47 76	25 021	16 777	0.2421	0.0021		abkarolution	

2. Click Copy Selected Row(s).

The row data is now ready for pasting (Ctrl v).

Copying Selected Cell(s)	 Right-click on a cell you want to copy. Optional: Single-click on a row or Ctrl click, Shift click, or press Ctrl A (to select multiple cells).
	A menu appears. (Figure 100)
	2. Click Copy Selected Cell(s).
	The cell data is now ready for pasting (Ctrl v).



Changing or Reverting Genotype Calls Reverting Calls changes them back to what they were originally called by the algorithm. No other history is saved, only its current and original values.

1. Click the **Change/Revert Calls** drop-down.

The following menu appears: (Figure 101)

Figure 101 Revert Calls drop-down menu
Change/Revert Calls
Change Calls By Text File
Revert Selected SNP(s) for All Samples
Revert All SNPs for All Samples

Changing Genotype Calls by Text File

- 1. Click Change Calls By Text File.
 - An Explorer window appears.
- 2. Navigate to the text file's location.

IMPORTANT! Your Change Call by Text file must start with the header *probeset_id* and use numeric or alphabetic codes for the genotype, as shown in Figure 102. It also cannot contain any annotation columns.

F	igure 102	Change	e Call b	oy Text	file exa	ample s	shown i	n Excel	(as a t	ab-deli	imited t	ext file)			
	А	В	С	D	E	F	G	Н	I.	J	K	L	М	N	0	Р
1	probeset_id	2877_Axio	2878_Axi	2879_Axio	2880_Axio	2881_Axio	2882_Axio	2883_Axio	2884_Axic	2885_Axio	2887_Axio	2888_Axio	2889_Axio	2890_Axio	2891_Axio	2892_A
2	AX-11699985	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA
3	AX-11700086	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA
4	AX-11700405	BB	BB	AB	BB	AB	AA	BB	BB	BB	AB	BB	AA	BB	AA	AA
5	AX-11700418	AB	AB	BB	BB	AB	AB	AB	AB	BB	BB	AB	BB	AB	BB	BB
6	AX-11700675	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB
7	AX-11700679	AB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB	BB
8	AX-11700939	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA

3. Click Open.

Your Genotype Calls are now changed and reflected in the Cluster Plot.

Reverting Selected SNPs for all Samples

- 1. Single-click on a SNP file or Ctrl click, Shift click, or press Ctrl A (to select multiple files) within the SNP Sample Table.
- 2. Click Revert Selected SNP(s) for All Samples.

Reverting all SNPs for all Samples

Use this feature to perform a master SNPs reset.

1. Click Revert All SNPs for All Samples.

IMPORTANT! Once *Revert Calls* is performed, the selected calls will be reverted to original calls. This cannot be undone.

Ŕ

Reanalyzing your SNP Summary Table Data

The **Reanalyze** drop-down menu (Figure 103) offers optional steps for post-processing SNP data. These functions utilize the output files from the previous genotyping and classification steps as input.

Reanalyze 🔻	Figur menu
	Reanaly
Regenerate SNP Metrics	Re
Run OTV Caller	Ru

Regenerate SNP Metrics

The **Regenerate SNP Metrics** operation allows you to recalculate SNP Metrics and ConversionType classifications, which you may want to do if you have edited any genotype calls, or if you want to change SNP QC thresholds, or if you want to generate additional metrics and classifications.

1. Click Regenerate SNP Metrics.

The following window appears: (Figure 104)

Figure 104 F window	Regenerat	e SNP N	/letrics		
Regenerate SNP Metri	5		×		
Select Biallelic Posterio	r File:				
Salast Multiallalis Past	vriar Filos		Browse		
Select Multialienc Poste	enor rile:		▼ Browse		
Select ps2snp File (Rec	ommended):				
			Browse Clear		
Generate advanced n	netrics 📃 Run P	S Supplement	al		
Select Threshold Con	igurations				
Human (Default)		Restore	Save Save As		
SNP QC					
	ental				
		ОК	Cancel		

1. Use the drop-down to select the appropriate Posterior File or click its **Browse** button.

An Explorer window appears.

- 2. Navigate to Posterior File you want to use, then click Open.
- 3. Choose an appropriate ps2snp File (recommended), as described in "Using the Analysis Settings Fields" on page 24.
- 4. (Optional) Click the **Generate advanced metrics** check box to calculate additional SNP-specific posterior metrics.
- 5. (Optional) Click the **Run PS Supplemental** check box to generate additional metrics and probeset ConversionTypes. Note: PS Supplemental performs further classification that may be needed for polyploid organisms, complex genomes, or inbred populations.
- 6. Select the Threshold Configuration you want to use, as described in "Customizing Thresholds" on page 28.
- 7. Click to expand and use the **SNP QC** drop-down menu selections and text fields to setup the regeneration of your SNP Metric.
- 8. To change the priority-order of the various assigned ConversionTypes, go to the SNP QC setting's priority-order column and click the Change List Order button.

Cł	hange the Priority Order					
Click on a name, then drag and drop it to its new position. After your list is set, click OK.						
	Name					
	PolyHighResolution					
NoMinorHom						
	OTV					
	MonoHighResolution					
	CallRateBelowThreshold					
	OK Cancel					

The following window appears: (Figure 105)

- **9**. Click and hold onto the selection you want to move, then drag and drop it into its new position. After you get the order of priority you want, click **OK**.
- 10. To change the recommended options, click Checklist.

The following window appears: (Figure 106)

Figure 106 Recommended window				
recommended X				
PolyHighResolution				
VoMinorHom				
OTV				
MonoHighResolution				
CallRateBelowThreshold				
OK Cancel				

11. Click to check/uncheck the available recommended options. Click a field's **1** button to return its value back to its default setting.

Note: If you have checked **Run PS Supplemental** check box, then the associated threshold configurations are relevant. Edit them as needed.

12. Click OK.

After the process is complete, your SNP Summary Table is updated.

Running OTV Caller The OTV Caller is intended for SNPs that have been classified as likely having offtarget variants (OTV), or for SNPs with unusually large Y-dimension variance (as identified by PS Supplemental option of the Regenerate SNP Metrics).

OTV Caller function performs post-processing analysis to identify miscalled clustering and identify which samples should be in the OTV cluster and which samples should remain in the AA, AB, or BB clusters. Samples in the OTV cluster are re-labeled as OTV.

1. Click Run OTV Caller.

The following window appears: (Figure 107)

Figure 107	Run OTV (Caller win	dow
Run OTV Caller			×
Select Posterior File	:		
Default			▼ Browse
Select Pid File:			
			Browse
		ОК	Cancel

1. Use the drop-down to select the appropriate Posterior File or click its **Browse** button.

An Explorer window appears.

- 2. Navigate to Posterior File you want to use, then click Open.
- **3**. Click the Select Pid File's **Browse** button. An Explorer window appears.
- 4. Navigate to Pid File you want to use, then click **Open**.
- 5. Click OK.



Managing your SNP List

Axiom Analysis Suite enables lists of SNPs to be saved within the application.

1. Click the Manage SNP List drop-down menu (Figure 108), then select the option you want.

Note: The Recommended SNP List is auto-generated and updated whenever SNP metrics are calculated. Avoid using this reserved (default) SNP List name for your custom lists.

Figure 108 Manage SNP List drop-down menu				
Manage SNP List				
Create SNP List from Table				
Export Saved SNP List to Text File				
Import SNP List to Batch				

Saving your Current SNP List

1. To save all SNPs currently displayed in the SNP Summary Table, click **Create SNP List from Table**.

The following window appears: (Figure 109)

Save SNP
×
-
Cancel

2. Enter a name, then click **OK**.

Exporting your SNP List

Before exporting a SNP List you must first create one. If no SNP Lists are detected, a message box appears. Click **OK** to acknowledge the message, then go to "Saving your Current SNP List" to create a SNP List.

1. Click Export Saved SNP List to Text File.

An Explorer window appears.

2. Navigate to an export location, enter a name, then click Save.

Importing a SNP List 1. Click Import SNP List to Batch.

An Explorer window appears.

2. Navigate to your SNP List location containing your tab-delimited text file.

Your first row/column header must be labeled **probeset_id**, as shown in Figure 110, otherwise an error message appears.

Figure 110 Required probeset_id column header						
	А	В				
1	probeset_	id 🧹				
2	AX-85040799					
3	AX-85040886					
4	AX-850409	932				
5	AX-85041189					
6	AX-85041517					
7	AX-850415	551				

3. Click Open.

Your imported SNP List now appears in the SNP Summary Table.

Using your Saved SNP List

Displaying SNPs in a SNP list

- 1. Click to select the probeset_id column, then right-click on it.
- 2. Click Filter.

The Filter window appears. (Figure 111)

Fig	jure 111	Filter window		
probe	eset_id		×	
9	In SNP List	•	• 3	
		ОК	Cancel	

3. Add your previously saved SNP List by selecting it from the drop-down list. (Figure 112)



4. Click OK.

Only the SNPs in your SNP List are displayed in the SNP Summary Table.

Displaying SNPs that are not in your SNP List

- 1. Click to select the **probeset_id** column, then right-click on it.
- 2. Click Filter.
- 3. Click the In SNP List drop-down, then click to select Not in SNP List. (Figure 113)
- 4. Click the Saved SNP List drop-down, select your saved SNP List, then click OK.



Your SNPs from the SNP List are no longer displayed in the SNP Summary Table.



Cluster Plot

The Cluster Plot displays the SNP calls for selected samples as a set of points in the clustering space used for making the calls. A visual inspection of select Cluster Plots aids in identifying problematic SNPs and enables you to manually change calls.

As shown in Figure 114, use the **Type** option above the plot to switch between **Signal** view (left plot), **Log2 Signal** view (not shown), and **Contrast** view (right plot).

Note: For bi-allele probesets (those that measure two alleles), only the **Contrast** view displays the ellipses that illustrate the prior and posterior knowledge of genotype cluster positions.



Multi-allele probesets measure more than two alleles. Unlike bi-allele probesets, multiallele probesets are genotyped in the Log2 Signal space. For multi-allele probesets, the ellipses that illustrate the prior and posterior knowledge of genotype cluster positions are only displayed when selecting plot Type **Log2 Signal**. See Figure 115 on page 90 for plot examples of a multi-allele probeset.



Note: Multi-allele probesets in the SNP Summary Table can be more easily identified by adding the annotation field Allele Count and/or Ordered Alleles. If a probeset has Allele_Count of 3 or greater, it is a multi-allele probeset.

Multi-allele probesets measure more than two alleles, and so more than two allele signal channels are present. The cluster plot only shows data for two allele signal channels. If multi-allele probesets are present in the current results, then the top of the cluster plot will also display the Draw By menu.

See Figure 116 for an example of using Draw By to select different pairs of allele signal channels for plotting. The default Draw By option is selected based on the probeset's most common genotype allele calls in the data set.



Using the Cluster Plot

Displaying a SNP Cluster Plot that Corresponds with a SNP

1. In the SNP Summary Table, click on row (SNP) of interest.

Note: Use the arrow keys on the keyboard to toggle through the list. As you toggle through the list, the Cluster Plot auto-updates to match your selected SNP.

2. Click the **Color By** and **Shape By** drop-down menus to select the combination (X and Y axis) view you want. See Table 12 on page 154 for Color By and Shape By definitions. Note: Your imported sample attributes are also available for use within the Color By and Shape By drop-down menus.



The appropriate legend appears within the plot. (Figure 117)

Setting New Scale Setting Ranges

1. Click Scale Settings.

The following window appears. (Figure 118)

Figure 118 window	Scale Settings
Scale Settings	*
Auto Scale	Max Vi 1
Min Y: 95	Max Y: 100
	Default
	OK Cancel

By default, the window displays your current range values.

- 2. Enter your new ranges in the appropriate text fields.
- 3. Click OK.

Click Default to return all values back to their factory settings.

Customizing Color By Settings 1. Click Configure.

The Color Scale Configuration window appears. (Figure 119)

Figure 119 Color By options							
Color Scale Con	figuration	×					
28.941	Auto Scale Cutoff Type: Above Cutoff Failing 28.941 Cutoff: 26.125 Min:	Above Cutoff Failing Above Cutoff Failing Below Cutoff Failing No Cutoff					
23.309	23.309	Ō					
	OK Cancel						

- 2. Use the provided text fields and color drop-down menus to customize your Color By selection.
- Auto Scale check box (when checked) uses the actual minimum (lower bound) and maximum (upper bound) as your min/max scale. Uncheck the Auto Scale check box to enter your min and max number scales in the provided fields. Note: If the Auto Scale check box remains unchecked, you must enter new scale limits for each subsequent analysis.
- Click the Cutoff Type drop-down menu to select your cutoff preference.
 - Above Cutoff Failing This presents a hard visual cutoff graph of all values that fail ABOVE the Cutoff value entered. The Above Cutoff data is represented by the color defined for Max. (Green in Figure 119)

 Below Cutoff Failing - This presents a hard visual cutoff graph of all values that fail BELOW the Cutoff value entered. The Below Cutoff data is represented by the color defined for Min. (Red in Figure 120)

Figure 120 Below Cutoff					
Color Scale Conf	figuration 🗙				
28.941 26.125 23.309	✓ Auto Scale Cutoff Type: Below Cutoff Failing ▼ Max: 28.941 26.125 ✓ Min: 23.309				
	Default				
L	OK Cancel				

 No Cutoff - This presents a smooth 3-point gradient of your defined Max, Min, and colors. (Figure 121).

Figure 121 No Cutoff				
Color Scale Cor	nfiguration	×		
28.941	✓ Auto Scale			
	No Cutoff	•		
26.125	28.941 Cutoff:			
	26.125			
23.309	23.309			
		Default		
	ОК	Cancel		

3. Click OK.

Your Cutoff preference, entered values, and color selections are now displayed on the graph and saved for future use. If needed, click the **Default** button to revert ALL values back to their factory setting.

Selecting Multiple Samples in a Cluster Plot

1. Drag the cursor around a group of samples to draw a pink-dotted closed loop around them, as shown in Figure 122.



2. Release the mouse button to select the group of lassoed samples. (Figure 123)



Changing a Sample's Call for a Single SNP

- 1. Highlight the sample or samples you want to modify, then right-click on them. A menu appears.
- 2. Click **Change Call**, then move your cursor to the right, then click to select a different call, an OTV (Off Target Variant), or No Call. (Figure 124) Note: If the array supports CN-aware Genotyping, additional Call Code choices appear.

Fig	gure 124	Chang	je C	all menu
ß	Copy ID(s)			
AAAAB	Change Call	•		AA
	Revert Call			AB
_				BB
				NoCall
				ΟΤV

The Call is now changed, but not the position. The image may or may not change, as it depends on the Color By and Shape By options you selected.

Reverting a Single Call

- Single-click to highlight the Call you want to revert back, then right-click on it. A menu appears.
- 2. Click Revert Call.

Reverting Multiple Calls

- 1. Drag the cursor around a group of samples to draw a pink-dotted closed lasso shape around them.
- 2. Release the mouse button to highlight your selected samples.
- 3. Click the **Revert Call** drop-down menu (Figure 125), then click to select **Revert Selected Call(s)**.

Figure 125Revert Calls drop-down menu	
Revert Calls	
Revert Selected Call(s)	Ī
Revert Current SNP for All Samples	



Displaying Cluster Model Data

By default, the **Prior**, **Posterior**, and **Special SNPs** drop-down selections are preset to best suit the currently displayed Cluster Plot.

1. Click the appropriate drop-down menu (Figure 126), then click to select a new setting.

Figure 126 Bi-Allele Cluste	er Model Data drop-down menus
Prior: Axiom_GW_Hu Posterior:	Default V Special SNPs: Axiom V
	Default
	Browse None
	None

- If you select **Browse**, an Explorer window appears. Navigate to your folder location as you normally would, then click **Open** to display your data within the graph.
- Selecting **None**, conceals (hides) the selected graph data.

Saving the Current Cluster Plot View

- Click the Save Image button.
 An Explorer window appears.
- 2. Navigate to where you want to save the .PNG file, enter a filename, then click **OK**.



Allele Translation

About Translations

For supported array types (e.g. PharmacoScan), Axiom Analysis Suite will provide the option to convert (translate) the genotype calls of an important subset of SNPs to functional allele calls using standardized nomenclature wherever possible. The software enables you to:

- Quickly identify possible rare alleles or missing data.
- Identify haplotype and SNP-level sequence variation in the test samples.
- Annotate the reported genotypes across translated SNPs to indicate genomic, mRNA, or peptide changes resulting from any observed variation.
- Predict general gene activity based on detected diplotypes.

See Appendix B, "About Allele Translation" on page 139 for more information.

Performing Allele Translation

The Perform Allele Translation operation is available only if the following conditions are met:

- The library folder must include the same library package (Array Type with revision number) used to generate the batch results.
- Inside the matched library package folder, the files with the extensions *.dc_annot.csv and *.translation must exist.

To perform an allele translation:

1. From an open batch in the Viewer, navigate to the SNP Summary Table, then click the **Perform Allele Translation** button, as shown in Figure 127.

Figure 127 SNP Summary Table window										
Summary Sample Table SNP Summary Table CN Summary Table Cluster Plot CN Region Plot										
Select Annotation	Select Annotation Manage SNP List V Change/Revert Calls V Reanalyze V Perform Allele Translation									
Apply View 🔻 S	Apply View 🔻 Save View Show/Hide Columns 👻 Export 🔻 Filters 👻									
probeset_id 🛧	ConversionType	CR	MinorAlleleFrequency	H.W.p- Value	FLD	HomFLD	HetSO	HomRO	Nclus	n_AA
AFFX-KIT-000001	PolyHighResolution	100	0.422	1	18.257	39.072	0.189	2.692	3	
AFFX-KIT-000002	PolyHighResolution	100	0.219	1	10.347	20.808	0.261	1.038	3	
AFFX-KIT-000003	PolyHighResolution	100	0.281	0.004	6.838	16.162	0.295	1.094	3	
AFFX-KIT-000004	PolyHighResolution	100	0.234	0.652	12.001	26.576	0.221	2.22	3	
AFFX-KIT-000005	NoMinorHom	100	0.062	1	5.846		0.123	0.87	2	
P Find in Table Row Count: 129544 Selected: 1										

The Perform Allele Translation window appears. (Figure 128)

Figure 128 Perform Allele Translation window
Perform Allele Translation
Input Files Annotation File: Translation File:
Metabolizer File:
Options
SNP List Filter:
If filtering removes markers needed to differentiate among multiple possible haplotypes Report only the first named haplotype in the Translation File Report combined name that includes all haplotypes that are no longer differentiated. Include Sample Attributes?
Select Translation Results Folder
* Output Root Path: C:\Users\Public\Documents\AxiomAnalysisSuite\Export\
* Export Folder Name: 2016-07-19-035442
OK Cancel

2. At the Perform Allele Translation window, use the provided drop-down menus to select an **Annotation File**, **Translation File**, and an optional **Metabolizer File**. Note: If you disagree with the phenotype interpretations, you may want to leave the Metabolizer File option blank. (Figure 128) A Phenotype report will not be created if this option is not used.

Allele Translation	Refer to Table 1 and Table 2 for descriptions of the available Allele Translation
Options	options.

Select Options	Description
SNP List Filter	Choose this option to translate only the genotypes of SNPs in a user-specified probeset list. Click the Browse button to select the marker list, or select from SNP lists you're already imported to the batch results. WARNING: If you supplied a custom optional SNP List File for Genotyping in Analysis Settings when setting up your analysis, you should either filter on the same marker list for Allele Translation, or filter on an even more restrictive list. Otherwise, you may see SNPs with NotAvailable calls in your translation reports. The presence of NotAvailable calls may increase the number of reported haplotype possibilities.
Report only the first named haplotype in the translation file (Default)	This option is only relevant IF you filter by a SNP List AND if the SNP List contains some (but not all) of the available probesets in a gene that is allele translated. If both conditions are met, then it is possible that your SNP List excludes a probeset needed to differentiate among two or more named haplotypes in the translation library file. Selecting this option means that only the first haplotype will be reported from the set of possible haplotypes that are non-distinguishable due to probeset exclusion. The haplotypes are ordered by name from left to right in the translation library file. For example, in gene CYP1A1, the *2C haplotype may be differentiated from the *1 haplotype by a variation in a single probeset. If this probeset is omitted, and the data indicates that both *1 and *2C are possible (due to a NoCall at that probeset), then only *1 is reported as a possibility (since *1 is listed before *2C in the translation library file). The disadvantage of selecting this option is that you may be excluding the actual haplotype for a tested sample. The advantage of selecting this option is that you may be colded not to translate, AND you agree that the selection of which haplotype to report is correct.

