# Automated Axiom<sup>™</sup> Analysis v2.0 USER GUIDE

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#### Revision history: MAN1000174

Revision	Date	Description
A00	May 2024	Initial release in Thermo Fisher Scientific document control system. MAN1000174 supersedes legacy Affymetrix publication number 703513 and Revision 3 is now A00 to comply with new numbering guidelines. v2.0 release: Documented new user interface and features, including the BloodGenomiX Reporter, Multiplate batching, Parallelization, and additional extension workflows.
2	January 2022	v1.1 release: Added Best Practices with Dual Workflow feature. Optional settings for Sample Name Mapping, Filter File, SNP List Filter, SNP Mapping File, and SNP Allele Swap are now enabled for arrays that have AxLE Export enabled. Option to include SNP Metrics in the AxLE SNP Map Export file.
1	October 2019	Initial release.

#### Important Software Licensing Information

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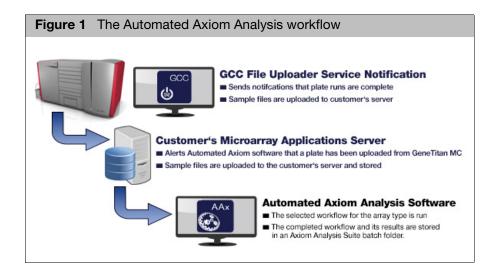
# **Getting started**

## Introduction

Automated Axiom Analysis (AutoAx) enables analysis of the CEL files upon completion of a plate scanning. It requires the GeneChip Console (GCC) File Uploader Service application that resides on the GeneTitan MC instrument workstation.

**IMPORTANT!** Automated Axiom users are expected to be familiar with Axiom Analysis Suite and have its User Guide (P/N MAN0027928) on hand for reference and detailed instructions.

Upon completion of plate scanning to the Microarray Applications, the GCC File Uploader Service sends a notification. Duplicates of the CEL and ARR files are uploaded to the designated folder on the Microarray Applications. AutoAx is alerted that CEL files are available for analysis, then it runs the analysis based on the workflow that was selected for that specific array type.



**Note**: Prior to the first GTMC instrument run, set up an analysis configuration and set it as the default workflow for each array that will be run. When Automated Axiom runs and encounters that array, it will automatically run it. No user intervention is required.

**Note**: Specific library files are required for Automated Axiom. Contact your Thermo Fisher support person for more information.



## Launching AutoAx

**IMPORTANT!** AutoAx has been validated to be compatible with Chrome. Using a different Internet browser is not recommended, as incompatibility issues may occur.

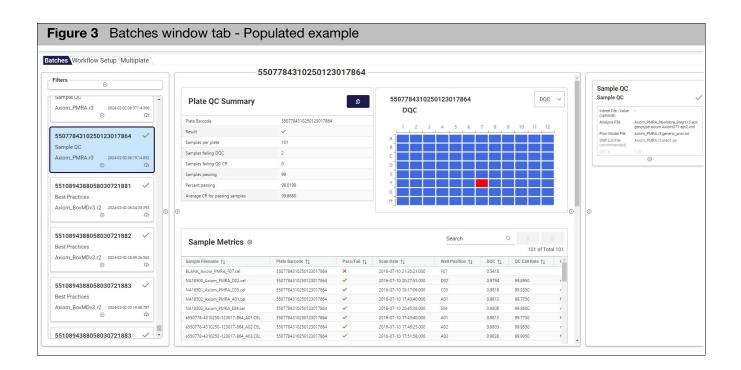
1. Click on the link provided by your IT team.

The Microarray Applications web page opens and displays the Automated Axiom Analysis application card. (Figure 2)

Figure 2         Microarray Applications web page				
appliedbiosystems	Microarray Applications			
AAx Automated Axiom Analysis				
For high throughput plate analysis of Axiom data. Product Information Release Notes Open				

- Click the Product Information link to learn more about the software.
- Click the Release Notes link for information about this v2.0 release.
- 2. Click Open.