 Table 1
 Allele Translation options

Table I Allele Hallslation options	Table 1	Allele	Translation	options
------------------------------------	---------	--------	-------------	---------

Select Options	Description
Report combined name that includes all haplotypes that are no longer differentiated	This option is only relevant IF you filter by a SNP List AND if the SNP List contains some (but not all) of the available probesets in a gene that is allele translated. If both conditions are met, then it is possible that your SNP List excludes a probeset needed to differentiate among two or more named haplotypes in the translation library file.
	Selecting this option means that a combined haplotype name will be reported using the set of possible haplotypes that are non-distinguishable due to probeset exclusion.
	For example, in gene CYP1A1, the *2C haplotype may be differentiated from the *1 haplotype by a variation in a single probeset. If this probeset is omitted, and the data indicates that both *1 and *2C are possible (due to a NoCall at that probeset), then "*1_or_*2C" is reported as a possibility.
	The advantage of selecting this option is that you are not excluding possible haplotypes. The disadvantage of selecting this option is that the report will include haplotypes that require a variant allele of a probeset you have decided to exclude for translation.
	Note: This option is only available if you do not need a phenotype report, as phenotyping requires haplotype names to not change (depending on the set of probesets used for translation). To enable this option that excludes the generation of a phenotype report, you must deselect the usage of the metabolizer library file. To do this, click (right of the Metabolizer File option).
Include Sample Attributes	Click this check box to include sample attributes in the translation reports. This option is enabled if you have imported sample attributes to your analysis results.

Table 2	More Allele	Translation	options
---------	-------------	-------------	---------

Select Translation Results Folder	Description
Output Root Path	The path to the output folder for the translation reports and log. Click its Browse button to set the path.
Export Folder Name	The new folder name for the translation results, whose default name has the date-time format YYYY-MM-DD_HHMMSS_translations. Confirm the default or enter a new folder name.

3. After completing the Allele Translation selections, click **OK**.

When the translation is finished, an Explorer window appears displaying the folder containing the translation reports and corresponding run log.

Translation Reports

The allele translation operation creates the following reports and supporting files:

- **Comprehensive Translation** Displays one row per translated SNP for each sample. Provides information on each SNP in addition to haplotype calls.
- Summary Translation An abbreviated version of the Comprehensive report, which displays at least one row for every translated gene for each sample. It also includes rows for every genotype where the translation identifies a variant call. It also includes rows listing SNPs with missing data. In the Summary report only, if no SNPs responsible for functional changes report a variant allele, then information for those SNPs is replaced with a comment to this effect. If a copy number state of zero is indicated, then information for SNPs in that gene is replaced with a comment to this effect, and the copy number haplotype code is reported in the Known Call field.
- **Phenotype Translation** Displays one row per phenotyped gene for each sample, based on the diplotypes from the source Comprehensive Translation report
- Uncalled probeset list A list of probesets with NoCall genotype calls from SNPs used for translation. This probeset list can then be importing into the Batch, so that you can filter the SNP Summary Table by the uncalled.ps SNP list. This provides a quick way to review the cluster plots of probesets with missing data, and possibly edit the calls directly to "fill in" the missing data in preparation for another round of allele translation reports.
- **Uncalled** A list of NoCall genotype calls from SNPs used for translation. This report is useful for identifying samples and SNPs for follow up genotyping.
- **MD5** An electronic signature that can be used to verify that the comprehensive and phenotype reports have not been modified. Interested users can contact devnet@affymetrix.com for information on accessing tools to verify the integrity of the translation results files.
- Log file A list of messages generated by the software as the data is processed. This file is useful for troubleshooting errors.

Comprehensive and Summary Translation Report

The basic layout of this report is shown in Figure 129.

Figure 129 Example: Basic layout of a Comprehensive and Summary Translation report								
			Marker Info					
	Gana ID	Hanlatuna Info	Marker Info					
	Gene iD	парютуре ппо	Marker Info					
Ð			Marker Info					
E E			Marker Info					
ne l	Gene ID	Hanlotyne Info	Marker Info	Sample Info				
≥	Celle ID	парютуре ппо	Marker Info					
LI			Marker Info					
a			Marker Info					
	Cono ID	Hanlotyne Info	Marker Info					
Gene iD	r aplotype into	Marker Info						
			Marker Info					
	Gene ID	Gene ID		Marker Info				
			Gene ID	Gene ID	Gene ID	Gene ID	Gene ID	Hanlotype Info
			Marker Info					
ē			Marker Info					
E			Marker Info					
ů	Gene ID	Haplotype Info	Marker Info	Sample Info				
ay			Marker Info					
			Marker Info					
a l	a		Marker Info					
	Gene ID	Haplotype Info	Marker Info					
	Gene ID		Marker Info					
			Marker Info					

Summary Translation Report

To make this report easier to read, bold and regular fonts are used. For example, genotype calls are represented in bold, as shown in Figure 130.

Fig	Figure 130 Example: Summary Translation report														
	🗐 2008-11-05_122550_translations _summary.rpt 📃 🚍														
	СНР			Unknown	Interpretat	Summaru	Relevant			Probe Set		Reference	Variant		Haploty
Index	Filename	Gene	Known Call	Call	ion Code	Flag	Alleles	Common Name		ID	Basecall	Base	Base	Call	Marker
0001-	TEST_000	1. SLC15A2	14A/14A		UNIQ	R503K	2, 4A, 4B	SLC15A2_34845	5A>G(R503)	AM_13307	A/A	G	A	Yar/Yar	Y
0001-	TEST_000	LUGT1A9	22/VT		UNIQ	*22-	*22	UGT1A9*22_inst	T-118	AM_12947	-п		Т	Ref/¥ar	Y
0001-	TEST_000	I. UGT1A9	*22/WT		UNIQ	A7S	A7S	UGT1A9_(rs675:	9892)	AM_12969	T/G	т	G	Ref/¥ar	N
0001-	TEST_000	1. UGT2B7	"1/"2E,"2/"5	"2B/UNK,	MULT+UNK	*2B	2B, 2E, 5	UGT2B7*2B3;	27>(rs7662(AM_13458	G/A	G	A	Ref/¥ar	Y
0001-	TEST_000	1. UGT2B7	"1/"2E,"2/"5	"2B/UNK,	MULT+UNK	*2A	2A, 2C, 2	UGT2B7*2A16	51>(rs76682	AM_13459	сл	С	т	Ref/¥ar	Y
0001-	TEST_000	1. UGT2B7	"1/"2E,"2/"5	"2B/UNK,	MULT+UNK	*2	2, 2B, 2D	UGT2B7*2_2100	DC>T(Y268I	AM_13465	сл	С	т	Ref/¥ar	Y
0001-	TEST_000	I. VKORC1	B2/B2		UNIQ	Q30R	Q30R	VKORC1_1120A:	>G(Q30R)	AM_11032	A/A	G	A	Yar/Yar	N
0001-	TEST_000	I. VKORC1	B2/B2		UNIQ	B-Hap	B1,B2,B3,E	VKORC1_2255(r	s2359612)	AM_11040	C/C	т	С	Yar/Yar	Y
0001-	TEST_000	I. VKORC1	B2/B2		UNIQ	B-Hap	B1,B2,B3,E	VKORC1_1173(rs	;9934438)	AM_11045	C/C	т	С	Yar/Yar	Y
0001-	TEST_000	I. VKORC1	B2/B2		UNIQ	B-Hap	B1,B2,B3,E	VKORC1_(rs992	23231)	AM_11054	G/G	A	G	Yar/Yar	Y
0001-	TEST_000	I. DCK			NoHAP	P1228	P122S	DCK_(P122S)		AM_13645	сл	С	т	Ref/¥ar	N
0001-	TEST_000	I. DCK			NoHAP	35708A>G	35708A>G	DCK_35708>(rs-	4643786)	AM_13646	T/C	Т	С	Ref/¥ar	N
0001-	TEST_000	I. SLCO1A2			NoHAP	T277N	T277N	SLC01A2_>(T27	'7N)	AM_10528	C/C	A	С	Yar/Yar	N
0001-	TEST_000	I. UGT1A1		UNK/UNK	UNDH	*60-	*28var,*60	UGT1A1*60_(rs4	124874)	AM_13018	G/G	т	G	Yar/Yar	Y
0001-	TEST_000	I. UGT1A1		UNK/UNK	UNDH	*28-	*28,*28var	UGT1A1*28_(rs3	84815109)	AM_13024	Stof(AT)	(TA)Sor6	(TA)?or	Yar/Yar	Y
0001-	TEST_000	1. UGT2B15			NoHAP	' 2	' 2	UGT2B15*2_>(rs	;1902023)	AM_13439	G/T	G	Т	Ref/¥ar	N
0001-	TEST_000	1. ABCG2	1811		UNIQ	All marke	ers respon	sible for func	tional cha	ages are R	ef/Ref				
0001-	TEST_000	I. CDA	"1C/"3	*17UNK	UNIQ+UNK	All marke	ers respon	sible for func	tional cha	ages are R	ef/Ref				
0001-	TEST_000	1. CES2	11/1		UNIQ	All marke	ers respo	sible for func	tional cha	nges are R	ef/Ref				
0001-	TEST_000	 CYP2A13 	114/114		UNIQ	All marke	ers respon	sible for func	tional cha	nges are A	ef/Ref				
0001-	TEST_000	I. CYP2A6	*1G/*1G		UNIQ	All marke	ers respon	sible for func	tional cha	ages are R	ef/Ref				
0001-	TEST_000	I. CYP2J2	11/1		UNIQ	All marke	ers respon	sible for func	tional cha	ages are R	ef/Ref				
0001-	TEST_000	I. FAAH			NoHAP	All marke	ers respon	sible for func	tional cha	ages are R	ef/Ref				
0001-	TEST_000	1. FM03	H1/H2A,H2E	37H7	MULT	All marke	ers respon	sible for func	tional cha	ages are R	ef/Ref				

Phenotype Translation Report

The basic layout of this report is shown in Figure 131.

Figure 131 Example: Phenotype Translation report

e	Gene ID	Phenotype Info	Haplotype Info	
E I	Gene ID	Phenotype Info	Haplotype Info	Convolo Info
È.	Gene ID	Phenotype Info	Haplotype Info	Sample Into
Le	Gene ID	Phenotype Info	Haplotype Info	
e	Gene ID	Phenotype Info	Haplotype Info	
l la	Gene ID	Phenotype Info	Haplotype Info	Convolo Info
λe	Gene ID	Phenotype Info	Haplotype Info	Sample Into
E .	Gene ID	Phenotype Info	Haplotype Info	

Phenotype Report

The basic layout of this report is shown in Figure 132.

Figure 132 Example: Phenotype report							
Index	CHP File (Gene	Phenotype Call	Gene Activity	Known Call	Unknown Call	Interpretation Code
0001-0020	test_01.c	CYP1A2	EM	normal/normal	*1F/*1F		UNIQ
0001-0022	test_01.c	CYP2A6	EM	normal/normal	•1/•1		UNIQ
0001-0024	test_01.c	CYP2B6	EM	normal/normal	* 1/ * 1		UNIQ
0001-0029	test_01.c	CYP2D6	PM	none/none	* 5/ * 5		UNIQ
0002-0020	test_02.c	CYP1A2	EM_or_IM	normal/reduced	*1A/*1L,*1C/*1F		MULT
0002-0022	test_02.c	CYP2A6	EM_or_IM	normal/reduced	* 1/ * 17		UNIQ
0002-0024	test_02.c	CYP2B6	EM_or_IM	normal/reduced	* 1/ * 6	*4/UNK	UNIQ+UNK
0002-0029	test_02.c	CYP2D6	EM_or_IM	normal/reduced	*2/*29	*2/UNK,*29/UNK	NC/PRA/NA
0004-0020	test_04.c	CYP1A2	EM	normal/normal	*1A/*1A		UNIQ
0004-0022	test_04.c	CYP2A6	EM	normal/normal	•1/•1		UNIQ
0004-0024	test_04.c	CYP2B6	IM	normal/none	* 1/ * 18		UNIQ
0004-0029	test_04.c	CYP2D6	EM	normal/normal	* 1/ * 1		UNIQ
0005-0020	test_05.c	CYP1A2	EM	normal/normal	*1A/*1F		UNIQ
0005-0022	test_05.c	CYP2A6	EM	normal/normal	* 1/ * 1		UNIQ
0005-0024	test_05.c	CYP2B6	IM	reduced/reduced	*6/*6		UNIQ
0005-0029	test_05.c	CYP2D6	IM	normal/none	* 2/ * 4	*1/UNK,*2/UNK,*4/	NC/PRA/NA
0009-0020	test_09.c	CYP1A2	EM	normal/normal	*1F/*1F		UNIQ
0009-0022	test_09.c	CYP2A6	EM	normal/normal	•1/•1		UNIQ
0009-0024	test_09.c	CYP2B6	IM	reduced/reduced	*6/*6		UNIQ
0009-0029	test_09.c	CYP2D6	IM	normal/none	*2/*4	*1/UNK,*10/UNK,UN	UNIQ+UNK

Opening Translation Report in MS Excel

- 1. Use Windows Explorer as you normally would to navigate to the export folder with the translation results.
- 2. Double-click the report (.rpt) to be viewed. You may be asked choose an application to open the report. Select Microsoft Excel, then step through the Text Import Wizard (use the tab-delimited default options).

The report header includes basic information that helps track study data and definitions of interpretation codes, as shown in Figure 133.

Fi	gure 133	Example: H	leader	for Comp	orehensive	and Summary	report		
	🕙 2008-11-05_122550_translations TRAINING r5_summary.rpt [Read-Only]								
	А	В	С	D	E	F	G	Н	
1	# For research u	use only. Not for a	liagnostic	purposes.					
2	#%dmet3-repo	rt-guid=00000467	34-122591	6774-00000194	453-0000014019-	0000029057			
3	#%Program=ap	t-dmet-translatio	n						
4	#%Version=1.0								
5	#%Date=Wed N	Jov 05 12:26:14 20	08						
6	#%TranslationF	ile=C:\Program Fi	les\Affyn	netrix\DMET C	onsole Library b	233 r5\DMET_Plus.200	81105.translatic	n	
7	#%MarkerList=								
8	#%HaplotypeR	eportOption=							
9	#%GenotypeOv	verrideFile=							
10	#Interpretation	n Code Description	n:						
11	#UNIQ: Unique	haplotype pair							
12	#MULT: Multipl	e haplotype pairs	possible	due to phase a	ambiguity				
13	#UNIQ+UNK: UI	nique annotated ł	naplotype	pair, with oth	er haplotype pa	irs requiring unannota	ited haplotypes	also possible	
14	14 #MULT+UNK: Multiple annotated haplotype calls possible, with other haplotype pairs requiring unannotated haplotypes also possible								
15	15 #UNDH: Only haplotype pairs with undefined haplotypes possible								
16	16 #NC/PRA/NotAvailable: NoCall, PossibleRareAllele, or NotAvailable call for one or more markers resulting in multiple haplotype pairs								
17	#NoHAP: No ha	plotypes defined	at this ge	ne					
18	Index	CHP Filename	Gene	Known Call	Unknown Call	Interpretation Code	Summary Flag	Relevant Alleles	Comm
10	0001 0002 02 ▶ ▶ 2008-11-	05_122550_transla	tions	*1A/*2			*1	*2 U1 Affo U2Affo	

Available Report Fields and Descriptions

Array Tracking

Refer to Table 3 for descriptions of the available Array Tracking fields.

Table 3 Array Tracking fields

Array Tracking fields	Description
Index	A row index in the format: [filename index]-[gene index within filename]-[Probe Set ID index within gene]. This field can be parsed for sorting or row filtering. For the phenotype report, the index is shortened to [filename index]-[gene index within filename]
Filename	Name of the sample file.

Gene-specific

Refer to Table 4 for descriptions of the available Gene-specific fields.

IMPORTANT! Haplotypes are not reported for genes whose Interpretation Code is NoHap.
 The fields described in the table below will therefore be empty for these genes. The exception is if the gene reports a gene deletion, in which case the associated haplotype names are reported.

Table 4 Gene-specific fields

Gene-specific fields	Description
Associated Gene	Gene symbol
Phenotype Call	In the Phenotype report, the predicted phenotype given the supplied Known Call diplotypes. Multiple comma-separated phenotypes are reported when multiple Known Call diplotypes are associated with different phenotypes. Most genes use the following terminology when the default metabolizer library file is selected: • UM = ultra-rapid metabolizer • RM = rapid metabolizer • NM = normal metabolizer • IM = intermediate metabolizer • PM= poor metabolizer
	 Variations on these terms also exist to describe some level of uncertainty: NM_or_IM = normal or intermediate metabolizer IM_or_PM = intermediate or poor metabolizer Not_PM = not a poor metabolizer unknown = unknown metabolizer state Some genes use different phenotype terms to be consistent with literature usage. Refer to the header of the phenotype report for additional information. Users are responsible for reviewing the *.metabolizer library file for accuracy! Users may modify the *.metabolizer file as needed, and are not restricted to this terminology. Refer to "Diplotype to Phenotype Translation" on page 144 for more information.

Table 4 Gene-specific fields

Gene-specific fields	Description
Gene Activity	In the Phenotype report, the predicted pair of gene activities given the supplied Known Call diplotypes. Multiple comma-separated activity pairs are reported when multiple Known Call diplotypes are associated with different activity pairs. Most genes use the following terminology when the default metabolizer library file is selected: • increased = increased gene function • normal = normal gene function • reduced: reduced gene function • no = no gene function • unknown = unknown or uncertain gene function
	Some genes use different phenotype terms to be consistent with literature usage. Users are responsible for reviewing the *.metabolizer library file for accuracy! Users may modify the *.metabolizer file as needed, and are not restricted to this terminology. Refer to "Diplotype to Phenotype Translation" on page 144 for more information.
Known Call	Haplotype pairs (diplotypes) identified in the gene of interest. When more than one pair of haplotypes is implicated (due to phase ambiguity in compound heterozygous samples), the reported diplotypes are separated by a comma.

Table 4	Gene-specific fields
Table 4	Gene-specific fields

Gene-specific fields	Description
Unknown Call	When the gene table includes haplotyping SNPs and a complete diplotype pair cannot be identified in a sample, one or more unknown haplotypes is assumed. This is designated as UNK in the report. Multiple haplotype pairs (diplotypes) that have unknown alleles are separated with a comma in this field. An example record might be in the format: *2/UNK,*13/UNK,*24/UNK,*32/UNK to indicate that there are at least 4 defined alleles consistent with the data, but each would require matching to a haplotype pattern that does not exist in the translation library file.
Interpretation Code	This diplotype interpretation code indicates whether one and only one unique haplotype pair is consistent with the data (UNIQ), whether there are multiple haplotype pairs consistent with the observed genotypes (MULT) and whether these are observed in conjunction with other unknown haplotypes (UNIQ+UNK or MULT+UNK). Additional codes indicate that no known haplotype pairs have been identified (UNDH) or if there is missing data leading to additional haplotype possibilities. The missing data could be NoCall or NotAvailable (NC/PRA/NA). The PossibleRareAllele call is not used by Axiom Analysis Suite.
	For genes for which copy number state is available, the following interpretation codes may also appear:
	• CN_HybridLoss = Partial gene deletion is detected, so haplotype pair calling is not available.
	• CN_HybridGain = Partial gene duplication is detected. If a non-wild-type allele is detected, the software can't determine whether the variant allele is on the partial copy of the gene.
	• CN_Gain = Gene duplication is detected, but the software cannot determine which haplotype(s) are duplicated. For this reason you won't see xN nomenclature in the reported haplotype pairs.
	• CN_NoCall = Copy Number state not reported, so there is less confidence in the reported haplotype pairs.
	• CN_Error = Genotypes from multiple Copy Number states are detected, so haplotype pair calling is not available. This can happen if you manually edit a genotype call in such a way as to change the SNP's copy number state. For example, if you change a "NoCall_1" genotype (a NoCall for a CN=1 sample) to a "BB" CN=2 genotype, its copy number has changed. This would cause allele translation problems if other SNPs for this sample for the same gene have genotypes of a different copy number state.

Refer to Table 5 for descriptions of the available Marker-specific fields.