The Automated Axiom web page opens (in the same browser window). The **Batches** window tab appears by default. (Figure 3)



# Navigating AutoAx

AutoAx consists of three large window tabs. The following chapters (starting with "Batches" on page 11) detail how to navigate and use each window tab's unique workflows, features, and options.

## About, Help, and Configurations menu

Click on (far upper right).
 About, Help, and Configurations appears.

### About

1. Click About.

An About box appears.

- 2. Click on the drop-down arrow to display the multiple sub-applications and their versions that make up this release.
- 3. Click **OK** to close the window.

### Help

1. Click Help.

The Thermo Fisher Scientific Microarray Analysis Software web page opens.



## Configurations

1. Click Configurations.

The Batch Download Options window appears. (Figure 4)

### Customizing a batch download

1. Click an available batch download option toggle to turn it on or off.

**Note:** Each option you toggle is auto-saved. This custom option set will also be used for future batch downloads.

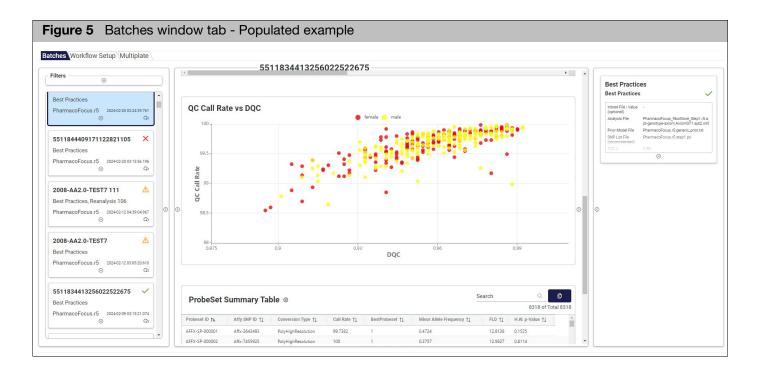
Figure 4	Batch Download Options	
Downloads	Batch Download Options	^
	Full Batch	On 🔵
	CEL/ARR files	Off
	Allele Translation	Off
	AxLE	Off
	ContaminationQC	Off
	HLA	Off
	Sex Check	Off
	VCF	Off
	Logs	Off

2. Click on the **Batches** tab to close the window.

# **Batches**



Use this window tab to view the comprehensive results of a successfully completed batch run, as shown in Figure 5.



# Using the Batches window tab

### **Batches pane**

- Batches are displayed in the far left pane. (Figure 6)
- The card with a blue background represents the batch currently displayed.



55108943880580	30721882 103 🗸
Best Practices, Rean	alysis 102
Axiom_BovMDv3.r2	2023-07-27 14:43:35.346
ି	۵ (C
55108943880580	30721882 🗸
Best Practices	
Axiom_BovMDv3.r2	2023-07-27 13:10:56.898
େ	۵ (G
55077843570060	50419716 🔥
Best Practices	
Axiom PMRA.r3	2023-07-27 11:13:20.377

Card symbols:



- Click the batch card's **Download** icon to copy its data to the **Downloads** folder.
- Click each card's down arrow button to display additional batch information and warning or error details.
- Click the pane's right arrow button to view all batch cards. (Figure 7) Click on the left arrow button (far middle right) to return to the default three pane view.

**Note:** If the batch is a multiplate batch, the barcode is replaced with a user defined batch name.

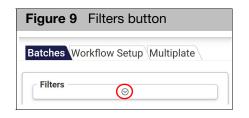
Click on the Switch to Table View button to view all cards in a table view. (Figure 8) Click on a column header(s) to sort the data. Click on the Cloud icon next to the batch to download it.

2

workflow Setup Multiplate						
lters						_
0	J					
5510894388058030721883	5510894388058030721883	~	5510894388058030721883	~	5510894388058030721883	C
Best Practices	Best Practices		Best Practices		Best Practices	
Axiom_BovMDv3.r2 2024-02-19 07:04:37.271	Axiom_BovMDv3.r2	2024-02-14 07:32:50.659	Axiom_BovMDv3.r2	2024-02-14 05:04:04:495	Axiom_BovMDv3.r2	2024-02-13 13:16:08.78
· · · · · · · · · · · · · · · · · · ·	0	Ģ		Ģ	•	G
5510894388058030721883	5510894388058030721883	~	5510894388058030721883	~	5510894388058030721883	
lest Practices	Best Practices		Best Practices		Sample QC	
xiom_BovMDv3.r2 2024-02-09 03:03:45.056	Axiom_BovMDv3.r2	2024-02-09 03:02:26.475	Axiom_BovMDv3.r2	2024-02-09 02:56:02.836	Axiom_BovMDv3.r2	2024-02-08 01:56:11.7
⊖ û.	0	G	⊘	G	0	a
5510894388058030721883	5510894388058030721883	~	5510894388058030721883	~	boymd	
lest Practices	Sample QC		Best Practices with Dual Workflow		Best Practices	15
xiom_BovMDv3.r2 2024-02-08 01:58:56.843	Axiom_BovMDv3.r2	2024-02-08 01:54:41.443	Axiom_BovMDv3.r2	2024-02-05 09:20:06.062	Axiom_BovMDv3.r2	2024-02-02 08:57:23.20
⊖ û.	0	Ģ		Q	0	۵ ۵
510894388058030721881 8	5510894388058030721881	~	5510894388058030721882	~	5510894388058030721883	
est Practices, Reanalysis 4	Best Practices		Best Practices	· · · ·	Best Practices	
Axiom_BovMDv3,r2 2024-02-02 07:37:17.608	Axiom_BovMDv3.r2	2024-02-02 06:04:33.393	Axiom_BovMDv3.r2	2024-02-02 05:59:26.965	Axiom_BovMDv3.r2	2024-02-02 03:19:58.78
≪xioin_bovwibv3.12 ⊙ ©	AXION_BOVMDV3.12 ⊙	202402-02 00.04.33.395 D	Ø	2020F02F02 03:39:20:903	ØX1011_B0000003.12 ⊗	202002-02-03-19-30-70

	panded (All) Ta						
						Search	Q 🕑
							286 of Total 286
ID †1	Array Type 🏗	Group Name 1	Workflow 1	Created 1	Analysis Status †↓		Results
228	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-29 06:03:49:520	success		ര
201	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-21 09:16:23.122	success		G
200	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-21 09:05:11.689	success		େ
199	Axiom_BovMDv3.r2	5510894388057030721314	Best Practices	2024-02-21 08:49:17.270	success		୍ୱ
195	Axiom_BovMDv3.r2	5510894388057030721314	Best Practices	2024-02-21 07:41:33.681	error		G
194	Axiom_BovMDv3.r2	5510894388057030721314	Best Practices	2024-02-21 07:40:08.287	error		(G)
184	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-21 03:04:30.607	success		G
180	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-20 03:17:53.442	error		G
178	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-20 03:17:10.798	warning		(Ga
171	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-19 07:04:37.271	success		(Ga
133	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-14 07:32:50.659	success		G
131	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-14 05:04:04:495	success		G
128	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-13 13:16:08.783	pending		G
88	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-09 03:03:45.056	success		G
87	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-09 03:02:26.475	success		G
82	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-09 02:56:02.836	success		G
74	Axiom_BovMDv3.r2	5510894388058030721883	Sample QC	2024-02-08 01:56:11.714	success		G
73	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-08 01:58:56.843	success		G
72	Axiom_BovMDv3.r2	5510894388058030721883	Sample QC	2024-02-08 01:54:41.443	success		a
20	Axiom_BovMDv3.r2	5510894388058030721883	Best Practices with Dual Workflow	2024-02-05 09:20:06.062	success		G
12	Axiom_BovMDv3.r2	boymd	Best Practices	2024-02-02 08:57:23:209	success		G
8	Axiom_BovMDv3.r2	5510894388058030721881 8	Best Practices, Reanalysis 4	2024-02-02 07:37:17.608	success		a
4	Axiom_BovMDv3.r2	5510894388058030721881	Best Practices	2024-02-02 06:04:33.393	success		G
3	Axiom_BovMDv3.r2	5510894388058030721882	Best Practices	2024-02-02 05:59:26.965	success		(A)
2	Axiom BovMDv3.r2	5510894388058030721883	Best Practices	2024-02-02 03:19:58.787	success		G,

**Filtering batches** 1. At the main Batches window tab, click the **down arrow** button under **Filters** (Figure 9) to display the filters.