IMPORTANT! Marker annotations will always be the same throughout the reports for a given SNP. Fields in the Marker Information section of the Comprehensive and Summary reports include the biological information at the SNP level, along with the interpreted genotypes identified in each sample

Table 5 Marker-specific fields

Marker-specific fields	Description
Summary Flag (marker annotation)	This annotation field contains an abbreviated name when structural or functional differences are known to result with mutations at the SNP locus. For triallelic SNPs, there may be more than one flag. For example, ABCB1_c.2677G>T>A(A893SorT) marker is triallelic and can result in different function changes in the protein. The two summary flags for the marker are thus reported "A893S,A893T".
	This flag is N (No) if the marker is not defining for a haplotype, and its genotype doesn't affect the structure or expression of the gene product. If the flag is N, the marker will not appear in the Summary report unless it reports NoCall.
Relevant Alleles (marker annotation)	This annotation field is the full listing of haplotype-based alleles defined in the gene table that contain the variant version of the marker. For non-haplotype-based SNPs, this is an abbreviated name indicating the protein change that results when the variant base is present.
Common Name (marker annotation)	A SNP identifier describing either the gene location, coding change or dbSNP rsID for the SNP. The Common Name is retrieved from the translation library file, and may not be the same as the Common Name seen within Axiom Analysis Suite (which instead uses the Common Name in the *.annot.db library file)
Probe Set ID	Unique identifier for the SNP.
Basecall	The observed bases, also known as the "raw" genotypes.
Reference Base	This field generally indicates the more common allele in biallelic SNPs. Certain genes use a particular GenBank entry as the "Reference genome" and the observed allele at each marker across the gene is then reported as Reference.
Variant Base	These are the alternate alleles for each SNP. When there is more than one variant allele (e.g. triallelic SNPs) the alternate alleles are reported together and separated by a comma (e.g. A,T).
Call	The first level of translation of the Basecall field, replacing the individual nucleotide calls with the associated reference (Ref) or variant (Var) allele state. For Basecalls associated with copy numbers less than 2, this field will show haplotype names as needed. This field will be empty if the Basecall value has an unclear call.
Haplotype Marker (marker annotation)	 Differentiates SNPs used to make haplotype calls or single-marker variant calls. Y = A flag to indicate that the Allele translation algorithm will match allele variants in blocks of SNPs defining haplotypes in the gene tables. Called haplotypes are reported in the "Known Calls" and Unknown Calls of the report. N = A flag to indicate that the haplotype background of a variant is not known. Genotyping results for these SNPs are only reported in the "Call" column.
Change for Variant (marker annotation)	Amino acid substitution or other structural change (such as splicing variant, promoter mutation, Frame shift mutation, etc.) caused by the presence of the variant allele.
Marker-specific fields	Description
--	--
cDNA Change (marker annotation)	Location of the mutation on a reference mRNA sequence.
Genome Position (marker annotation)	This is the chromosomal position of the mutation.
dbSNP RS ID (marker annotation)	The dbSNP identifier for the marker.

Table 5 Marker-specific fields

Tracking Edited Genotype Calls Refer to Table 6 for descriptions of the available fields for tracking edited genotype calls.

IMPORTANT! Fields for tracking genotyping changes are recorded in the Change Tracking portion of the translation reports (located immediately before the User Defined Sample Information section of the report).

Table 6 Fields for tracking edited genotype calls

Tracking Edited Genotype Calls	Description
Original Basecall	This field reports the original genotype for the sample. The field is not empty when the user has edited the call within Axiom Analysis Suite.
Override Comment	This field reports edited in AxAS if the call has been edited within Axiom Analysis Suite.

Uncalled Report

Refer to Table 7 for descriptions of the available fields for uncalled reports.

IMPORTANT! The Reference Allele and Variant Allele values of the Uncalled report are the same as the Reference Base and Variant Base values of the Comprehensive and Summary reports.

Table 7 Uncalled Report

Uncalled Report	Description
Filename	Name of the sample file.
Gene	Gene symbol.
Common Name	The Common Name defines the gene and positional information about the genetic change tested with the probeset.
Basecall	The Uncalled report contains all the NoCall genotypes from SNPs used for allele translation.
Override Comment	User-specified annotation field that enables an audit trail of the source of genotyping results done outside of Axiom Analysis Suite. In the Uncalled report, this field is primarily useful when creating a copy of this file to use as an Override file. The Override file may be useful for Affymetrix Power Tool users.
Reference Allele	Reference base indicates the allele in a reference sequence known to be present at this genetic location. Generally this is the more common allele at SNPs with low minor allele frequency (<1%).
Variant Allele	The variant base(s) defined by the marker are alternative known genotypes known to be present at this genetic location. For triallelic SNPs, the reporting format is: A,T for ABCB1_68883G>T(S893A) because two specific mutations are known to occur at this genomic location (G>A and G>T).

Exporting



Using the Sample Table Export Options

1. Click the **Export** drop-down.

Your Export options appear. (Figure 134)



- 2. Click **Export Current Table** or **Export All Data**. A **Save As** window appears.
- 3. Click on an existing folder or click New Folder to choose a new save location.
- Type a filename for the table, then click Save.
 The table data is now saved as a tab-delimited .txt file.

Using the SNP Summary Table Export Options

 Click the Export drop-down. The following window appears: (Figure 135)



6

Exporting the	To export the columns and rows currently displayed in the table:		
Current Table	1. Click Export Current Table.		
	An Explorer window appears.		
	2. Enter a filename, then click Save .		
	The current table data is now saved and exported as a tab-delimited text file.		
Exporting All Data	To export all columns and rows, including hidden and filtered data:		
	1. Click Export All Data.		
	An Explorer window appears.		
	2. Enter a filename, then click Save .		
	All data in the table (displayed or not) is saved and exported as a tab-delimited text file.		
Exporting Signature	Use this option to export only the signature SNPs in your data.		
SNPs	1. Click Export Signature SNPs.		
	An Explorer window appears.		
	2. Enter a filename, then click Save .		
	All data is now saved a tab-delimited text file.		
Exporting			

Genotyping Data

IMPORTANT! Not all options are available and are dependent on the export format you select and **its applicable format restrictions.**

1. Click Export Genotyping Data.

The following window appears: (Figure 136)

Figure 136	Export Genotype Data window			
Export Genotyping D	ata			×
Result Output Form	nats:	📃 Include Pedigree Info	rmation	
Call Output Format	is: 🔿 Forward Strand Base Call 🔘 Call Codes 🔿 Numeric Call Codes	i ☑ Include '/' Separator B	Between Alleles	
Exported Data: 🔲	Confidence 🔲 Signal	Include CN Region C	alls	
Input and Output	Files			
SNP List Filter:				• 🗙
Output Location:	C:\Users\Public\Documents\AxiomAnalysisSuite\Export			
Output Name:				.txt
Annotation File:				
Axiom_PMRA.na35.annot.db			Browse	
SNP Identifier				
Probe_Set_ID			-	
Select Annotation Column(s) to Add:				
Check/Unchecl	k All			
 affy_snp_id_an Chromosome Chromosome Chromosome Strand dbSNP RS ID 	not Start Stop			
Display the second seco	e			•
			ОК	Cancel

Result Output Formats

Figure 137 Result Output Format selections			
Result Outpu	t Formats: 🍥 TXT 🔘 VCF 🔘 PLINK (PED) 🔘 PLINK (TPED)	Include Pedigree Information	

- 1. Click the radio button to select the Result Output Format you want use.
- 2. Optional: If you selected a PLINK format, make sure you click the **Include Pedigree Information** check box. Not checking this box may require special handling (within PLINK) to make your exported output work properly.

Note: PLINK format specifies that all markers be biallelic, therefore multi-allele probesets will not be exported to PLINK files.

IMPORTANT! If you click the Include Pedigree Information check box with your PLINK format, make sure your Sample Attributes include an Index ID and Pedigree Information (Family ID, Individual ID, Father ID, Mother ID, Sex, and Affection Status).

3. If you have used OTV Caller, some of the calls may be OTV. To export these OTV calls, click the **TXT** and **Call Codes** radio buttons or **TXT** and **Numeric Call Codes** radio buttons. Note: For all other formats, OTV calls are treated as No Calls.

Call Output Formats

1. Click the radio button to select the appropriate Call Output Format (Figure 138) you want to use.

Figure 138 Call Output Format selections

Call Output Formats: O Forward Strand Base Call 💿 Call Codes O Numeric Call Codes 📝 Include '/' Separator Between Alleles

If you select Forward Strand Base Call, you have the option of not including the '/' separator between alleles. Note: Excluding separators is NOT advised when exporting probesets with multi-base alleles.

IMPORTANT! If exporting Numeric Call Codes, the exported data file's header rows includes a table mapping numeric call code to call code. The format of these header rows is:

#%%call-code-N=call_code:numeric_call_code:code_ploidy

The assignment of numeric call codes to call codes is NOT guaranteed to be the same across all supported array types.

Exported Data Selections

1. Click inside the check box(es) to check the additional type(s) of Exported Data (Figure 139) you want to include.



Note: Multi-allele probesets will not export signal values. Normalized signal values for multi-allele and bi-allele probesets are available in the AxiomGT1.summary.txt file inside the batch results folder.

If you selected the VCF output format and copy number analysis was performed, you have the option to not include CN Region Calls from the VCF file.

Input and Output Files

(Figure 140)

Figure 140	Input and Output Files selections	
Input and Output	Files	
SNP List Filter:		 • ×
Output Location:	$C: \label{eq:constraint} C: \label{eq:constraint} C: \label{eq:constraint} Users \label{eq:constraint} C: \label{eq:constraint} Users \label{eq:constraint} C: \label{eq:constraint} Users \label{eq:constraint} V: $	
Output Name:		.txt
		J

SNP List Filter (Optional)

1. To restrict the output to a list of SNPs (probeset_IDs) contained in a file, click the SNP List Filter field's **Browse** button (Figure 140).

An Explorer window appears.

2. Navigate to the SNP Filter List location, then click Open.

The SNP Filter List field is now populated. (Figure 141)

	Figure 141	SNP List Filter and Output path	
ſ	Input and Output	Files]
	SNP List Filter:	C:\Users\Public\Documents\AxiomAnalysisSuite\Export\C	- ×
	Output Location:	C:\Users\Public\Documents\AxiomAnalysisSuite\Export\	
L	Output Name:		.txt

3. Click the SNP List Filter's drop-down menu to view and select a previously saved SNP List. (Figure 141)

Output Location (Required)

1. Click the Output Location field's Browse button.

An Explorer window appears.

2. Navigate to an output location, create a new folder if needed, then click **Select Folder** button.

The Output Location path is displayed. (Figure 141)

Output Name (Required)

1. Use the output name already in the Output Name field, or click inside the field to enter a new name. Note: Your output name's file extension reflects the Results Output Format you selected in Step 1.

Changing the SNP Identifier

IMPORTANT! Only SNPs that have a value for the selected annotation are exported. For example, markers that have a dbSNP RSID are exported, while markers without a dbSNP RSID are not exported.

If the selected SNP Identifier has more than 1 probeset mapped to it, it will have multiple entries in the exported file.

1. Click the drop-down arrow, then click to select the SNP Identifier you want to use. (Figure 142)

Figure 142	SNP Identifier	
SNP Identifier Probe_Set_ID		

6

Changing the Current Annotation File (Optional)

1. To change the currently displayed Annotation File, click the Annotation File field's **Browse** button (Figure 143).

An Explorer window appears.

2. Navigate to the appropriate Annotation File location, then click **Open**. Your newly selected Annotation file is displayed.

Adding and Removing Annotation Columns

 Click the check box next to the Annotation Column(s) you want to add to your format results or click to uncheck/remove a column. If you want to add or remove ALL available annotations, click the Check/Uncheck All check box. (Figure 143)

Annotation File: Axiom_GW_Hu_SNP.na34.annot.db Select Annotation Column(s) to Add: Check/Uncheck All Affy_SNP_JD Chr_id Stat Stop Strand dbSNP_RS_JD dbSNP_Loctype In_Hapmap Strand_Vs_dbSNP Probe_Count Cytoband ChrX_PAR Flank Allele_A Allele_B Ref_Allele Ref_Allele	Figure 143 Select Annotation Column(s) pane
Axiom_GW_Hu_SNP.na34.annot.db	Annotation File:
Select Annotation Column(s) to Add: Check/Uncheck All Affy_SNP_ID Chr_id Start Stop Stard dbSNP_RS_ID dbSNP_Loctype In_Hapmap Strand_Vs_dbSNP Probe_Count Cytoband ChrX_PAR Flank Allele_A Allele_B Ref_Allele	Axiom_GW_Hu_SNP.na34.annot.db
Check/Uncheck All Affy_SNP_ID Chr_id Start Stop Strand dbSNP_RS_ID dbSNP_RS_ID dbSNP_Loctype In_Hapmap Strand_Vs_dbSNP Probe_Count Cytoband ChrX_PAR Flank Allele_B Ref_Allele	Select Annotation Column(s) to Add:
 Affy_SNP_ID Chr_id Start Stop Strand dbSNP_RS_ID dbSNP_Loctype In_Hapmap Strand_Vs_dbSNP Probe_Count Cytoband ChrX_PAR Flank Allele_A Allele_B Ref_Allele 	Check/Uncheck All
Alt_Allele Associated_Gene Genetic_Map Microsatellite Heterozygous_Allele_Frequencies Allele_Frequency_Count Allele_Frequencies Minor_Allele Minor_Allele Minor_Allele_Frequency OMIM Biomedical	Affy_SNP_JD Affy_SNP_JD Chr_id Start Stop Strand dbSNP_RS_JD dbSNP_Loctype In_Hapmap Strand_Vs_dbSNP Probe_Count Cytoband ChrX_PAR Flank Allele_A Allele_B Ref_Allele Att_Allele Att_Allele Genetic_Map Microsatellite Heterozygous_Allele_Frequencies Allele_Frequencies Allele_Frequencies Minor_Allele Minor_Allele Minor_Allele Frequency OMIM Biomedical Anotation_Notes

- 2. After the Export Genotype Data form is complete, click OK.
- 3. Your newly exported data now reside in the output location you defined in Step 1 on page 115.

Exporting Cluster Plots to PDF

This exporting option generates a specific number of cluster plots from the selected Conversion Type choices.

1. Click Export Cluster Plots to PDF.

The following window appears: (Figure 144)

Figure 14 window	4 Report Settings		
Report Settings	×		
File Name:	Browse		
Picture Setting	gs:		
O All SNPs free	om Current Table		
Random SI	NPs from Current Table		
SNP Count	200		
Row Count	3		
Col Count	4		
	Default		
Conversion Ty	pes:		
Check/Uncheck All			
CallRateBelowThreshold			
MonoHighResolution			
NoMinorHom			
Other			
OTV Debd Vieb Break time			
	UK Cancel		

2. Click **Browse**.

An Explorer window appears.

- 3. Navigate to a desired location, then enter a name for your PDF report.
- 4. Click Save.

You are returned to the **Report Settings** window.

- 5. In the Picture Settings section, click either:
 - All SNPs from Current Table
 - Random SNPs from Current Table
- 6. Click inside the applicable **Count** field(s) to enter how many cluster pots you want to export.

Click the **Default** button to revert your modified counts back to their original states.

7. Click to check a Conversion Type(s) you want to add to the report or click the **Check/Uncheck All** check box, then click **OK**.

A Please Wait message and progress bar appear. Allow several minutes if multiple Conversion Type PDF report(s) were selected.

An Explorer window (where you saved the PDF Report location in Step 3) appears.

8. Double-click on the PDF Report you want to view.

The PDF Report opens. (Figure 145)

6



Note: One plot is made for each probeset. For multi-allele probesets, the most informative pair of allele signal channels in the data set is used for plotting, based on the distribution of genotype calls.

External Tools



Axiom Analysis Suite auto-detects any previously installed application that may further analyze your genotype results. When a compatible application is detected, an **External Tools** window tab is generated, as shown in Figure 146.

Figure 146 External Tools window tab	
Sum Sample SNP Summar CN Summar External External Tools	^
The tools listed below are designed to further analyze the genotype results. These tools may require that this application be closed to provide access to the result file.	
Axiom CNV Tool	
CDCB Export Tool	
Long Format Export Tool	

IMPORTANT! The Axiom Analysis Suite can be left open/active if you are using an external application to further view your data. However, you must exit and close the Axiom Analysis Suite before editing its data with an external application.



Axiom CNV Tool 1.1

1. Click the **Axiom CNV Tool** button. The application opens. (Figure 147)

Figure 147 CNV Tool - Main window	
CNV Tool	
Axiom CNV Tool	1 2
Data Directory Annotation File	
Specify Gender Information (required if Data Directory does not contain AxiomGT1 report bt file) Select Gender File	
Specify Reference Data Select Ref File	
Export Nexus Format Export PennCNV Format Export One Sample Per File (Nexus Format) Configure Nexus Run	Stop

For details on how to use the Axiom CNV Tool 1.1 application, refer to its User Guide (**P/N 703216**).

Council on Dairy Cattle Breeding (CDCB) Export Tool

1. Click the CDCB Export Tool button.

The application opens. (Figure 148)

cuments\AxiomAnalysisSuite\Output\20170201 Buffs	
	2
Buffalo.r2FinalReport.txt	
Buffalo.r2SampleSheet.csv	
are being used by the Axiom Analysis Suite close that software to proceed with the export.	
	Buffalo.r2FinalReport.txt Buffalo.r2SampleSheet.csv are being used by the Axiom Analysis Suite close that software to proceed with the export.

For details on how to use the CDCB Export Tool application, refer to its User Guide (**P/N 703465**).

7

Axiom Long Format Export Tool

 Click the Long Format Export Tool button. The application opens. (Figure 149)

ppliedbiosystems	Axiom Long Format Export Tool (?) (i) 🗕	
Analysis Results	C:\Users\Public\Documents\AxiomAnalysisSuite\Output\20170201 Buffalo	
Array Name	Axiom_Buffalo.r2	
Annotation File	Axiom_Buffalo.r2.annot.db	
SNP Name Mapping File		
Genotypes Export File	20170216_Axiom_Buffalo.r2.txt	1
	The analysis results are being used by the Axiom Analysis Suite software. You must close that software to proceed with the export.	

For details on how to use the Long Format Export Tool application, refer to its User Guide (**P/N 703455**).



Predefined Region CN Analysis Variations

Predefined Copy Number Analyses

If the array content and library package supports it, the Genotyping and Best Practices workflows perform predefined region copy number analysis. Copy number analysis is restricted in that the copy number regions are pre-specified, with a fixed set of probesets used to estimate a single copy number state for each region. This form of copy number analysis is done within the Genotyping and Best Practices workflows.

Axiom Analysis Suite supports two variations of restricted copy number analysis. These analysis variations are determined by the array library package and are compared in Figure 150.

An					/sis Variation			
Workflow Stage		Analysis Description		Predefined CN Genotyping †	CN-Aware Genotyping ‡			
		Sample QC	yes	yes	yes			
Best Practices	Sample QC	Identify Copy Number (CN) control samples that pass Sample QC	not done	female samples used as CN plate controls	CN controls identified by comparing measured with reference genotype calls			
		Normalize plate signals using CN control samples that pass CN QC. Compute CN state in pre-defined regions. Samples that fail CN QC will report a CN state of NoCall.	not done	yes	yes			
	Genotyping	Supply CN results to genotyping engine	not done	no	CN-Aware Genotyping ‡ yes CN controls identified by comparing measured with reference genotype calls yes yes yes			
		Final genotyping	yes	yes, including OffTargetVariant calls				

With Predefined CN Genotyping

- CN states will be reported for the predefined regions specified by the library package
- Unless overridden by the user, the control samples used for per-plate signal correction for CN analysis are selected from the set of samples determined to be female and passing sample QC



- Unless overridden by custom library file settings, genotyping probesets will report diploid genotype codes (such as "AA", "AB", "BB", "NoCall"), whether or not they fall within CN regions reporting deletions.
- OTV Caller is automatically run if any SNPs are assigned to the Conversion Type "OTV". Some of the resulting genotype calls may be "OTV", indicating a detected Off Target Variant.