2. Click on the radio button(s) (Figure 10) to customize your filtering options, then click the **up arrow** button to hide your filter(s).

Figure 10	Filters opti	ons
Filters Sort By ID Array Type Name Workflow Created Analysis Status	Order Ascending Descending	Period All Cast 30 Days Cast 90 Days
	0	

## Viewing a selected batch

# Plate QC summary table

The Plate QC Summary table (Figure 11) contains detailed QC information for each plate that is run in the batch. **Note:** Displayed information varies with each analysis run.

Figure 11 Plate QC Summary table				
Plate QC Summary	C			
Plate Barcode	5510894388058030721883			
Result	$\checkmark$			
Samples per plate	78			
Samples failing DQC	0			
Samples failing QC CR	0			
Samples passing	78			
Percent passing	100			
Average CR for passing samples	99.4120			

The Plate QC Summary table columns and its descriptions are detailed in Table 1.

Column	Description
Plate Barcode	Barcode of the plate that was analyzed in the batch.
Result	Based on QC thresholds set in analysis configuration template. <b>Note:</b> A plate is either Pass or Fail.
Samples per plate	Count of samples in the plate.
Number of Dual Workflow samples	Records the samples that went through the Dual Workflow analysis.
Samples failing DQC	Count of samples with DQC values that were lower than threshold set for Dish QC.
Samples failing QC CR	Count of samples with QC CR values that were lower than threshold set for QC Call Rate.
Samples passing	Count of samples that pass DQC, QC Call Rate and Plate QC.
Percent passing	Percent of samples that pass DQC, QC Call Rate and Plate QC.
Average CR for passing samples	Average call rate for samples that pass QC.
Number of samples failing ContaminationQC *	Count of samples that did not pass ContaminationQC thresholds. * BloodGenomiX Reporter analyses only.
Number of BGX Reports used *	Number of BGX reports used per plate. * BloodGenomiX Reporter analyses only.
Sex Check *	Number of samples with confirmed computed sex. * BloodGenomiX Reporter analyses only.

### Table 1 Plate QC Summary table

## Copying the table

- 1. Click the button to copy the table.
- 2. Open Word or an Excel file.
- 3. Press **Ctrl v** to paste the table data.

### **Plots**

1. The DQC plot appears by default. Click the drop-down (Figure 12) to select a different plot.

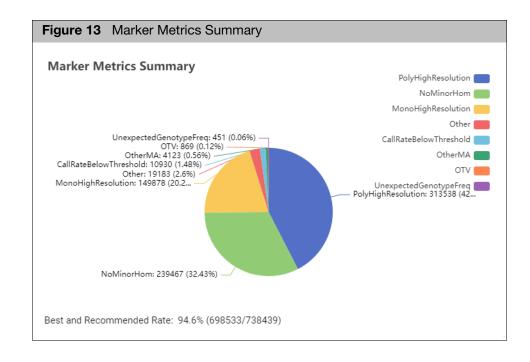
Fig	ure	12	Plo	ots c	drop	o-do	wn						
5	50778 D	43562 QC	20004	29199	976							DQC QC Ca	O II Rate
	1	2	3	4	5	6	7	8	9	10	11	DQC	
A													_
В													0
С													0
D													
E													
F													
G													
н													

Each sample value is shown in its well location.

- Wells with failed samples are shown in red.
- A white well denotes an empty well or one that was not calculated.
- Mouse over a well to view its details.

**Note:** Batches that include a Copy Number will display MAPD and WavinessSD plate plots. BloodGenomiX Reporter analyses will display ContaminationQC plate plots.

1. Hover over the **Marker Metrics Summary** (i) button (Figure 13) to display its pop up window.



2. Click outside the pop up window to hide it.

# Sample metrics table

Marker metrics

summary

The Sample Metrics table's (Figure 14) default columns and descriptions are detailed in Table 2.

2

Figure 14 Sample	e Metrics table					
Sample Metrics				Search	Q	
						384 of Total 384
Sample Filename †↓	Plate Barcode ↑↓	Pass/Fail †↓	Scan Date ↑↓	Well Position ↑↓	DQC †↓	QC Call Rate ↑↓
551282-4428910-120522-828_A01.CEL	5512824428910120522828	~	2021-01-19 14:47:53.000	A01	0.9704	99.5568
551282-4428910-120522-828_A02.CEL	5512824428910120522828	~	2021-01-19 14:50:08.000	A02	0.9723	99.9229
551282-4428910-120522-828_A03.CEL	5512824428910120522828	~	2021-01-19 14:50:54.000	A03	0.9783	98.7283
551282-4428910-120522-828_A04.CEL	5512824428910120522828	~	2021-01-19 14:51:41.000	A04	0.9704	99.9422
551282-4428910-120522-828_A05.CEL	5512824428910120522828	~	2021-01-19 14:52:15.000	A05	0.9783	99.9422
551282-4428910-120522-828_A06.CEL	5512824428910120522828	~	2021-01-19 14:52:49.000	A06	0.9802	99.9422
551282-4428910-120522-828_A07.CEL	5512824428910120522828	~	2021-01-19 14:53:22.000	A07	0.9822	99.9422
551282-4428910-120522-828_A08.CEL	5512824428910120522828	~	2021-01-19 14:53:56.000	A08	0.9783	99.7688
551282-4428910-120522-828_A09.CEL	5512824428910120522828	~	2021-01-19 14:54:29.000	A09	0.9684	99.8459
551282-4428910-120522-828_A10.CEL	5512824428910120522828	~	2021-01-19 14:55:01.000	A10	0.9763	99.8844
551282-4428910-120522-828_A11.CEL	5512824428910120522828	~	2021-01-19 14:55:34.000	A11	0.9743	99.8073
551282-4428910-120522-828_A12.CEL	5512824428910120522828	~	2021-01-19 14:56:07.000	A12	0.9822	99.9807
551282-4428910-120522-828_A13.CEL	5512824428910120522828	~	2021-01-19 14:56:41.000	A13	0.9546	99.7110
551282-4428910-120522-828_A14.CEL	5512824428910120522828	~	2021-01-19 14:57:13.000	A14	0.9822	100
551393 44399340 139533 639 A15 CE	5540004400010100500000		0001 01 10 14 57 45 000	***	0.0004	00.0000

Table 2 Sample Metrics table \*

Column	Description
Sample Filename	Name of CEL file.
Pass/File	<ul> <li>Sample quality control status.</li> <li>The Sample QC step deems it meets the minimum thresholds for DQC, QC call_rate, and average call rate for passing samples.</li> <li>The sample did not pass Sample QC.</li> </ul>
Plate Barcode	Barcode of the array plate.
Scan Date	Date the array was scanned (year-month-day).
Well Position	Location of sample in the array plate e.g A1, B1, etc.
DQC	Dish QC of sample. <b>Note:</b> Dish QC measures the amount of overlap between two homozygous peaks created by non-polymorphic probes. A DQC of 1 equals no overlap (Good), while a DQC of 0 denotes a complete overlap (Bad).
QC Call Rate (QCCR)	QC Call Rate of a sample. <b>Note:</b> The QC call rate is computed over a representative set (usually 20k) of well-behaved probesets. A Low QC call rate over well-behaved probesets suggest sample quality issues.
Call Rate	Call rate of a sample. <b>Note:</b> The Call rate is the percentage of samples assigned a genotype (other than a No Call).
Computed Sex	Computed sex of the sample.
Dual Workflow *	Records the samples that went through the Dual Workflow analysis. <b>Note:</b> A [●] appears in the Dual Workflow column to indicate the sample met the Dual Workflow criteria and was analyzed. * Best Practices with Dual workflow only.

\* The columns that appear in the Sample Metrics table are Workflow specific.