With CN-aware Genotyping

- CN states will be reported for the predefined regions specified by the library package
- Unless overridden by the user, the control samples used for per-plate signal correction for CN analysis are identified by high concordance between measured and reference genotypes for expected control samples, and should also pass sample QC
- SNPs in a CN=0 region will be called as "ZeroCN"
- SNPs in a CN=1 region will be called with haploid genotype codes such as "A", "B", "NoCall_1"
- SNPs in a CN=2 or higher region will continue to be called with diploid genotype codes such as "AA", "AB", "BB", "NoCall"
- Special SNPs (those on chromosomes Y, MT, and non-PAR X) will also be assigned hemizygous and ZeroCN genotype codes when expected copy number is less than two
- CN-aware genotyping can be applied to probesets between two measured CN regions. For example, if two adjacent regions predict the same CN state, genotyping probesets between those two regions can be assigned genotype call codes consistent with that CN state.

Note: Examples of library packages that support these copy number analyses include; CarrierScan for Predefined CN genotyping, and PharmacoScan for CN-aware genotyping.



Genotyping with CN Analysis

Setting Up a Genotyping with CN Analysis

Before setting up a genotyping with predefined CN analysis, click the New Analysis window tab, then click the Array Type drop-down menu to confirm an array such as PharmacoScan or CarrierScan is available, as shown in Figure 151. If it is, continue to "Selecting a Mode (Workflow)".

Figure 151 Pret	ferences window tab	
Mode: Best Practices Worl	Array Type: PharmacoScan Analysis Settings	iles Import CEL Files by Txt Remove Selected Files Threshold Settings
File Name Click "Import CEL Files" to import files for analysis.	Select Analysis Configurations PharmacoScan.r1 (Default) Restore Save Save As Save As Save As Genotyping	Select Threshold Configurations PharmacoScan (Default) Restore Save Save As Sample QC CN QC SNP QC
Output Folder: C:\Users\P	ublic\Documents\AxiomAnalysisSuite\Output\ Brov	wse Batch Name: Run Analysis

Selecting a Mode	From the main Axiom Analysis Suite window tab, click the Mode drop-down.
(Workflow)	1. Select Best Practices Workflow.
	Best Practices Workflow performs and combines the Sample QC Workflow and Genotyping Workflow. For more information, see "Overview and Use of the Best Practices Workflow" on page 137.
Importing CEL Files	1. Click Import CEL Files . The Add CEL Files window appears
	2. Navigate to your CEL file location.

- **3**. Single-click on a CEL file or Ctrl click, Shift click, or press Ctrl A (to select multiple files).
- 4. Click Open.

The CEL Files pane populates and displays your selected CEL files. (Figure 152)

	Figure 152 Cell File pane
	CEL Files: 24
1	File Name
I	HG00127_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG00366_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG01770_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	HG02086_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	HG02301_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG02373_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	HG02589_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG02679_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	HG03021_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03058_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03225_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03460_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03521_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03556_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03598_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	HG03643_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	NA18541_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	NA19118_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	NA19315_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	NA19318_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	NA19395_DB_10uLmPCRspike_Pharmacoscan_24_2016
I	NA20289_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	NA20291_DB_10uLmPCRspike_Pharmacoscan_24_2016
1	NA20581_DB_10uLmPCRspike_Pharmacoscan_24_2016

After importing your CEL files, the Analysis and Threshold Setting panes autopopulate with default settings and additional user-configurable fields and settings specific to the library package, as shown in Figure 153.

For information on typical Axiom array analysis and threshold fields, see "Setting Up an Analysis" on page 19 and "Setting Up Threshold Settings" on page 28.

12	2	2	3	U
		r	ų	
		1	17	
	6	62	53.	

		_						
Figure 153 Example: Auto-populated Ana	aly	/S	is and Threshold S	etti	ng pa	nes		
Analysis Settings		1	hreshold Settings					
Select Analysis Configurations	*	5	elect Threshold Configurations				*	
PharmacoScan 24F.r6 (Default)		ſ	PharmacoScan (Default)	Rest	ore Save	Save As	ח ו	
Sample OC		0	A Sample OC					
GT Analysis File:			Namo		ottings		1	
PharmacoScan 24F SNPSpecificPriors Step1.r6.apt-genotype-axiom.AxiomGT1.apt2			Name	-	ettings			
Prior Model File:			DQC	2 .	• 0.88	5		
PharmacoScan_24F.r6.AxiomGT1			QC call_rate	2 .	• 98	ຄ		
SNP List File:			Percent of passing samples	2	• 95	5		
PharmacoScan_24F.r6.step1			Average call rate for passing camples	>	98.5	5		
Gender File (optional):		ŀ	a set to a s	-				
🗙			Control comparisons	<u>></u> .	- 30	*7		Available for
Hints/Inbred File (optional):			Control concordance	2 .	• 93	5		CN-aware
🔿 Inbred 💿 Hints 🛛 📖 🗙				_		_	11	genotyping
Control Reference Calls File:		ľ				_	1	analysis variation
PharmacoScan_24F.r6.signatureSNPs.refs.cn_controls	=		Name		ettings			
Genotyping		I	MAPD	≤ .	• 0.35	5		
(N Control CEL List File (ontional):		0	Waviness SD	<u>د</u>	• 0.1	າ		
						-	•	
CN Analysis File:							, Ш	Available for
PharmacoScan 24F.r6.apt-copynumber-axiom-ssa.AxiomCN1.apt2			Name		settings			CN-aware
CN Reference Model File:			species-type	Diplo	oid 🔻	5		genotyping and
PharmacoScan_24F.r6 🗙			cr-cutoff	2	• 95	5		Predefined CN
CN Bins File:			fld-cutoff	2 .	- 3.6	5		analysis variations
PharmacoScan_24F.r6.cnbins 🗙			hat an autoff	<u> </u>	- 02	-		
GT Analysis File:			net-so-cuton		-0.2	- 1		
PharmacoScan_24F_SNPSpecificPriors_Step2.r6.apt-genotype-axiom.AxiomGT1.apt2 🔻			het-so-XChr-cutoff	2 .	-0.2	າ		
Prior Model File:			het-so-otv-cutoff	2	-0.3	5		
PharmacoScan_24F.r6.AxiomGT1			hom-ro-1-cutoff	2	-0.5	5		
Multi-allele Background Prior Model File:				_	1.1			
PharmacoScan_24F.r6.AxiomGT1.mmb			nom-ro-2-cutoff	2.	-1.5	*1		
Multi-allele Pairwise Prior Model File:			hom-ro-3-cutoff	2	-1.5	5		
PharmacoScan_24F.r6.AxiomGT1.mmp			hom-ro	true	•	5		
Multi-allele Prior Model File:			hom hat	true				
PharmacoScan_24F.r6.AxiomGT1.mm			nom-net	line	- -	, -,		
SNP List File (optional):	-		num-minor-allele-cutoff	2 .	• 2	5	-	
		_						



Analysis Settings with Copy Number Options

IMPORTANT! Only experienced users should modify default analysis settings.

Sample QC

- **GT Analysis File**: Parameters file for the genotyping step that calculates QC Call Rate.
- **Prior Model File**: Defines prior knowledge of SNP cluster locations. This file has the same format as a posteriors file, which is generated by the genotyping step. This means that you can "train" on a custom data set, and use the updated knowledge of cluster locations as a "seed" to possibly improve future genotyping batches. This file must contain two row entries for the GENERIC and GENERIC:1 probesets (if there are any probesets to be genotyped that are not listed in this file).
- **SNP List File**: A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.
- **Gender File**: A file specifying the desired gender of every sample. If supplied, software will use values in this file instead of the computed gender. Gender impacts genotyping of chromosome X and Y SNPs.
- **Hints/Inbred File**: If a hints file, a file of expected genotype calls. This is used to influence the predicted cluster locations, which influences the final calls. In the Sample QC section, it influences the QC Call Rate.
- **Control Reference Calls File**: For CN-aware genotyping only, a recommended file containing the expected signature SNP calls of the CN control samples and used by Sample QC to identify the control samples among the supplied CEL files.

Genotyping

• **CN Control CEL List File**: This optional file identifies the CEL files that are to be used for per-plate tuning of CN signals. The file has the same format as file **[Batch Name]\CNData\CNcontrolSamples.pass.txt**, which is generated by a Sample QC step. The following table describes how the Workflow and this input option interact to select the controls used for CN analysis.

Workflow	CN Control CEL List File Input	Chosen Controls for CN Analysis			
Best Practices	None	Passing controls identified by Sample QC step			
Genotyping	None	No controls, therefore no plate signal tuning is done.			
Best Practices	User-supplied	User-supplied controls			
Genotyping	User-supplied	User-supplied controls			

- CN Analysis File: Parameters file for the CN Analysis step.
- CN Reference Model File: Reference information for CN Analysis step.



- **CN Bins File**: Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
- GT Analysis File: Parameters file for the final genotyping step.
- **Prior Model File**: Defines prior knowledge of bi-allelic SNP cluster locations. This file has the same format as a posteriors file, which is generated by the genotyping step. This means that you can "train" on a custom data set, and use the updated knowledge of cluster locations as a "seed" to possibly improve future genotyping batches. This file must contain two row entries for the GENERIC and GENERIC:1 probesets (if there are any probesets to be genotyped that are not listed in this file).
- If the library package supports multi-allele SNPs, then there are three additional model files for the three stages of multi-allele genotyping: Multi-allele Background Prior Model File, Multi-allele Pairwise Prior Model File, and Multi-allele Prior Model File.
- **SNP List File**: A file of probeset IDs to genotype. For Genotyping it defines the probesets for which genotypes will be reported.
- **Gender File**: A file specifying the known gender of every sample. If supplied, software will use values in this file instead of the computed gender. Gender impacts genotyping of chromosome X and Y SNPs.
- **Hints/Inbred File**: If a hints file, a file of expected genotype calls. This is used to influence the predicted cluster locations, which influences the final calls.
- **Posterior File Name**: The desired file output of a genotyping analysis, specifying identified SNP probeset cluster locations, variance, and relative weight. One use of this file is as a prior model file for future genotype analyses.
- **ps2snp File**: If multiple probeset designs exist on the array for a given SNP (for example, one forward and one reverse strand design), then the ps2snp file is used by the SNP classification step to identify the best performing probeset for the SNP, using the priority-order setting in the SNP QC section in the New Analysis tab. This text file has two tab delimited columns with the headers probeset_id and snpid (**snpid = affy_snp_id**).
- **Genotype Frequency File**: If the library package supports a check for unexpectedly high call frequency for specific genotypes, this optional file specifies the maximum expected frequency for reviewed genotypes.



Threshold Configurations with CN Options

IMPORTANT! Only experienced users should modify default threshold settings.

Sample QC

Control Comparisons (CN-aware genotyping only): For identifying control samples, this is the minimum number of SignatureSNP probesets compared to a reference.

Control Concordance (CN-aware genotyping only): For identifying control samples, this is the minimum percent concordance of SignatureSNP calls to a reference.

CN QC

MAPD: Median Absolute Pairwise Difference of log2ratio signals of adjacent copy number (CN) probesets must be below this value to make CN calls.

Waviness SD: Waviness Standard Deviation of log2 ratio signals of copy number (CN) probesets must be below this value to make CN calls.

SNP QC

Refer to Table 11 on page 149 for SNP QC Threshold name definitions.

Assigning an Output 1. Click the Output Folder path's Browse button. (Figure 154)

Folder Path

Figure 154	Output Folder field
Output Folder: C:\User	rs\Public\Documents\AxiomAnalysisSuite\Output\ Browse Browse
	An Explorer window appears.
	 Navigate to the recommended path C:\Users\Public\Documents\AxiomAnalysisSuite\Output, then click Select Folder.
	Your selected output folder path is now displayed.
Assigning a Batch Name	The batch file is produced while your analysis is running and includes all the necessary files needed to view your analysis in the Axiom Analysis Suite Viewer.
	1. Enter a name in the Batch Name field. (Figure 155)
	Figure 155 Enter a Batch

 Figure 155
 Enter a Batch

 Name
 Run Analysis

Note: A folder (with the same name as your entered batch name) is auto-generated during the analysis process. This folder includes all the necessary files needed to view your analysis results in the Viewer.

Running your Genotyping with CN Analysis

1. Click Run Analysis

The Dashboard window tab appears. (Figure 156)

F	Figure 156 Dashboard window tab									
	New Analysis Dashboard Preferences									
	Workflows									
	Batch Name	Date Created	Workfl	Array Type	Status	Elapsed	Status	Warning	Action	
	20160304_pscan	3/4/2016 10:06:32 AM	Best Practices Workflow	PharmacoScan.r1	Success	10 minutes			Open]

After Success is displayed in the Status column (Figure 156), click Open.

The **Open** button will not appear if a processing error occurs. If a message appears stating that no samples passed QC, click the **Open Selected Result(s)** button to review the analysis result that may need troubleshooting.

The Axiom Analysis Suite Viewer opens in a new window (Figure 157) and displays your completed analysis results.

Note: To make comparisons between your completed data analyses easier, open additional Viewer windows. To do this, click the **Dashboard** window tab, then click on the **Open** button again.

Viewing your Genotyping and CN Results

After processing arrays that support predefined CN or CN-aware genotyping, the following is added to the Axiom Analysis Suite Viewer:

- New entries in the Summary Report, Sample Table, and SNP Summary Table.
- For CN-aware genotyping, the Cluster Plot also displays hemizygous and ZeroCN clusters for SNPs in CN regions.
- CN Summary Table, with Export CN Data option.
- A CN Region Plot window tab.
- Exported VCF formats can now include CN and SNP calls.



Summary Report After successfully running a Sample QC or Best Practices workflow, the Summary report's Plate QC Summary section features two additional fields for the CN-aware genotyping analysis variation, as shown in Figure 157.

- Number of controls found This metric is a count of samples identified as controls based on high signature SNP concordance to expected calls in the Control Reference Calls library file.
- Controls in normal wells The Control Reference Calls library file lists the expected plate wells for the control samples. This metric will report "Yes" if all the detected control samples are in the expected plate wells. It is OK to put the control samples in any wells you choose.

Figure 157 Viewe	er wind	dow ta	abs								
Summary Sample Table S	NP Summa	ry Table	CN Sum	nary Table	e Cluste	r Plot C	N Region	Plot			
Export to File											
Analysis Summary										*	
 Batch Name: MyBatch Array Package Name: F Array Type Name: Phar Array Display Name: Pf Workflow Type: Best Pr Date Created: 7/14/2010 	PharmacoSc macoScan narmacoSca actices Worl 5 4:24:38 AN	an.r3 n.r3 «flow A									
Sample Summary Number of input samples: Samples passing DQC: 16 Samples passing DOC an	: 167 57 out of 16 d OC CR: 10	7 56 out of 10	67							=	
Samples passing DQC, Q Number of failing sample: Number of input samples Number of Samples Gence Average QC CR for the p Gender Calls Counts: fem	C CR and P s: 1 without QC otyped: 166 assing samp ale=90 male	late QC: 16 informatio les: 99.791 =77 unknov	6 out of 16 n: 0 wn=0	7 (99.401%	6)						
Plate QC Summary											Columna
Plate Barcode	Result	Number of files in a batch	Number of files failing dish QC	Number of files failing QC Call rate	Number of samples that passed	Percent of passing samples	Average call rate for passing	Number of controls found	Controls in normal wells		reserved for
5507454301008100717698 5507454301008100717699 5507454301008100717701	PASSED PASSED PASSED	24 24 24	0	0	24 24 24	100 100	samples 99.845 99.842 99.659	2 2 2 2	Yes Yes Vec		genotyping
5507454301008100717703 5507454301008100717704 5507454301008100717705	PASSED PASSED PASSED	24 24 23	0	0	24 24 22	100 100 95.652	99.859 99.75 99.744	2 2 2	Yes Yes Yes		analysis variation.
View details	PHOOED	24	0	0	24	100	77.034	2	TES		
CN Summary								1			Columns
Plate Barcode	Number of files analyzed	Number of samples failing CNQC	Number of samples passing CNQC	Number of controls analyzed	Number of controls failing CNQC	Number of controls for CN tuning	CN tuned using controls				reserved for CN-aware
5507454301008100717698 5507454301008100717699 5507454301008100717701	24 24 24	0	24 24 24	2 2 2	0	2 2 2	yes yes yes				genotyping and pre-
5507454301008100717703 5507454301008100717704 5507454301008100717705 5507454301008100717706	24 24 22 24	0 0 0 0 0	24 24 22 24	2 2 2 2	0 0 0 0 0	2 2 2 2	yes yes yes yes				defined CN
SNP Metrics Summary											variations.
- 17 4 CONTR 100544										Ψ 	

After successfully running a Genotyping or Best Practices workflow, the Summary report's **CN Summary** section features a table of information for each plate, as shown in Figure 157. Refer to Table 8 for descriptions of each CN Summary Report column.



CN Summary Report	Description
Number of files analyzed	Count of CEL files supplied to the Genotyping step (which includes CN analysis).
Number of samples failing CNQC	Count of CEL files that fail copy number quality control checks, which is the result of a comparison of measured MAPD and waviness SD metrics against thresholds. Samples that fail CN QC report a CN state of NoCall. For the CN-aware genotyping analysis variation, SNPs from samples that fail CN QC are called without the benefit of CN-aware genotyping.
Number of samples passing CNQC	Count of CEL files that pass copy number quality control checks.
Number of controls analyzed	Count of CEL files supplied to the Genotyping step that are identified as controls.
Number of controls failing CNQC	Count of control CEL files that fail copy number quality control checks, which is the result of a comparison of measured MAPD and waviness SD metrics against thresholds. Controls that fail CN QC report a CN state of NoCall. For the CN-aware genotyping analysis variation, SNPs from samples that fail CN QC are called without the benefit of CN-aware genotyping.
	Note: For the CN-aware genotyping analysis variation, Non-PAR X, Y, and mitochondrial SNPs use gender and other information to report non-diploid calls, regardless of CN QC status.
Number of controls for CN tuning	Count of control CEL files that pass copy number quality control checks.
CN tuned using controls	If Number of controls for CN tuning > 0, then this value is "Yes". This means that CN signals are adjusted based on the signal measured in the control samples from the same plate. If the controls are appropriate for the samples being processed, this adjustment can improve CN calling accuracy.

Table 8 CN Summary Report

Sample Table After successfully running a CN analysis, additional columns appear within the Sample Table, as shown in Figure 158.

Summary Sample Table SNP Summary Table CN summary Table Cluster Plot CN Region Plot Cluster Plot CN Region Plot Scatter Plot Box Plot Plate View Concordance Reanalyze Selected Samples Import Sample Attributes Revert Calls Apply View Save View Sove View	Figure	Figure 158 Sample Table after a CN Analysis run																				
Scatter Plot Box Plot Plate View Concordance Reamayze Selected Samples Import Sample Attributes Revert Calls Apply View Save View Show/Hide Columns Export Filters Sample Pass/Fail DQ QC call_rate call_val Percent of plate- rate for passing passing passing partices MAPD WavinessD CN CN samples Passing call CN tuned ontrol for using control for using contr	Summary	Summary Sample Table SNP Summary Table CN Summary Table Cluster Plot CN Region Plot																				
Apply/View Save View Show/Hide Columns Export Filters Affyrmetric-plate- barcode offyrmetric-plate- plate- samples Operation samples MAPD WavinessD CN passes CN passes Control MAPD Control MAPD Number of CN tuning controls for samples H600185_JS Pass 0.978 99.935 99.85 29 27.415 male 550745430100810071768 AVerage call Percent of passing samples 0.274 0.033 yes yes yes no 0 2 yes es no 0 2 yes no 0 2 yes es	Scatter Plot B	catter Plot Box Plot Plate View Concordance Reanalyze Selected Samples Import Sample Attributes 🔹 Revert Calls 💌																				
Proprint Base Control Control Comput. affymetric-plate plate-rote for passing parcede Percent of plate-rote for passing amples Percent of passing CN passes CN passes Control Number of CN tuning choicen HG00185_JS Pass 0.978 99.935 99.85 29 27.415 male 550745430100810017698 6A5 99.844 100 0.264 0.043 yes yes no 0 2 yes H600185_JS Pass 0.956 99.825 9.812 27.229 female 550745430100810017698 6D5 99.844 100 0.27 0.037 yes yes no 0 2 yes yes no 0 2 yes yes no 0 2 yes yes yes no 0 2 yes yes ye	Annly View 🔻	Apply View V Save View Show/Hide Columns V Export V Filters V																				
Sample Filename Pass/Fail DQC QC call_rate het_rate QC comput affymetric-plate- barcode offymetric- plate- pl	Арріу вією	ppy area leave rise another common type interview of the second sec																				
HG00185_JS Pass 0.978 99.355 99.85 29 27.415 male 5507454301000100717698 A05 99.844 100 0.276 0.043 yes yes yes yes no 0 2 yes HG00018_JS Pass 0.954 99.835 99.737 29.701 27.223 female 5507454301000100717698 C07 99.844 100 0.27 0.033 yes yes no 0 2 yes H600081_S Pass 0.966 99.842 2427 24.312 male 5507454301000100717698 D09 99.844 100 0.226 0.041 yes yes yes no 0 2 yes H601018_JS Pass 0.969 99.842 100 0.256 0.041 yes yes yes no 0 2 yes H60119_JS Pass 0.971 99.835 99.873 99.873 99.873 99.873 23.805 7644301008100717698 E07 99.844 100 0.249 0.035 yes yes no 0 2 yes H602072_JS <	Sample Filename	Pass/Fail	DQC	QC call_rate	call_rate	QC het_rate	het_rate	QC comput	affymetrix-plate- barcode	affy plate - peg	Average call rate for passing samples	Percent of passing samples	MAPD	WavinessSD	CN passes MAPD	CN passes WavinessSD	CN passes QC	Control chosen	Control used	Number of controls for CN tuning	CN tuned using controls	
HG00313_J Pass 0.964 99.835 99.73 29.701 27.229 female 5507454301008100717688 C07 99.844 100 0.27 0.037 yes yes yes no 0 2 yes H600041_S Pass 0.559 99.825 99.825 99.825 25.288 25.305 female 5507454301008100717688 B05 99.844 100 0.221 0.033 yes yes yes no 0 2 yes H601085_JS Pass 0.969 99.875 99.842 28.37 female 5507454301008100717688 B09 99.844 100 0.225 0.041 yes yes yes no 0 2 yes H601198_JS Pass 0.979 99.842 28.37 female 5507454301008100717698 E07 99.844 100 0.256 0.041 yes yes yes no 0 2 yes H602072_JS Pass 0.963 99.87 99.832 25.313 23.44 female 550745430100810071	HG00185_JS	Pass	0.978	99.935	99.85	29	27.415	male	5507454301008100717698	A05	99.844	100	0.264	0.043	yes	yes	yes	no	0	2	yes	4
HG0004U_J Pass 0.959 99.825 99.812 25.298 23.805 female 5507454301008100717698 B05 99.844 100 0.291 0.033 yes yes yes no 0 2 yes Pss HG00083_J Pass 0.969 99.845 99.842 24.827 24.312 male 5507454301008100717698 D09 99.844 100 0.225 0.0.01 yes yes yes no 0 2 yes HG01085_J Pass 0.97 99.845 28.885 male 5507454301008100717698 E07 99.844 100 0.258 0.004 yes yes no 0 2 yes HG01191_S Pass 0.97 99.843 23.392 26.837 female 5507454301008100717698 E07 99.844 100 0.267 0.038 yes yes yes no 0 2 yes HG01191_S Pass 0.963 99.87 99.832 23.33 23.44 female 5507454301008100717698 E07 99.844 100 0.275	HG00313_JS	Pass	0.964	99.835	99.737	29.701	27.229	female	5507454301008100717698	C07	99.844	100	0.27	0.037	yes	yes	yes	no	0	2	yes	
H-G0088_J_S Pass 0.66 99.842 24.827 24.312 male 5507454301008100717688 D09 99.844 100 0.225 0.036 yes yes no 0 2 yes H601085_J_S Pass 0.69 99.87 99.805 29.316 28.88 male 5507454301008100717698 B9 99.844 100 0.258 0.041 yes yes no 0 2 yes H601195_J_S Pass 0.974 99.815 99.843 20.936 20.837 fmale 5507454301008100717698 E07 99.844 100 0.258 0.041 yes yes yes no 0 2 yes H601219_J_S Pass 0.963 99.879 99.832 25.33 23.44 fmale 5507454301008100717698 E07 99.844 100 0.257 0.038 yes yes no 0 2 yes H602086_J_S Pass 0.56 99.87 99.77 2.649 23.647 fmale 5507454301008100717698 E09 99.844 100 0.27	HG00404_JS	Pass	0.959	99.825	99.812	25.298	23.805	female	5507454301008100717698	B05	99.844	100	0.291	0.033	yes	yes	yes	no	0	2	yes	=
HolD108_JS Pass 0.969 99.87 99.805 29.316 28.885 male 5507454301008100717698 B09 99.844 100 0.256 0.041 yes yes no 0 2 yes H601189_JS Pass 0.37 99.95 99.834 28.389 26.387 female 5507454301008100717698 E07 99.844 100 0.224 0.035 yes yes no 0 2 yes H60119J_S Pass 0.963 99.874 99.832 27.487 female 5507454301008100717698 E05 99.844 100 0.257 0.038 yes yes no 0 2 yes H602005_J_S Pass 0.566 99.879 99.879 99.874 100 0.257 0.038 yes yes no 0 2 yes H602005_J_S Pass 0.566 99.879 99.879 99.873 23.383 female 5507454301008100717698 E09 99.844 100 0.226 0.042 yes no 0 2 yes H602258J_S	HG00583_JS	Pass	0.96	99.865	99.842	24.827	24.312	male	5507454301008100717698	D09	99.844	100	0.295	0.036	yes	yes	yes	no	0	2	yes	
HG01159_S Pass 0.97 99.005 99.834 28.39 26.837 female 5507454301008100717698 E07 99.844 100 0.249 0.035 yes yes no 0 2 yes H601159_S Pass 0.974 99.813 23.33 23.34 female 5507454301008100717698 E07 99.844 100 0.238 0.043 yes yes no 0 2 yes H602015_S Pass 0.956 99.879 99.833 23.33 24.4 female 5507454301008100717698 E07 99.844 100 0.257 0.038 yes yes no 0 2 yes H602016_S Pass 0.956 99.879 99.77 25.649 23.964 female 5507454301008100717698 E07 99.844 100 0.276 0.033 yes yes no 0 2 yes H602058_J_S Pass 0.956 99.78 24.376 female 5507454301008100717698 E07 99.844 100 0.27 0.03 yes yes	HG01085_JS	Pass	0.969	99.87	99.805	29.316	28.885	male	5507454301008100717698	B09	99.844	100	0.258	0.041	yes	yes	yes	no	0	2	yes	
HolD151_JS Pass 0.974 99.915 99.842 23.96 27.048 frame 5507454301008100717698 E05 99.844 100 0.030 0.043 yes yes no 0 2 yes H602072_JS Pass 0.956 99.87 99.832 23.313 23.44 female 5507454301008100717698 B07 99.844 100 0.257 0.038 yes yes no 0 2 yes H602076_JS Pass 0.956 99.78 99.78 23.944 female 5507454301008100717698 E09 99.844 100 0.267 0.038 yes yes no 0 2 yes H602085_JS Pass 0.956 99.78 99.75 24.367 female 5507454301008100717698 E09 99.844 100 0.27 0.033 yes yes no 0 2 yes H602055_JS Pass 0.956 99.75 24.41 22.424 male 5507454301008100717698 F05 99.844 100 0.27 0.038 yes yes	HG01198_JS	Pass	0.97	99.905	99.834	28.399	26.837	female	5507454301008100717698	E07	99.844	100	0.249	0.035	yes	yes	yes	no	0	2	yes	
H602072_JS Pass 0.963 99.87 99.833 25.333 23.44 female 5507454301008100717698 B07 99.844 100 0.257 0.038 lyes yes yes no 0 2 yes H60208_JS Pass 0.966 99.87 99.77 25.649 23.964 female 5507454301008100717698 A09 99.844 100 0.265 0.035 lyes yes yes no 0 2 yes H602035_JS Pass 0.956 99.78 99.375 25.33 23.467 female 5507454301008100717698 E09 99.844 100 0.276 0.038 lyes yes no 0 2 yes H602255_JS Pass 0.956 99.78 94.75 24.342 rane 5507454301008100717698 F05 99.844 100 0.27 0.03 lyes yes yes no 0 2 yes H602555_JS Pass 0.958 98.42 12 2.244 male 5507454301008100717698 F05 99.844 100 0.276 0.036 lyes yes yes no 0 2 ye	HG01519_JS	Pass	0.974	99.915	99.843	29.396	27.048	female	5507454301008100717698	E05	99.844	100	0.308	0.043	yes	yes	yes	no	0	2	yes	
HG02086_JS Pass 0.956 99.87 99.87 25.649 23.964 frame 5507454301000100717688 A09 99.844 100 0.265 0.035 yes yes no 0 2 yes HG02133_S Pass 0.969 99.89 99.835 50.7454301000100717698 E09 99.844 100 0.265 0.042 yes yes no 0 2 yes HG02133_S Pass 0.969 99.895 99.75 24.336 female 5507454301000100717698 E09 99.844 100 0.276 0.03 yes yes no 0 2 yes HG02155_S Pass 0.958 99.879 93.71 24.336 27.45301000100717698 F05 99.844 100 0.276 0.031 yes yes no 0 2 yes HG02345_S Pass 0.958 99.879 99.872 24.056 22.94 male 5507454301000100717698 F05 99.844 100 0.276 0.036 yes yes no 0 2 yes	HG02072_JS	Pass	0.963	99.87	99.833	25.313	23.44	female	5507454301008100717698	B07	99.844	100	0.257	0.038	yes	yes	yes	no	0	2	yes	
H602133_J_C Pass 0.969 99.835 25.13 23.838 female 5507454301008100717698 E09 99.844 100 0.226 0.042 yes yes no 0 2 yes H602133_J_C Pass 0.956 99.78 24.336 23.467 female 5507454301008100717698 E09 99.844 100 0.27 0.03 yes yes no 0 2 yes H60258J_C Pass 0.956 99.78 24.14 12.824 male 5507454301008100717698 E05 99.844 100 0.27 0.03 yes yes no 0 2 yes H602360_C Pass 0.958 99.82 99.725 24.056 22.894 male 5507454301008100717698 E05 99.844 100 0.27 0.036 yes yes no 0 2 yes NA18541_JS Pass 0.969 99.78 24.344 female 5507454301008100717698 E05 99.844 100 0.27 0.036 yes yes no 0 2 yes	HG02086_JS	Pass	0.956	99.87	99.77	25.649	23.964	female	5507454301008100717698	A09	99.844	100	0.265	0.035	yes	yes	yes	no	0	2	yes	
HG02589_JS Pass 0.956 99.75 24.362 23.467 frame 5507454301008100717698 D07 99.844 100 0.27 0.03 yes yes no 0 2 yes HG02953_JS Pass 0.956 99.789 24.141 22.847 frame 5507454301008100717698 PG 99.844 100 0.278 0.03 yes yes no 0 2 yes HG03460_JS Pass 0.958 99.879 24.141 22.824 male 5507454301008100717698 PG 99.844 100 0.278 0.03 yes yes no 0 2 yes NA18541_JS Pass 0.979 99.875 24.844 frame 5507454301008100717698 DG5 99.844 100 0.27 0.036 yes yes no 0 2 yes NA18541_JS Pass 0.969 99.74 92.52 25.393 24.244 frame 5507454301008100717698 HG5 99.844 100 0.254 0.036 yes yes no 0 2 yes	HG02133_JS	Pass	0.969	99.89	99.835	25.133	23.838	female	5507454301008100717698	E09	99.844	100	0.236	0.042	yes	yes	yes	no	0	2	yes	
H602353_JC Pass 0.968 99.875 99.782 24.141 22.824 male 5507454301008100717688 F05 99.844 100 0.776 0.031 yes yes no 0 2 yes H603460_JC Pass 0.958 99.829 99.725 24.056 22.894 male 5507454301008100717688 F05 99.844 100 0.276 0.036 yes yes yes no 0 2 yes NA18541_JS Pass 0.97 9.875 98.44 100 0.27 0.036 yes yes yes no 0 2 yes NA18541_JS Pass 0.97 98.875 23.89 female 5507454301008100717688 F05 99.844 100 0.27 0.036 yes yes yes no 0 2 yes NA180541_JS Pass 0.969 99.874 99.824 24.346 female 5507454301008100717688 H05 99.844 100 0.254 0.038 yes yes no 0 2 yes NA19095_T04 Pass 0.68 99.9 99.763 23	HG02589_JS	Pass	0.956	99.78	99.751	24.336	23.467	female	5507454301008100717698	D07	99.844	100	0.27	0.03	yes	yes	yes	no	0	2	yes	
Hol3460/S Pass 0.958 99.82 99.725 24.056 22.894 male 5507454301000100717698 C05 99.844 100 0.326 0.036 jves yes yes no 0 2 yes NA1864_1_S Pass 0.99 99.875 99.804 female 5507454301000100717698 DC5 99.844 100 0.27 0.036 jves yes no 0 2 yes NA18642_T0S Pass 0.969 99.835 99.782 25.393 24.289 female 5507454301008100717698 DC5 99.844 100 0.27 0.036 jves yes no 0 2 yes NA19095_T04 Pass 0.969 99.874 24.289 female 5507454301008100717698 GC5 99.844 100 0.27 0.036 jves yes no 0 2 yes NA19095_T04 Pass 0.969 99.763 23.80 23.175 female 5507454301008100717698 F07 99.844 100 0.241 0.03 yes yes no 0 2 yes NA191915_T04 Pass <t< td=""><td>HG02953_JS</td><td>Pass</td><td>0.968</td><td>99.855</td><td>99.798</td><td>24.141</td><td>22.824</td><td>male</td><td>5507454301008100717698</td><td>F05</td><td>99.844</td><td>100</td><td>0.278</td><td>0.03</td><td>yes</td><td>yes</td><td>yes</td><td>no</td><td>0</td><td>2</td><td>yes</td><td></td></t<>	HG02953_JS	Pass	0.968	99.855	99.798	24.141	22.824	male	5507454301008100717698	F05	99.844	100	0.278	0.03	yes	yes	yes	no	0	2	yes	
NA18641_S Pass 0.97 99.875 99.804 25.188 24.244 fermine 5507454301008100717698 D05 99.844 100 0.27 0.036 yes yes no 0 2 yes NA18642_T05 Pass 0.969 99.835 99.762 25.393 24.289 fermie 5507454301008100717698 G05 99.844 100 0.254 0.036 (yes yes no 0 2 yes NA19095_T04 Pass 0.969 99.763 23.805 23.175 fermie 5507454301008100717698 607 99.844 100 0.24 0.033 (yes yes yes no 0 2 yes NA19095_T04 Pass 0.968 99.9 99.763 23.805 23.175 fermie 5507454301008100717698 609 99.844 100 0.241 0.033 (yes yes yes no 0 2 yes NA19151_C/L Pass 0.962 99.915 99.844 100 0.241 0	HG03460_JS	Pass	0.958	99.82	99.725	24.056	22.894	male	5507454301008100717698	C05	99.844	100	0.326	0.036	yes	yes	yes	no	0	2	yes	
NA18642_T05 Pass 0.969 99.875 99.782 25.393 24.289 fermale 5507454301008100717698 G05 99.844 100 0.254 0.036 yes yes no 0 2 yes NA19095_T04 Pass 0.968 99.964 507454301008100717698 H05 99.844 100 0.254 0.033 yes yes no 0 2 yes NA19015_T04 Pass 0.968 99.9 99.763 23.055 23.175 fermale 5507454301008100717698 F07 99.844 100 0.241 0.033 yes yes no 0 2 yes NA19114_JC	NA18541_JS	Pass	0.97	99.875	99.804	25.188	24.244	female	5507454301008100717698	D05	99.844	100	0.27	0.036	yes	yes	yes	no	0	2	yes	
NA19095_T04 Pass 0.969 99.704 99.65 24.281 23.465 female 5507454301008100717698 H05 99.844 100 0.264 0.033 yes yes no 0 2 yes NA19095_T04 Pass 0.968 99.9 99.763 23.805 23.175 female 5507454301008100717698 F07 99.844 100 0.241 0.03 yes yes no 0 2 yes NA19114_JS Pass 0.962 99.915 99.842 100 0.241 0.03 yes yes no 0 2 yes NA19315_Ctru Pass 0.962 99.915 99.842 100 0.241 0.03 yes yes no 0 2 yes	NA18642_T05	Pass	0.969	99.835	99.782	25.393	24.289	female	5507454301008100717698	G05	99.844	100	0.254	0.036	yes	yes	yes	no	0	2	yes	
NA19114_JS Pass 0.968 99.9 99.763 23.805 23.175 [female 5507454301008100717698 F07 99.844 100 0.241 0.03 [yes yes no 0 2 yes NA19315_Cth Pass 0.962 99.915 99.84 23.732 [female 5507454301008100717698 609 99.844 100 0.241 0.03 [yes yes no 0 2 yes	NA19095_T04	Pass	0.969	99.704	99.65	24.281	23.46	female	5507454301008100717698	H05	99.844	100	0.264	0.033	yes	yes	yes	no	0	2	yes	
NA19315_Cth Pass 0.962 99.915 99.48 24.281 23.732 female 55074543010001071768 009 99.844 100 0.254 0.024 yes yes yes yes 1 2 yes	NA19114_JS	Pass	0.968	99.9	99.763	23.805	23.175	female	5507454301008100717698	F07	99.844	100	0.241	0.03	yes	yes	yes	no	0	2	yes	
	NA19315_Ctrl.	Pass	0.962	99.915	99.84	24.281	23.732	female	5507454301008100717698	G09	99.844	100	0.254	0.024	yes	yes	yes	yes	1	2	yes	
NA19318_Ctrl Pass 0.304 99.684 99.684 99.646 24.602 23.757 male >>0745430100810071/698 HU9 99.844 100 0.295 0.038 yes yes yes yes 1 2 yes	NA19318_Ctrl	Pass	0.964	99.684	99.646	24.662	23.757	male	5507454301008100717698	H09	99.844	100	0.295	0.038	yes	yes	yes	yes	1	2	yes	

Refer to Table 9 for descriptions of each added Sample Table column.

Sample Table	Description
MAPD	Median Absolute Pairwise Difference of log2ratio signals of adjacent copy number (CN) probesets.
WavinessSD	Waviness Standard Deviation of log2ratio signals of adjacent copy number (CN) probesets
CN passes MAPD	"Yes" if the sample's MAPD value is not greater than the MAPD threshold used by CN QC.
CN passes WavinessSD	"Yes" if the sample's WavinessSD value is not greater than the WavinessSD threshold used by CN QC
CN passes QC	"Yes" if the sample passes both MAPD and WavinessSD threshold tests. If "no", the sample reports a CN state of NoCall. For the CN-aware genotyping analysis variation, SNPs from samples that fail CN QC are called without the benefit of CN-aware genotyping.
	Note: For the CN-aware genotyping analysis variation, Non-PAR X, Y, and mitochondrial SNPs use gender and other information to report non-diploid calls, regardless of CN QC status.
Control chosen	"Yes" if the control was listed in the CN Control CEL List file supplied for CN analysis.
Control used	"1" if the control was used to adjust CN signals of all samples in the same plate. The value will be 1 if Control chosen = 1 and CN passes QC = Yes.

A

Sample Table	Description
Number of controls for CN tuning	A count of control samples used to adjust the CN signals for the given sample. This value will be the same for all samples on the same plate.
CN tuned using controls	If Number of controls for CN tuning > 0, then this value is "Yes". This means that CN signals are adjusted based on the signal measured in the control samples from the same plate. If the controls are appropriate for the samples being processed, this adjustment can improve CN calling accuracy.

 Table 9
 Added Sample Table columns after running a CN Analysis

SNP SummaryAfter successfully running a CN-aware genotyping analysis, additional columns
appear within the SNP Summary Table, as shown in Figure 159.

F	Figure 159 SNP Summary Table after a CN Analysis run																			
	Summary Sample Table SNP Summary Table CN Summary Table Cluster Plot CN Region Plot																			
	Select Annotation Manage SNP List 🔻 Change/Revert Calls 💌 Reanalyze 💌 Perform Allele Translation																			
E	Apply View 🔻 Save View Show/Hide Columns 👻 Export 👻 Filters 💌																			
	probeset_id ConversionType CR MinorAlleleFrequency H.W.p- Value FLD HomFLD HetSO HomRO Nclus n_AA n_AB n_BB n_A n_B n_CN0 n_N hemizygous HomHet Call Modified																			
7	AFFX-KIT-000001	PolyHighResol	100	0.464	0.306	20.649	44.464	0.322	2.754	3	39	76	51	0	0	0	0	0	0 False	*
4	AFFX-KIT-000002	PolyHighResol	99.398	0.33	0.482	8.127	17.968	0.199	0.854	3	76	69	20	0	0	0	1	0	0 False	
14	AFFX-KIT-000003	PolyHighResol	100	0.22	0.007	6.331	13.592	0.153	0.76	3	107	45	14	0	0	0	0	0	0 False	
14	AFFX-KIT-000004	PolyHighResol	100	0.319	0.145	14.997	32.701	0.315	2.126	3	81	64	21	0	0	0	0	0	0 False	
1	AFFX-KIT-000005	PolyHighResol	99.398	0.079	1	5.144	11.911	0.128	1.072	3	140	24	1	0	0	0	1	0	0 False	
1	AFFX-KIT-000008	PolyHighResol	100	0.322	0.532	5.368	11.118	0.132	1.123	3	19	69	78	0	0	0	0	0	0 False	
1	AFFX-KIT-000009	NoMinorHom	100	0.075	0.602	20.912		0.247	4.47	2	0	25	141	0	0	0	0	0	1 False	
1	AFFX-KIT-000012	PolyHighResol	100	0.368	0.388	19.026	39.889	0.24	2.763	3	69	72	25	0	0	0	0	0	0 False	
1	AFFX-KIT-000013	PolyHighResol	99.398	0.136	0.19	5.259	10.952	0.193	0.84	3	125	35	5	0	0	0	1	0	0 False	
1	AFFX-KIT-000014	PolyHighResol	100	0.099	0.663	19.334	39.101	0.554	2.898	3	2	29	135	0	0	0	0	0	0 False	-
	P Find in Table Now Count: 129544 Selected: 1 1 <th1< th=""> 1 1 <th1< th=""></th1<></th1<>																			

Refer to Table 10 for descriptions of each added SNP Summary Table column.

Table 10 Added SNP Summary Table columns after CN-aware genotyping is performed

SNP Summary Table	Description
n_A	Count of hemizygous "A" calls.
n_B	Count of hemizygous "B" calls.
n_CN0	Count of ZeroCN calls.
CopyNumIssue (hidden by default)	Copy Number Issue is "1" if the probeset is assigned to the "Other" ConversionType because the haploid or ZeroCN clusters are in an unusual location compared to each other or to the diploid clusters.



CN Summary Table and CN Region Plot

The Genotyping and Best Practices workflows perform predefined region copy number analysis (if the array content and library package support it). If this is the case, a **CN Summary Table** and **CN Region Plot** window tab appear in the Axiom Analysis Suite Viewer, as shown in Figure 160.



CN Summary Table (Overview)

- The CN Summary Table gives a count of samples called with a given copy number state (CN_State) for a given copy number region (CN_Region).
- CN_States that cannot be reported are displayed as empty with a gray background. If the CN_State can be reported but no samples in the batch report that CN State, then the count is 0. As long as the default CN Bins library file is used during analysis setup, the only samples that will report a CN_State of NoCall are those that fail Copy Number QC.
- Selecting a specific CN_Region row in the table updates the associated CN Region Plot.
- The button **Export Copy Number Data** copies several CN reports to another location specified by the user. Note that CN_States for each CN_Region for each sample are also available in a VCF format export of genotyping data, available from the SNP Summary Table tab.

CN Region Plot (Overview)

- The CN Region Plot displays the MedianLog2Ratio of each genotyped sample for the copy number region selected in the CN Summary Table, grouped by affymetrix-plate-barcode.
- Selecting a sample or samples in the CN Region Plot also selects the same samples in the Sample Table and the Cluster Plot, and vice versa.
- Genes with a CN of 2 typically have a MedianLog2Ratio centered at 0, which indicates that the signals of probesets used for CN estimation are similar to the expected signals for a CN=2 sample.
- CN States are called by comparing the MedianLog2Ratio to the thresholds defined in the CN Bins library file selected during analysis setup. CN States cannot be edited in the Viewer.
- Sometimes between-plate assay variation can shift the observed MedianLog2Ratio values away from 0 for CN=2 samples. Fortunately, if the assay includes appropriate control samples on the assay plate, then the copy number signals can be tuned using the control samples. If CN plate correction was done, then the default "Shape By" metric "CN tuned using controls" identifies which samples had their CN measurements adjusted using the plate controls.

Overview and Use of the Best Practices Workflow

The recommended genotyping method is to use information from a batch of samples to improve the calling of individual samples. To this end, the cluster locations in signal space for each probeset (see the Cluster Plot) adapt in a Bayesian fashion to the supplied data. Prior knowledge of cluster locations influences the final calls. The more samples that are supplied in a batch, the more the final cluster locations will be influenced by the supplied data.

The dynamic nature of this genotyping algorithm means that if a given sample's CEL file is genotyped in a group of 24 CEL files, or in a group of 2400 CEL files, you can expect that some SNPs may experience a changed call (call <-> NoCall, or sometimes call 1 <-> call 2). This is more likely to happen if the SNP's data quality is not great (close clusters), if there are strong between-plate effects in cluster positions, or for very low Minor Allele Frequency SNPs where good information on rare genotype cluster positions may not be available before genotyping. Calling accuracy of both rare and common SNPs is improved even for small batch sizes if enough prior knowledge of cluster locations is available.

The batch nature of the genotyping means that if there are some samples of poor data quality in a group of good quality samples, then sometimes the poor quality samples will harm the calling accuracy of the good samples. For this reason, Axiom Analysis Suite's "Best Practices Workflow" can be thought of as a two-step process:

Step 1 Sample QC: Identify and exclude the poor quality sample CEL files. **Step 2 Genotyping**: Genotype only the sample CEL files that pass Step 1.

- Step 1: The Sample QC Workflow performs the following steps:
 - Genotype a small set of SNPs used to uniquely identify each sample ("Signature SNPs"). This method uses static calling boundaries, so a given sample's calls are not influenced by other samples.
 - DishQC: Calculate DishQC metric and exclude CEL files with too small a DishQC from next step. For this method, a given sample's metrics are not influenced by other samples.
 - QC Call Rate: Initial cluster genotyping on remaining samples for selected QC SNPs, to identify and remove any additional CEL files with low QC call rates. For this method, batch information from other samples influences a given samples QC call rates. Genotypes are not stored.
 - Plate QC: Compare the average QC Call Rate of passing samples within a plate against the threshold "Average call rate for passing samples". If the metric is below this threshold, all the samples on the plate will fail Plate QC, and will not be genotyped.
 - Identify Copy Number plate controls:
 - For CN-aware genotyping arrays like PharmacoScan, auto-identify reagent control samples by comparing measured SignatureSNP genotypes from all CEL files against reference genotypes. A control is identified if [number of Signature SNPs with a call is >= "Control comparisons"] AND [Concordance % of signature SNP calls compared with reference genotypes is >= "Control concordance"]. Control samples that pass all sample QC checks will be used in the subsequent Genotyping workflow, unless overridden by the user.
 - For predefined CN region arrays like CarrierScan, select as CN controls all female samples passing sample QC.



- **Step 2:** Genotyping can be run by itself or as part of the Best Practices Workflow. When run as part of the Best Practices Workflow, only CEL files passing Sample QC are genotyped. When run by itself, the user has full control of which samples to genotype. Genotyping does the following steps:
 - For arrays supporting predefined region CN analysis or CN-aware genotyping, collate normalized signals for CN probesets of interest and perform predefined-region CN analysis. This step also calculates CN-specific QC metrics MAPD and wavinessSD. Samples that fail CN QC have their CN states reset to NoCall. If control samples are supplied (by user or by Sample QC step), then plate-based signal correction is performed. Control samples that fail CN QC are not used for plate-based signal correction.

IMPORTANT! If you plan to run only the Genotyping workflow, and if the CN Control CEL List File is an Analysis Settings input option, this file should be supplied. If it is not, then plate signal correction is not performed and CN results may suffer.

The file **[Batch Name]\CNData\CNcontrolSamples.pass.txt** (from the Sample QC step) is supplied for CN analysis during a Best Practices Workflow.

- For CN-aware genotyping arrays like PharmacoScan, CN results are supplied as an input to the next genotyping step.
- Perform genotyping, using either default or user-supplied input options. Genotypes are saved.
- Generate summary statistics on each SNP.



About Allele Translation

Overview

Human genome sequence variation, which includes both single nucleotide polymorphisms (SNPs) as well as more complex structural variation in the form of insertions, duplications and deletions, underlies each individual's response to drugs. Products like PharmacoScan and DMET Plus are designed to enable comprehensive and accurate genotyping of specific polymorphisms involved in drug-metabolizing enzymes and transporters. Axiom Analysis Suite enables conversion of genotype calls to clinically-recognized star nomenclature via Allele Translation. This section explains the organization of the translation reports to help you interpret the translation data. Key concepts such as phase ambiguity and the impact of missing data on haplotypebased allele calling are described.

Gene Table Layout for Haplotyping

To appreciate how haplotyping operates, it is essential to describe the organization of the gene tables (Figure 161 and Figure 162) in the translation library file (*.translation).

F	Figure 161 Biological annotations [Example: Gene table data for markers in CYP1A1]											
				Switch								
				Design			cDNA					
			Probe Set	Strand to	dbSNP RS		Nucleotide	Genome				
	CYP1A1	Reference Link	ID	Report	ID	Defining	Position	Position	Change	Common Name		
	CYP1A1	PMID: 11295847	AM_10774	Y	rs56313657	•6	993G>T	Ch15:75013804	M331	CYP1A1*6_1635G>T(M331I)		
	CYP1A1	PMID: 15618738	AM_10771	Y	rs72547510	•7	1275_1276ir	Ch15:75013093	E426Fra	CYP1A1*7_2345insT		
	CYP1A1	PMID: 15618738	AM_10770	Y	rs72547509	•8	1343T>A	Ch15:75013026	1448N	CYP1A1*8_2413T>A(I448N)		
	CYP1A1	PMID: 8895751	AM_10769	Y	rs1799814	•4	1382C>A	Ch15:75012987	T461N	CYP1A1*4_2452C>A(T461N)		
	CYP1A1	PMID: 9070254	AM_10768	Y	rs1048943	*2C	1384A>G	Ch15:75012985	1462V	CYP1A1*2C_2454A>G(I462V)		
	CYP1A1	PMID: 11295847	AM_10766	N	rs41279188	•5	1390C>A	Ch15:75012979	R464S	CYP1A1*5or*9_2460C>A>T(R464SorC)		
	CYP1A1	PMID: 11295847	AM_10766	N	rs41279188	•9	1390C>T	Ch15:75012979	R464C	CYP1A1*5or*9_2460C>A>T(R464SorC)		
	CYP1A1	PMID: 15618738	AM_10765	Y	rs56240201	*10	1429C>T	Ch15:75012940	B477V	CYP1A1*10_2499C>T(R477V)		
	CYP1A1	PMID: 15618738	AM_10762	N	rs1800031	•3	*595T>C	Ch15:75012235	3'UTR	CYP1A1*3_3204T>C(3'UTR)		
	CYP1A1	PMID: 11295847	AM_10778	N	rs4646422	G45D	134G>A	Ch15:75015305	G45D	CYP1A1_134G>A(G45D)		
	CYP1A1	PMID: 11295847	AM_10776	N	rs34260157	R279V	835C>T	Ch15:75014049	R279V	CYP1A1_1390C>T(R279W)		
	CYP1A1	PMID: 9353182	AM_10775	Y	rs4987133	I286T	857T>C	Ch15:75014027	1286T	CYP1A1_1412T>C(I286T)		
	CYP1A1	PMID: 15618738	AM_10772	Y	rs2856833	F381L	1143C>A	Ch15:75013563	F381L	CYP1A1_1876C>A(F381L)		
	CYP1A1	GBID: NM_00049	AM_10767	Y	rs2278970	A463G	1388C>G	Ch15:75012981	A463G	CYP1A1_2458C>G(A463G)		

Figure 162 Haplotype d	escripti	ions [Ex	kample	e ge	ne ta	able	dat	a foi	r ma	rker	s in	CYF	21A1]			
Common Name	Haplotype	Reference	Variant	-1	*2C	•3	-4	•5	•6	•7	•8	•9	*10	G45E	R279	1286T	' F3
CYP1A1 6_1635G>T(M331I)	Y	G	Т						Т								
CYP1A1"7_2345insT	Y	-	Т							Т							
CYP1A1"8_2413T>A(I448N)	Y	Т	A								A						
CYP1A1"4_2452C>A(T461N)	Y	С	A				A										
CYP1A1*2C_2454A>G(I462V)	Y	A	G		G												
CYP1A1*5or*9_2460C>A>T(R464SorC)	Y	С	A					A									
CYP1A1*5or*9_2460C>A>T(R464SorC)	Y	С	Т									Т					
CYP1A1*10_2499C>T(R477W)	Y	С	Т										Т				
CYP1A1"3_3204T>C(3'UTR)	Y	Т	С			С											
CYP1A1_134G>A(G45D)	N	G	A											A			
CYP1A1_1390C>T(R279W)	N	С	Т												Т		
CYP1A1_1412T>C(I286T)	N	Т	С													С	
CYP1A1_1876C>A(F381L)	N	С	A														A
CYP1A1_2458C>G(A463G)	N	С	G														

Biological annotations: The first set of columns in the table are annotations for the markers in each of the translated genes. Haplotype descriptions: The columns beyond the Common Name field contain information used for interpretation and translation of the gene file.

Following the columns enumerating the Reference and Variant alleles, haplotypes and markers in the gene are listed. In this example, CYP1A1 has 10 haplotypes described and they are named in the column headers. In addition, there are five additional markers for rare variants that can also be identified in this gene. Notice that the first haplotype described in this table is CYP1A1*1A, and that all markers except the last five are haplotyping markers (see the Haplotype field). Markers are characterized as "non-haplotyping" if their state in every reportable haplotype is not known. For example, a variant has been identified in this gene resulting in a non-synonymous change in the protein (CYP1A1_134G>A(G45D)), but the haplotype background of that variant is not available in the literature references used (for example, the Karolinska reference database; http://www.cypalleles.ki.se/).

Figure 161 illustrates the way that haplotypes are called. Notice that the haplotype names appear in the header of each gene table (for CYP1A1, when all markers are tested, this is *1, *2C, *3, *4, etc). Only differences from the reference haplotype appear in the haplotype columns, and the change for the altered base is indicated. For example, CYP1A1*2C contains one difference relative to CYP1A1*1, specifically a mutation 5'-prime to the gene, defined by the probe "CYP1A1*2C_2454A>G(I462V)".

The Defining field in the translation file lists the effect that a variant allele of this marker has. For example, because a variant at the final probe in the table, "CYP1A1_2458C>G(A463G)", results in a structural change in the protein, this marker is flagged with the name of the amino acid change that results: alanine at position 463 of the protein is a glycine in this variant (A463G). Although not shown in this gene, if a marker contains a variant allele in multiple haplotypes, then the marker does not uniquely define a single haplotype. That marker would then have an "N" (No) in the Defining field.

Haplotype field names with a # prefix, and rows with a # prefix in the Probe Set ID field are "commented out", and not used for translation.

Biological Annotations in Translation Reports

The primary function of the translation reporting is to summarize genotypes into commonly recognized variant names. In the case of the CYP450 core gene set, this translates to the Star-nomenclature followed by standardized nomenclature committee direction. Similar names are used by other steering committees such as the two Phase II enzyme genes, N-acetylase genes (NAT1 and NAT2) or the UGT-transferase gene families. Wherever possible, we have attempted to use a standard naming convention for the markers. To facilitate interpretation of the genotyping results, the translation reports provide:

- Reference publications, sequences or dbSNP identifiers for following previously published information about the variant site
- Precise genomic location in a recent genome build for identifying confirmatory genotyping assays
- Notation of protein changes that may result from the mutations in the panel. This field may also indicate whether the variant allele is strategically positioned in the promoter region or causes changes in splice junction sequences in the gene.
- Description of the initial star-allele which the variant was identified. Generally, this corresponds to the Summary flag entry
- Alternative alleles at each marker and whether the defined allele is the Reference base or Variant (corresponding to the altered gene form)

Along with the identified genotypes of the sample, this information provides biological evidence supporting haplotype calls.

Impact of Phase Ambiguity in Haplotyping

It is not unusual for individuals to be heterozygous at more than one defining marker (compound heterozygote genotype) in a gene. When this happens, multiple haplotype pairs may be consistent with resulting profiles, as shown in Figure 163.



In this example, it is clear that the child could have inherited both variant alleles from one parent (in that case their diplotype would be *1/*3A), or could have inherited one variant allele from each of the two parents (in that case the diplotype would be *3B/*3C). Although the *3A haplotype is less common than either *3B or *3C, the translation reports list both potential haplotype pairs in the output reports. One reason for this is that the phenotypes may differ between the two alternative genetic configurations. In this case, three of the four alleles of TPMT have reduced activity: *3A, *3B and *3C, whereas the reference allele, *1 is a normally functioning allele. Phase ambiguity is relatively common in genes with common polymorphisms. Figure 164 on page 143 lists the multiple possible calls due to phase ambiguity that were observed in six HapMap populations, and how often they occurred.

Gene	Call 1	Call 2	Call 3	Call Rate						
CYP1A2	*1A/*1L	*1C/*1F		22.6%						
CYP2B6	*1/*7	*5/*6		2.0%						
CYP2D6	*2/*64	*10/*17		0.3%						
FMO2	*2A/*3	*1/*2C		0.5%						
NAT1	*4/*11	*11C/*30		1.8%						
NAT2	*4/*5E	*5/*6		9.2%						
NAT2	*4/*6J	*6/*7		3.7%						
NAT2	*4/*14D	*6/*14		2.0%						
NAT2	*4/*14F	*5/*14		1.3%						
NAT2	*4/*7D	*7/*14		0.2%						
SLC22A2	*1/*3D	*3A/*6		7.4%						
SLC22A2	*1/*2B	*2A/*3A		6.2%						
SLC22A2	*1/*3E	*2A/*3D	*2B/*6	5.5%						
SLC22A2	*2B/*3D	*3A/*3E		1.3%						
SLCO1B1	*1b/*17	*15/*21		5.9%						
SLCO1B1	*1a/*14	*1b/*4		3.2%						
SLCO1B1	*1a/*15	*1b/*5		3.2%						
SLCO1B1	*1a/*17	*5/*21		2.5%						
TPMT	*1/*3A	*3B/*3C		1.3%						
UGT1A1	*1/*28+60	*28/*60		1.5%						
UGT1A1	*1/*27+28+60+93	*27/*28+60+93		0.5%						
UGT1ACOMMON	*76+79/*IA	*76/*79		8.0%						
UGT2B15	*1/*5	*2/*4		12.9%						

Figure 164 Example: Observed phase ambiguities in DMET Plus in a data set of six HapMap populations with 597 individuals and no children.

It is worth pointing out that the predicted phenotypes of some of these alternative diplotype calls are identical, and in these cases the Phenotype report will then report a single phenotype. When phase ambiguity is encountered and the Phenotype report does not resolve the multiple calls to a single Phenotype Call, follow-up metabolic screening may be merited to differentiate the actual genetic configuration of the test samples.

Diplotype to Phenotype Translation

Allele translations include a Phenotype report if the required metabolizer library file has been selected. The Phenotype report further translates the reported diplotypes (star allele pairs) from a subset of genes in the Comprehensive report into one of several phenotypes (e.g. "Poor Metabolizer"). As the software reads the comprehensive.rpt file, it will try to match Known Call diplotype values for each gene of each sample to one row of the metabolizer library file table. If a match is found, the associated phenotype and allele activities are written to the phenotype.rpt. If a match is not found, a Phenotype Call of "unknown" is reported. More information on this software feature is available in the DMETTM Plus Allele Translation white paper.



IMPORTANT! Users are responsible for reviewing the metabolizer library file for accuracy!

Phenotype Call and Gene Activity interpretations for a Known Call are supported by differing levels of evidence from in vivo and/or in vitro research studies. Refer to metabolizer library file for a list of references. The actual phenotype and gene activities may be dependent on the substrate and dose.

If you do not want to generate a phenotype report, leave the Metabolizer File option blank in the Perform Allele Translation dialog.

If you want to report phenotypes for only a subset of genes, there are two ways to accomplish this:

 Import a custom SNP List into the workspace containing probesets from only the genes of interest. At the point when you normally perform allele translation, select the option to filter to just probesets in this list.

Or

2. Create a copy of the metabolizer file that only contains the genes of interest.

If you want to change what phenotypes are reported for a particular combination of diplotypes, or you would like to change what is written to the header of the Phenotype Translation report, you will need to create and use a custom version of the .metabolizer library file. Instructions for doing this follow.
Creating a Custom Metabolizer Library File

If you choose to create a custom metabolizer table, start with the default metabolizer file as a template, then save a copy of this file using a new name.

IMPORTANT! Use caution if editing the metabolizer file with Microsoft Excel. For example, Excel inserts quotation marks around text containing commas, which may make the file unreadable by Axiom Analysis Suite.

Before using the file with Axiom Analysis Suite, open it in another text editor and remove any unexpected text such as quotation marks.

To be recognized by Axiom Analysis Suite, the file:

- Must have the file extension *.**metabolizer**, where the * indicates your custom text.
- Must be encoded in ANSI, not Unicode or other encoding.
- Must exist in the library folder used by Axiom Analysis Suite.
- Must be selected for use from the **Configuration** -> **Options** menu, in the Translations tab.
- Must be properly formatted. It is recommended that you use a file comparison utility to verify that the only changes between the original and modified files are expected changes.

If you want to add phenotype reporting (for genes not currently in the metabolizer library file) the gene names and star allele names you want to add must exist in the ***.translation** library file used to generate the ***_comprehensive.rpt** file.

B

Metabolizer Library File Format

The **.metabolizer** library file is a tab-delimited text file that can be edited in any text editor. This file consists of a header section followed by a single table. Any rows from the start of the file until the beginning of the main table are considered header rows, and must begin with a pound or hash sign (#). Header rows are optional. Header rows beginning with #%**Info=** is added to the header of the ***_phenotype.rpt** file, so you can put custom text into your reports. The first row that does not begin with # must use the names shown below, be tab separated, and contain only lowercase letters.

gene allele_1 allele_2 phenotype activity_1 activity_2

Additional field names can be added to the first table row, but they will not be used. After the first table row, all rows require a value for the following fields:

gene allele_1 allele_2 phenotype

The **activity_1** and **activity_2** fields can be left blank. See Figure 165 on page 147 for field descriptions.

ary File Format	

Figure 165 field descriptions for the Metabolizer library file						
Metabolizer field	Descripti	on				
gene	The gene be found i report.	name as repor in the *.translat	ted in the co ion library file	mprehensiv e needed to	e report. These generate the c	e values can also comprehensive
allele_1 allele_2	The haplotype name of an allele for a gene as reported in the comprehensive report, e.g. '*2'. A Known Call in the comprehensive report is usually a single pair of alleles, e.g.'*1/*2'. To have this call be matched to a specific row in the metabolizer table, only one of the rows is needed in the following table:					
		gene	allele_1	allele_2	phenotype	
		CYP2D6	*1	*2	NM	
		CYP2D6	*2	*1	NM	
	Axiom Ana above exa	alysis Suite wil ample) AND IF	l report an ei the duplicate	ror IF it dete e rows repor	ects duplicate r t disagreeing p	rows (as in the ohenotypes.
phenotype	The value that should be reported for the associated 'gene allele_1/allele_2' call. The string should be short with no commas, quotes, or whitespace characters. Common phenotype names are:					
		phenotype	definition			
		UM	Ultra-rapic	l metabolize	PL	
		RM	Rapid met	abolizer		
		NM	Normal m	etabolizer		
		IM	Intermedia	ate metaboli	zer	
		PM	Poor meta	abolizer		
		unknown	Unknown	metabolizer		
activity_1 activity_2	The reported gene activity or function level for an allele, e.g. 'normal' or 'decreased'. Activity_1 is for allele_1, and activity_2 is for allele_2. The values in these fields are used to populate the Gene Activity field in the phenotype report, e.g. 'normal/decreased'. If you leave these fields empty, the phenotype report will display '/' for the Gene Activity.					
Optional fields	Axiom An Additional	alysis Suite wil fields may be	l ignore addi used to anno	tional fields otate each ro	in the metaboli ow.	zer file.

Reference Databases Used in Translation Data Curation

The databases used to curate the allele translation gene tables include:

- PharmGKB Stanford University Pharmacogenomics reference database http://www.pharmgkb.org
- Karolinska cytochrome P450 gene standard nomenclature http://www.cypalleles.ki.se
- Database of NAT genes (Democritus University of Thrace) http://nat.mbg.duth.gr
- Database of UGT genes https://www.pharmacogenomics.pha.ulaval.ca/ugt-alleles-nomenclature
- Drug interaction database (University of Indiana) http://medicine.iupui.edu/clinpharm/ddis
- PubMed On-line National Library of Medicine publication database http://www.ncbi.nlm.nih.gov/pubmed

Definitions



Threshold Names

Use the table of definitions below to help select thresholds (when setting up an analysis or regenerating SNP metrics).

Table 11 Threshold Names

Threshold Group	Threshold Name	Description
Sample QC	DQC	A sample's dish QC value must be greater than or equal to 'DQC' to pass sample QC.
Sample QC	QC call_rate	A sample's call rate value must be greater than or equal to 'QC call_rate' to pass genotyping QC.
Sample QC	Percent of passing samples	If a plate's percent of passing samples is smaller than this number, all samples on the plate will show a warning in the Sample Table.
Sample QC	Average call rate for passing samples	A plate's average QC call rate of passing samples must be greater than or equal to this number to pass plate QC.
Sample QC	Control comparisons	CN-aware genotyping parameter. For identifying control samples, this is the minimum number of SignatureSNP probesets compared to a reference.
Sample QC	Control concordance	CN-aware genotyping parameter. For identifying control samples, this is the minimum percent concordance of SignatureSNP calls to a reference.
CN QC	MAPD	For arrays that support copy number (CN) analysis, the Median Absolute Pairwise Difference of log2ratio signals of adjacent copy number (CN) probesets must be less than this value to make CN calls.
CN QC	Waviness SD	For arrays that support copy number (CN) analysis, the Waviness Standard Deviation of log2 ratio signals of copy number (CN) probesets must be less than this value to make CN calls.
SNP QC	species-type	Species type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.
		Note: Selecting 'Human' will assign a probeset with two clusters that are both homozygous to a non-recommended ConversionType like 'Other'.
SNP QC	cr-cutoff	Minimum acceptable call rate.

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Table 11	Threshold Names
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Threshold Group	Threshold Name	Description
SNP QC	fld-cutoff	For autosomal probesets, minimum acceptable FLD value for cluster separation.
SNP QC	het-so-cutoff	Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.
SNP QC	het-so-XChr-cutoff	For probesets on the non-pseudoautosomal regions of chromosome X, the minimum acceptable value for the correctness of the Size (Y position) offset of the female heterozygous cluster.
SNP QC	het-so-otv-cutoff	Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster, possibly indicating a fourth cluster below the heterozygous cluster (OTV).
SNP QC	hom-ro-1-cutoff	Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 1 genotype cluster.
SNP QC	hom-ro-2-cutoff	Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.
SNP QC	hom-ro-3-cutoff	Minimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.
SNP QC	hom-ro	Flag indicating whether the metric HomRO is used in classification.
SNP QC	hom-het	If hom-het flag is FALSE, a probeset with two clusters that are homozygous and heterozygous is assigned a non- recommended ConversionType like 'Other'. Hom-het flag should be TRUE for human and diploid species for a large batch of samples, and should be FALSE for highly inbred samples such as polyploid species.
SNP QC	num-minor-allele-cutoff	Minimum minor allele count for categorizing a probeset as PolyHighResolution.
SNP QC	hom-ro-hap-1-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 1 haploid genotype cluster.
SNP QC	hom-ro-hap-1-XChr-cutoff	For non-PAR X probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 1 haploid genotype cluster.
SNP QC	hom-ro-hap-1-YChr-cutoff	For Y probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 1 haploid genotype cluster.

Table 11 Threshold Names

Threshold Group	Threshold Name	Description
SNP QC	hom-ro-hap-1-MTChr-cutoff	For mitochondrial probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 1 haploid genotype cluster.
SNP QC	hom-ro-hap-2-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 2 haploid genotype clusters.
SNP QC	hom-ro-hap-2-XChr-cutoff	For non-PAR X probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 2 haploid genotype clusters.
SNP QC	hom-ro-hap-2-YChr-cutoff	For Y probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 2 haploid genotype clusters.
SNP QC	hom-ro-hap-2-MTChr-cutoff	For mitochondrial probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters from 0 (Ratio Offset) when a probeset has 2 haploid genotype clusters.
SNP QC	hom-hap-X-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Contrast (X position) of the haploid clusters relative to the homozygous clusters.
SNP QC	hom-hap-Y-lower-cutoff	For autosomal probesets, maximum acceptable value for the correctness of the Size (Y position) of the haploid clusters relative to homozygous clusters.
SNP QC	hom-hap-Y-upper-cutoff	For autosomal probesets, maximum acceptable value for the correctness of the Size (Y position) of the haploid clusters relative to homozygous clusters.
SNP QC	CN0-hap-X-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Contrast (X position) of the ZeroCN cluster relative to the haploid clusters.
SNP QC	CN0-hap-X-YChr-cutoff	For Y probesets, minimum acceptable value for the correctness of the Contrast (X position) of the ZeroCN cluster relative to the haploid clusters.
SNP QC	CN0-hap-Y-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Size (Y position) of the ZeroCN cluster relative to the haploid clusters.
SNP QC	CN0-hap-Y-YChr-cutoff	For Y probesets, minimum acceptable value for the correctness of the Size (Y position) of the ZeroCN cluster relative to the haploid clusters.
SNP QC	CN0-dip-X-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Contrast (X position) of the ZeroCN cluster relative to the diploid clusters.

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Table 11 Threshold Names

Threshold Group	Threshold Name	Description
SNP QC	CN0-dip-Y-cutoff	For autosomal probesets, minimum acceptable value for the correctness of the Size (Y position) of the ZeroCN cluster relative to the diploid clusters.
SNP QC	aaf-XChr-cut	For non-PAR X probesets, maximum acceptable difference in male versus female A-allele frequency. Any probeset that fails is categorized as Other.
SNP QC	fld-XChr-cut	For non-PAR X probesets, minimum acceptable FLD value for diploid cluster separation. Note: FLD is computed differently for non-PAR X probesets than for autosomal probesets.
SNP QC	homfld-XChr-cut	For non-PAR X probesets, minimum acceptable FLD value for male haploid cluster separation.
SNP QC	homfld-YChr-cut	For Y probesets, minimum acceptable FLD value for male haploid cluster separation.
SNP QC	sign-diff-hom-1-cutoff	For multi-allele probesets, minimum acceptable difference between mean log2 signal and background when there is 1 homozygous genotype cluster.
SNP QC	sign-diff-hom-2-cutoff	For multi-allele probesets, minimum acceptable difference between mean log2 signal and background when there are 2 homozygous genotype clusters.
SNP QC	min-mean-cp2-cutoff	For multi-allele probesets, minimum acceptable mean log2 allele signal for samples that appear to have at least 2 copies of that allele.
SNP QC	max-mean-cp2-cutoff	For multi-allele probesets, maximum acceptable mean log2 allele signal for samples that appear to have at least 2 copies of that allele.
SNP QC	min-genotype-freq-samples	Minimum count of samples to support the genotype frequency p-value calculation. This parameter is used if a genotype frequency file is supplied.
SNP QC	genotype-p-value-cutoff	Minimum acceptable value for the genotype frequency p-value calculation. Probesets not meeting this threshold may be categorized as 'UnexpectedGenotypeFreq'. This parameter is used if a genotype frequency file is supplied, and if the count of genotyped samples is at least min-genotype-freq-samples.
SNP QC	priority-order	Priority order of probeset conversion types when performing probeset selection.
SNP QC	recommended	Probesets having ConversionTypes in this recommended set will be included in the recommended probeset list.
SNP QC	y-restrict	Y-restrict is the maximum vertical distance 2 points in 2 clusters can be when matching up points for calculating the edge metric values BB_dis_x_adj and AA_dis_x_adj . Y-restrict is not used to set probeset ConversionType. It is only used during 'Regenerate SNP Metrics' when the 'Generate advanced metrics' option is selected.

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Table 11 Threshold Names

Threshold Group	Threshold Name	Description
PS Supplemental	homfld-cut	Minimum acceptable FLD value for homozygous cluster separation.
PS Supplemental	bb-varx-cut	Maximum acceptable Contrast (X position) variance for BB cluster. Not used if bb-varx-z-cut-enabled is true.
PS Supplemental	bb-vary-cut	Maximum acceptable Size (Y position) variance for BB cluster. Not used if bb-vary-z-cut-enabled is true.
PS Supplemental	ab-varx-cut	Maximum acceptable Contrast (X position) variance for AB cluster. Not used if ab-varx-z-cut-enabled is true.
PS Supplemental	ab-vary-cut	Maximum acceptable Size (Y position) variance for AB cluster. Not used if ab-vary-z-cut-enabled is true.
PS Supplemental	aa-varx-cut	Maximum acceptable Contrast (X position) variance for AA cluster. Not used if aa-varx-z-cut-enabled is true.
PS Supplemental	aa-vary-cut	Maximum acceptable Size (Y position) variance for AA cluster. Not used if aa-vary-z-cut-enabled is true.
PS Supplemental	bb-varx-z-cut-enabled	If true, bb-varx-z-cut is used.
PS Supplemental	bb-vary-z-cut-enabled	If true, bb-vary-z-cut is used.
PS Supplemental	ab-varx-z-cut-enabled	If true, ab-varx-z-cut is used.
PS Supplemental	ab-vary-z-cut-enabled	If true, ab-vary-z-cut is used.
PS Supplemental	aa-varx-z-cut-enabled	If true, aa-varx-z-cut is used.
PS Supplemental	aa-vary-z-cut-enabled	If true, aa-vary-z-cut is used.
PS Supplemental	bb-varx-z-cut	Maximum acceptable Contrast (X position) Z-score for BB cluster. Not used if bb-varx-z-cut-enabled is false.
PS Supplemental	bb-vary-z-cut	Maximum acceptable Size (Y position) Z-score for BB cluster. Not used if bb-vary-z-cut-enabled is false.
PS Supplemental	ab-varx-z-cut	Maximum acceptable Contrast (X position) Z-score for AB cluster. Not used if ab-varx-z-cut-enabled is false.
PS Supplemental	ab-vary-z-cut	Maximum acceptable Size (Y position) Z-score for AB cluster. Not used if ab-vary-z-cut-enabled is false.
PS Supplemental	aa-varx-z-cut	Maximum acceptable Contrast (X position) Z-score for AA cluster. Not used if aa-varx-z-cut-enabled is false.
PS Supplemental	aa-vary-z-cut	Maximum acceptable Size (Y position) Z-score for AA cluster. Not used if aa-vary-z-cut-enabled is false.
PS Supplemental	clustermin	Minimum number of samples in a cluster for the new variance or Z-score value to be calculated.
PS Supplemental	n-minor-hom-cut	Minimum number of minor hom samples for PolyHighResolution classification. PolyHighResolution probesets where the number of minor hom samples is less than n-minor-hom-cut are categorized as nMinorHom.

Table 11 Threshold Names

Threshold Group	Threshold Name	Description
PS Supplemental	hetv-maf-cut	Maximum acceptable heterozygosity parameter. PolyHighResolution probesets with het rate > MAF*hetv-maf- cut are categorized as UnexpectedHeterozygosity.
PS Supplemental	variance-class	List of categories that supplemental filters are used on.

Sample Table

Use the table of definitions below for the fields in the Sample Table.

 Table 12
 Sample Table

Selection	Definition
Sample Filename	CEL file name.
Pass/Fail	Sample quality control status. A sample is called 'Pass' by the Sample QC step if it meets the minimum thresholds for DQC, QC call_rate, and average call rate for passing samples.
DQC	DishQC measures the amount of overlap between two homozygous peaks created by non-polymorphic probes. DQC of 1 is no overlap, which is good. DQC of 0 is complete overlap, which is bad.
call_rate	Percentage of autosomal SNPs with a call other than NoCall. 'SpecialSNP' probesets are excluded, as they are mostly non-autosomal.
QC call_rate	Percentage of autosomal SNPs with a call other than NoCall (measured at the Sample QC step).
het_rate	Percentage of SNPs called AB (i.e. the heterozygosity) for autosomal SNPs.
QC het_rate	Percentage of SNPs called AB (i.e. the heterozygosity) for autosomal SNPs (measured at the Sample QC step).
computed_gender	Computed gender for the sample.
QC computed_gender	Computed gender for the sample (measured at the Sample QC step).
affymetrix-plate-barcode	Plate barcode number.
QC affymetrix-plate-barcode	Plate barcode number (measured at the Sample QC step).
affymetrix-plate-peg-wellposition	Well position of the plate's peg.
QC affymetrix-plate-peg-wellposition	Well position of the plate's peg (measured at the Sample QC step).
Average call rate for passing samples	Average QC Call Rate of passing samples within the plate to which this sample belongs.
Percent of passing samples	Percentage of samples passing sample QC within the plate to which this sample belongs.
cel_filepath	CEL file path.
cel_file_identifier	CEL file identifier.

Table 12 Sample Table

Selection	Definition
affymetrix-array-id	Command Console array identifier.
total_call_rate	Call rate at the default or user-specified threshold for all SNPs.
QC total_call_rate	Call rate at the default or user-specified threshold for all SNPs (measured at the Sample QC step).
total_het_rate	Percentage of SNPs called AB (i.e., the heterozygosity) for all SNPs.
QC total_het_rate	Percentage of SNPs called AB (i.e., the heterozygosity) for all SNPs (measured at the Sample QC step).
hom_rate	Percentage of SNPs called AA or BB (i.e. the homozygosity) for autosomal SNPs.
QC hom_rate	Percentage of SNPs called AA or BB (i.e. the homozygosity) for autosomal SNPs (measured at the Sample QC step).
total_hom_rate	Percentage of SNPs called AA or BB (i.e. the homozygosity) for all SNPs.
QC total_hom_rate	Percentage of SNPs called AA or BB (i.e. the homozygosity) for all SNPs (measured at the Sample QC step).
cluster_distance_mean	Average distance to the cluster center for the called genotype.
QC cluster_distance_mean	Average distance to the cluster center for the called genotype (measured at the Sample QC step).
cluster_distance_stdev	Standard deviation of the distance to the cluster center for the called genotype.
QC cluster_distance_stdev	Standard deviation of the distance to the cluster center for the called genotype (measured at the Sample QC step).
allele_summarization_mean	Average of the allele signal estimates (log2 scale).
QC allele_summarization_mean	Average of the allele signal estimates (log2 scale) (measured at the Sample QC step).
allele_summarization_stdev	Standard deviation of the allele signal estimates (log2 scale).
QC allele_summarization_stdev	Standard deviation of the allele signal estimates (log2 scale) (measured at the Sample QC step).
allele_deviation_mean	Average of the absolute difference between the log2 allele signal estimate and its median across all arrays.
QC allele_deviation_mean	Average of the absolute difference between the log2 allele signal estimate and its median across all arrays (measured at the Sample QC step).
allele_deviation_stdev	Standard deviation of the absolute difference between the log2 allele signal estimate and its median across all arrays.
QC allele_deviation_stdev	Standard deviation of the absolute difference between the log2 allele signal estimate and its median across all arrays (measured at the Sample QC step).
allele_mad_residuals_mean	Average of the median absolute deviation (MAD) between observed probe intensities and probe intensities fitted by the model.





Table 12 Sample Table

Selection	Definition
QC allele_mad_residuals_mean	Average of the median absolute deviation (MAD) between observed probe intensities and probe intensities fitted by the model (measured at the Sample QC step).
allele_mad_residuals_stdev	Standard deviation of the median absolute deviation (MAD) between
	observed probe intensities and probe intensities inted by the model.
QC allele_mad_residuals_stdev	Standard deviation of the median absolute deviation (MAD) between observed probe intensities and probe intensities fitted by the model (measured at the Sample QC step).
A_signal_mean	Average of control A probe raw intensities in the AT channel.
T_signal_mean	Average of control T probe raw intensities in the AT channel.
G_signal_mean	Average of control G probe raw intensities in the GC channel.
C_signal_mean	Average of control C probe raw intensities in the GC channel.
AT_B	AT channel background, which is the average signal of the GC control probes in AT channel.
GC_B	GC channel background, which is the average signal of the AT control probes in GC channel.
AT_S	AT channel signal, which is the average signal of the AT control probes in AT channel.
GC_S	GC channel signal, which is the average signal of the GC control probes in GC channel.
AT_SBR	AT channel signal to background ratio, defined as AT_S/AT_B.
GC_SBR	GC channel signal to background ratio, defined as GC_S/GC_B.
AT_B_IQR	AT channel interquartile range (middle 50%) of background intensities, measured using GC control probes.
GC_B_IQR	GC channel interquartile range (middle 50%) of background intensities, measured using AT control probes.
AT_S_IQR	AT channel interquartile range (middle 50%) of signal intensities, measured using AT control probes.
GC_S_IQR	GC channel interquartile range (middle 50%) of signal intensities, measured using GC control probes.
CV_AT	AT channel median coefficient of variation of replicate probe signals for control probesets.
CV_GC	GC channel median coefficient of variation of replicate probe signals for control probesets.
AT_FLD	Fisher's Linear Discriminant between signal and background in the AT channel, defined as [median_of_AT_probe_intensities – median_of_GC_probe_intensities] ² / [0.5*(AT_S_IQR ² + AT_B_IQR ²)].



Table 12 Sample Table

Selection	Definition
GC_FLD	Fisher's Linear Discriminant between signal and background in the GC channel, defined as [median_of_GC_probe_intensities – median_of_AT_probe_intensities] ² / [0.5*(GC_S_IQR ² + GC_B_IQR ²)].
log_diff_qc	A cross channel QC metric, defined as mean(log(AT_SBR))/ std(log(AT_SBR))+mean(log(GC_SBR))/std(log(GC_SBR)), where signal and background are calculated for control non-polymorphic probes after intensity normalization.
saturation_AT	Fraction of features in the AT channel with intensity greater than or equal to 3800. Features likely to saturate the scanner in the long exposure image will instead be measured in the short exposure image, and all feature signals are scaled using a high dynamic range exposure merging technique.
saturation_GC	Fraction of features in the GC channel with intensity greater than or equal to 3800. Features likely to saturate the scanner in the long exposure image will instead be measured in the short exposure image, and all feature signals are scaled using a high dynamic range exposure merging technique.
cn-probe-chrXY-ratio_gender_meanX	Average probe intensity (raw, untransformed) of X chromosome nonpolymorphic probes.
cn-probe-chrXY-ratio_gender_meanY	Average probe intensity (raw, untransformed) of Y chromosome nonpolymorphic probes.
cn-probe-chrXY-ratio_gender_ratio	Gender ratio Y/X = cn-probe-chrXY-ratio_gender_meanY / cn-probe- chrXY-ratio_gender_meanX.
cn-probe-chrXY-ratio_gender	Predicted gender, based on the value of cn-probe-chrXY- ratio_gender_ratio.
reagent_version	Reagent version used for processing the arrays, based on data intensity values. Note: You can only perform batch genotyping analysis on CEL files processed using the same reagent version.
reagent_discrimination_value	Value assigned to the reagent, and used to determine reagent_version.
user_color	User-supplied color to associate with the sample. For more information, see "Setting User Colors" on page 55.



Concordance Columns

Use the table of definitions below for the columns in the Concordance table.

Table 13Concordance Columns

Column Name	Description
Base	The first sample in the comparison.
Reference	The second sample in the comparison.
#SNPs Called	Number of SNPs common to both sample and reference files with genotype calls.
#Concordant SNP's	Number of called SNPs that have the same genotype call.
%Concordance	Percentage of called SNPs that have the same genotype call.

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Annotations and Columns

Use the table of definitions below to help select the annotations you can add to the SNP Summary Table.

 Table 14
 Annotations and Columns

Column Name	Description
Probe Set ID	The Affymetrix unique identifier for the set of probes used to detect a particular Single Nucleotide Polymorphism (SNP probe sets only).
Affx SNP ID	The Affymetrix unique identifier for the set of probes used to detect a particular Single Nucleotide Polymorphism (SNP). (SNP probe sets only, not available for Axiom [™] Genome-Wide Human Array).
dbSNP RS ID	The dbSNP ID that corresponds to this probe set or SNP. The dbSNP at the National Center for Biotechnology Information (NCBI) attempts to maintain a unified and comprehensive view of known single nucleotide polymorphisms (SNPs), small scale insertions/deletions, polymorphic repetitive elements, and microsatellites from TSC and other sources. The dbSNP is updated periodically, and the dbSNP version used for mapping is given in the dbSNP version field. For more information, see: http://www.ncbi.nlm.nih.gov/SNP/ (SNP probe sets only).
Chromosome	The chromosome on which the SNP is located on the current Genome Version.
Chromosome Start	The nucleotide base start position where the SNP is found. The genomic coordinates given are in relation to the current genome version and may shift as subsequent genome builds are released.
Chromosome Stop	The nucleotide base stop position where the SNP is found. The genomic coordinates given are in relation to the current genome version and may shift as subsequent genome builds are released.
Strand	Genomic strand that the SNP resides on.
Cytoband	Cytoband location of the SNP derived from the SNP physical map and the chromosome band data provided by UCSC.
Strand Vs dbSNP	Indicates whether the SNP is on the same or reverse strand as compared to dbSNP (SNP probe sets only).
ChrX pseudo-autosomal region	SNPs on the X Chromosome which are mapped to the two pseudo-autosomal region have a value of 1 or 2 in this field. All other SNPs are indicated by 0. A value of "1" indicates that the marker maps to the PAR-1 region and a value of "2" indicates that the marker maps to the PAR-2 region. A value of "0" indicates that the marker does not map to either of the two PAR regions.
Probe Count	The total number of probes in the probe set.
Flank	The nucleotide sequence surrounding the SNP. This is a 33-mer sequence with 16 nucleotides on either end of the SNP position. The alleles at the SNP position are provided in the brackets (SNP probe sets only).

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Column Name	Description
Allele A, Allele B	At array (or underlying database) design time, the following naming convention is used to assign allele nucleotide bases to the "Abstract" allele codes "A" and "B":
	 SNPs are fixed on the forward strand of the design-time reference genome. For AT or CG SNPs (SNP alleles are A/T or C/G), the alleles are named in alphabetical order (A and C are the "A" alleles, in these cases);
	3. For non-AT and non-CG SNPs, allele A is A or T, allele B is C or G;
	 4. For indels, allele A is -, allele B is the insertion. 5. For multi-base alleles, the alleles are named in alphabetical order. (For [AGT/TTA], AGT would be "Allele A". For [GGT/TTA], GGT would be "Allele A".)
Ref Allele, Alt Allele	The reference allele and alternative alleles are specified according to the current reference genome build. The value of Ref Allele could be "-", which indicates an insertion after the specified position. Otherwise it is the sequence of the allele of the marker which matches the current reference genome. The value of Alt Allele could be "-", which indicates that the variant is a deletion with respect to the current genome build. Otherwise it is the sequence(s) of the allele(s) of the marker which does not match the current reference genome. If neither allele of the marker matches the current genome build sequence, then the value of the Ref Allele is set to "." and the value of the Alt Allele(s) is set to <allele_1>/<allele_2>, where <allele_1> and <allele_2> are the alleles of the marker. If the current genome build position of the marker is unknown then the value of the Ref Allele and Alt Allele are set to "" to denote missing information.</allele_2></allele_1></allele_2></allele_1>
Associated Gene	SNPs were associated with human genes by comparing the genomic locations of the SNPs to genomic alignments of human mRNA sequences. In cases where the SNP is within a known gene, NetAffx reports the association. Additionally, for genes with exon or CDS annotations, NetAffx reports whether or not the SNP is in an exon, and in the coding region. If the SNP is not within a known gene, NetAffx reports the closest genes in the genomic sequence, and the distance and relationship of the SNP relative to the genes. A SNP is upstream of a gene if it is located closer to the 5' end of the gene and is downstream of a gene if it is located closer to the 3' end of the gene.
Genetic Map	Describes the genetic location of the SNP derived from three separate linkage maps (deCODE, Marshfield, or SLM). The physical distance between the markers is assumed to be linear with their genetic distance. The genetic location is computed using the linkage maps from the latest physical location of the SNP and the neighboring microsatellite markers (SNP probe sets only).
Microsatellite	Describes the nearest microsatellite markers (upstream, downstream and overlapping) for the SNP.
Enzyme Fragment	Lists the enzyme, the restriction fragment containing the SNP and the fragment length. The Whole Genome Assay protocol detects SNPs that are contained within the genomic restriction fragments to simplify the sequence background for genotyping arrays (not available for Axiom Genome-Wide Human Array).
Copy Number Variation	When available, a description of Copy Number Variation Region (CN) probe sets as described by the Database of Genomic Variants (not available for Axiom Genome-Wide Human Array).

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Table 14	Annotations and Columns
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Column Name	Description
SNP Interference	This column is for Copy Number probe sets. It indicates whether or not a known SNP overlaps a copy number probe (CN probe sets only, not available for Axiom Genome-Wide Human Array).
In Final List	This column annotates extended content for genotyping arrays. A value of "1" indicates that the marker is included in the final version of the library file and a value of "0" indicates that the marker is not included in the final version of the library file (SNP probe sets only, not available for Axiom Genome-Wide Human Array).
% GC	The fraction of bases that are G or C in a window of 250,000 bases to each side of the SNP or CN position. All positions that are nearer to the end than 250,001 are set to the value of the position at 250,001 from that end. Position and chromosome values for SNPs and CN probes were mapped to the position of bases in the FASTA files for the build of the genome used in this release of NetAffx, and these bases were then used for all calculations (not available for Axiom Genome-Wide Human Array).
Heterozygous Allele Frequencies	Describes the heterozygous frequency of the allele from Yoruba, Japanese, Han Chinese and CEPH studies using the Affymetrix genotyping arrays. (SNP probe sets only)
Allele Sample Size	Sample size used for Allele Frequency estimates (SNP probe sets only).
Allele Frequencies	Describes the major and minor frequency of the allele from Yoruba, Japanese, Han Chinese and CEPH studies using the Affymetrix genotyping arrays (SNP probe sets only).
Minor Allele	Indicates the Minor Allele of a SNP (SNP probe sets only).
Minor Allele Frequency	The Minor Allele Frequency of a SNP (SNP probe sets only).
OMIM ID	Furnishes OMIM and Morbid Map IDs and their respective gene titles. This database contains information from the Online Mendelian Inheritance in Man [®] (OMIM [®]) database, which has been obtained under a license from the Johns Hopkins University. This database/product does not represent the entire, unmodified OMIM [®] database, which is available in its entirety at www.ncbi.nlm.nih.gov/omim/.
Ordered Alleles	A list of alleles alphabetically ordered by abstract allele code. For bi-allele probesets, the order is "Allele A // Allele B" For multi-allele probesets, the order is "Allele A // Allele B // Allele C // Allele D", with the number of alleles equal to Allele Count.
Allele Count	The number of measured alleles. For bi-allele probesets the count is 2. For multi-allele probesets, the count is 3 or more.



SNP Summary Table Definitions

Use the table of definitions below to help select your SNP Summary Table columns.

Note: Most SNP Summary Table metrics are calculated only for the subset of samples reported by the column 'gender_metrics'. Refer to the gender_metrics description for more information.

Note: Some of the metrics defined in the following table are not displayed by default in the SNP Summary Table. Select **Show/Hide Columns** or **Apply View > All Columns View** to access more metrics. Some metrics are reported only after selecting the option(s) **Generate advanced metrics** or **Run PS Supplemental**, when choosing to **Regenerate SNP Metrics**. Some metrics are reported only if multi-allele probesets are present, or if CN-aware genotyping was performed.

Note: A few metrics are calculated only for multi-allele probesets. Most metrics are calculated only for biallele probesets.

 Table 15
 SNP Summary Table Metrics

Column Name	Description
ConversionType	Probeset classification
CR	Call rate (CR) is the percentage of samples with a genotype call other than "No Call" for the SNP. Note: Call Rate on non-pseudoautosomal regions of chromosome X is reported only for female samples. Call Rate on chromosome Y is reported only for male samples.
MinorAlleleFrequency	The allele frequency for the A allele is calculated as:
	$P_A = \frac{(\# AA \ Calls + 0.5 \ *AB \ Calls)}{Total \ \# \ Calls}$
	Where the Total # Calls does not include the No Calls. The B allele frequency is .
	PB = 1 - PA
	The minor allele frequency is the Min(PA, PB).

 Table 15
 SNP Summary Table Metrics

Column Name	Description
H.W.p-Value	Hardy Weinberg p-value is a measure of the significance of the discrepancy between the observed ratio or heterozygote calls in a population and the ratio expected if the population was in Hardy Weinberg equilibrium. There are two statistical tests used for HWE. When AA, AB, and BB counts are
	all >=10, a Chi-squared test is used. When one or more of the AA, AB, and BB counts are <10, an Exact test is used. An Exact test means that the p-value is calculated exactly and not approximated from a population distribution.
	$x^{2} = \frac{(f^{2}aa - fa)^{2}}{f^{2}aa} + \frac{(2faafbb - fab)^{2}}{2faafbb} + \frac{(f^{2}bb - fb)^{2}}{f^{2}bb}$
	Where: $fa = \frac{(\#AA \ Calls \)}{Total \ \# \ Calls}$
	$fb = \frac{(\#BB \ Calls)}{Total \ \# \ Calls}$
	$faa = \frac{(\#AA \ Calls + 0.5 * \# AB \ Calls)}{Total \# Calls}$ $(\#BB \ Calls + 0.5 * \# AB \ Calls)$
	$fbb = \frac{(\#BB Calls + 0.5 + \#AB Calls)}{Total \# Calls}$
	$fab = \frac{(\#AB \ Calls)}{Total \ \# \ Calls}$
	$PHW = CDF(x^2)$
	Where CDF is the Cumulative Distributive Function for the chi-squared distribution.
	The Exact test used is the one implemented in R package "HardyWeinberg" for more information see: [2] Haldane, J., 1954. An exact test for randomness of mating. J. Genet. 52 631-635.
	[3] Levene, H., 1949. On a matching problem arising in genetics. Ann. Math. Stat. 20 91-94.
H.W.statistic	H.W.statistic is 1 if H.W.p-Value is calculated using an exact test. H.W.statistic is 0 if the chi-squared test is used. Refer to H.W.p-Value definition (above) for more information.
minGenotypeFreqPval	The minimum probability that any of the clusters present for a probeset have more observations than the supplied expected values. If the ConversionType is 'UnexpectedGenotypeFrequency' then minGenotypeFreqPval is smaller than the SNP QC parameter genotype-p-value-cutoff. The metric minGenotypeFreqPval is available for supported arrays if a genotype frequency file is supplied for SNP QC, and if the genotyped sample batch size is at least as large as the SNP QC parameter 'min-genotype-freq-samples'.



Table 15	SNP Summary Table Metrics
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Column Name	Description
minGenotypeFreqCluster	The genotype cluster for which minGenotypeFreqPval is reported.
FLD	Fisher's Linear Discriminant (FLD) is a measure of the cluster quality of a probeset. High-quality probeset clusters have well-separated centers, and the clusters are narrow. FLD is measured from the SNP posteriors produced during genotyping. FLD is undefined if either the heterozygous or one of the homozygous clusters is empty.
HomFLD	HomFLD is a version of FLD computed for the homozygous genotype clusters. HomFLD is undefined for probesets without two homozygous clusters.
HomFLD_hap	HomFLD_hap is a version of HomFLD computed for the haploid genotype clusters, but only for samples specified by gender_metrics. HomFLD_hap is undefined for probesets without two haploid clusters.
HetSO	Heterozygous Strength Offset measures how far the heterozygous cluster center sits above the homozygous cluster centers in the Size dimension (Y position). Low HetSO values are produced either by misclustering events or by the inclusion of samples that contain variations from the reference genome. Most well-clustered diploid SNPs have positive HetSO values.
HomRO	Homozygote Ratio Offset is the distance to zero in the Contrast dimension (X position) from the center of the homozygous cluster that is closest to zero. If there is only one homozygous cluster, HomRO is the distance from that cluster center to zero on the Contrast dimension.
HomRO_hap	HomRO_hap is a version of HomRO computed for haploid clusters, but only for samples specified by gender_metrics. HomRO_hap will report -10 otherwise.
nMinorAllele	The count of minor alleles, which is one for each heterozygous call, one for each minor haploid call, and two for each minor homozygous call.
Nclus	The number of genotype clusters.
n_AA	The number of AA calls.
n_AB	The number of AB calls.
n_BB	The number of BB calls.
n_A	The number of A calls (haploid).
n_B	The number of B calls (haploid).
n_CN0	The number of ZeroCN calls (zero copy number).
n_NC	The number of NoCall calls, including NoCall_1 (haploid).
AA.meanX	Average Contrast (X position) for AA cluster.
AA.meanY	Average Size (Y position) for AA cluster.
AB.meanX	Average Contrast (X position) for AB cluster.
AB.meanY	Average Size (Y position) for AB cluster.
BB.meanX	Average Contrast (X position) for BB cluster.

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Table 15	SNP Summary Table Metrics
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Column Name	Description
BB.meanY	Average Size (Y position) for BB cluster.
hemizygous	Hemizygous flag is 1 if the probeset measures chromsome Y or mitochondrial DNA, indicating that diploid genotypes are not possible. Otherwise the flag is 0.
BB_dis_x_adj	Smallest Contrast (X position) distance between any BB cluster sample and any neighbor cluster sample, adjusted to only comparing the sample pairs that have a smaller Size (Y position) distance than y_restrict parameter.
AA_dis_x_adj	Smallest Contrast (X position) distance between any AA cluster sample and any neighbor cluster sample, adjusted to only comparing the sample pairs that have a smaller Size (Y position) distance than y_restrict parameter.
freq_diff	A-allele frequency difference between male and female samples.
HomHet	HomHet flag is 1 if, when two diploid genotype clusters are present, one cluster is homozygous and the other is heterozygous. Otherwise the flag is 0.
BB.varX	Contrast (X position) variance for BB cluster.
BB.varY	Size (Y position) variance for BB cluster.
AB.varX	Contrast (X position) variance for AB cluster.
AB.varY	Size (Y position) variance for AB cluster.
AA.varX	Contrast (X position) variance for AA cluster.
AA.varY	Size (Y position) variance for AA cluster.
BB.varX.Z	Contrast (X position) variance Z-score for BB cluster.
BB.varY.Z	Size (Y position) variance Z-score for BB cluster.
AB.varX.Z	Contrast (X position) variance Z-score for AB cluster.
AB.varY.Z	Size (Y position) variance Z-score for AB cluster.
AA.var.X.Z	Contrast (X position) variance Z-score for AA cluster.
AA.var.Y.Z	Size (Y position) variance Z-score for AA cluster.
CopyNumIssue	CopyNumIssue flag is 1 if the probeset is categorized as 'Other' because of unusual locations of the haploid or ZeroCN clusters. Otherwise the flag is 0.
BestProbeset	BestProbeset flag is available when multiple probesets are mapped to the same SNP (Affy-SNP-ID) by a ps2snp file. A probeset is selected based on the priority order of the conversion types. BestProbeset flag is 1 when it is the best or only probeset for a SNP. Otherwise the flag is 0.
BestandRecommended	BestandRecommended flag is 1 if BestProbeset is 1 and the ConversionType belongs to the Recommended set of conversion types. Otherwise the flag is 0.

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Column Name	Description
gender_metrics	Not all probeset metrics are reported using all samples. Number of clusters (Nclus) is calculated on all samples. See the Call Rate (CR) definition for samples used. The remaining metrics are calculated using the sample set described in gender_metrics. If gender_metrics is 'all', all samples are used. If gender_metrics is 'female', only female samples are used. If gender_metrics is 'male', only male samples are used. If gender_metrics is 'diploid', then haploid and ZeroCN samples are excluded before calculation.
MinMean_cp_2	For multi-allele probesets, the minimum mean log2 allele signal for samples that appear to have at least two copies of that allele. If this value is not between the thresholds for the SNP QC parameters 'min-mean-cp2-cutoff' and 'max-mean-cp2-cutoff', then the probeset fails this QC test.
minSigBgndDiffHom	For multi-allele probesets, minimum difference between mean log2 signal and background for the homozygous genotype clusters. If this value is less than the threshold for the appropriate SNP QC parameter 'sign-diff-hom-1-cutoff' or 'sign-diff-hom-2-cutoff', then the probeset fails this QC test.
count_ma_A, count_ma_B, count_ma_C,	For multi-allele probesets, the count of the associated allele. For example, if there are only two samples with the calls AA and AC, then count_ma_A is 3 and count_ma_C is 1.
Call Modified	Call Modified flag is True if any calls for this probeset are changed since the batch results were first created. If no calls are changed the flag is False.

Table 15SNP Summary Table Metrics

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