### **Sample Metrics Table Settings**

- 1. Click on the Sample Metrics Sicon.
- 2. At the pop up window (Figure 15), click on the toggles to show or hide the columns listed. **Note:** Columns Visibility preferences must be set for each workflow and **Sample Filename** cannot be toggled off.

Figure 15 Sample Metrics Settings window	s Table
Sample Metrics Table Setting	s
Columns Visibility	
Sample Filename Plate Barcode Pass/Fail Scan Date Well Position DQC QC Call Rate Call Rate Call Rate Call Rate Computed Sex MAPD WavinessSD CN passes MAPD CN passes MAPD CN passes QC Control Chosen Control Used Number of controls for CN tuning CN tuned using controls	11111111111111111111111

3. Click outside the pop up window to close it and save your toggled selections.

### Filtering the Sample Metrics Table

1. Enter a word or value in the Search field (upper right).

If a match is found, only your search results are displayed. To clear (return the table to its original populated state), highlight and delete your search entry.

### Sorting the Sample Metrics Table

- Click on a column header to sort it either in ascending or descending order.
   A small arrow appears on the header indicting the sorting order.
- 2. Click the header again to change the sorting order.

Ľ

button.

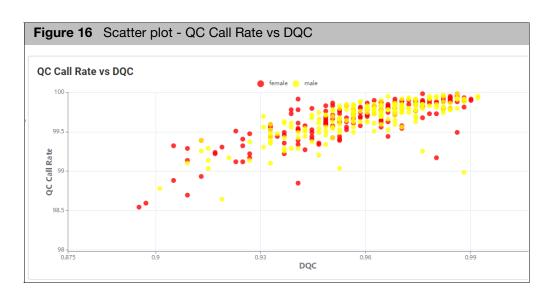
### Copying the table

1. Click to select the row(s) you want to copy, then click the

2. Open Word or an Excel file.

3. Press Ctrl v to paste the table data.

**Scatter plot** A **QC Call Rate** vs **DQC** Scatter plot appears below the Sample Metrics table. Plots are color-keyed by gender, as shown in Figure 16.



## ProbeSet Summary table

The ProbeSet Summary table's (Figure 17) columns and descriptions are detailed in Table 3.

ProbeSet S	Summary Tab	le 🎯			Search	h	Q 🗘
	,					2	0768 of Total 207
Probeset ID 🏦	Affy SNP ID ↑↓	Conversion Type ↑↓	Call Rate †↓	BestProbeset †↓	Minor Allele Frequency ↑↓	FLD †↓	H.W. p-Value ↑↓
AFFX-SP-000001	Affx-2643493	PolyHighResolution	100	1	0.4554	20.3033	0.0988
AFFX-SP-000002	Affx-7455925	PolyHighResolution	99.4751	1	0.3799	15.7325	0.3042
AFFX-SP-000003	Affx-3102334	PolyHighResolution	100	1	0.4987	17.5557	0.4421
AFFX-SP-000004	Affx-14063572	PolyHighResolution	99.7375	1	0.4697	16.8367	0.0365
AFFX-SP-000005	Affx-6193137	PolyHighResolution	99.2126	1	0.4987	13.6868	0.9999
AFFX-SP-000006	Affx-11800709	PolyHighResolution	100	1	0.4869	15.6303	0.8884
AFFX-SP-000007	Affx-13282062	PolyHighResolution	99.4751	1	0.4789	15.7838	0.5260
AFFX-SP-000008	Affx-21222369	PolyHighResolution	99.2126	1	0.4960	15.0759	0.6816
AFFX-SP-000009	Affx-30124231	PolyHighResolution	100	1	0.4357	13.3447	0.0183
AFFX-SP-000010	Affx-17140745	PolyHighResolution	99.2126	1	0.4974	12.2666	0.1813
AFFX-SP-000011	Affx-28077386	PolyHighResolution	100	1	0.4357	16.5186	0.0707
AFFX-SP-000012	Affx-13205808	PolyHighResolution	99.4751	1	0.3958	18.4828	0.0017
ACEV CD 000010	Aff. 01071455	DeluHighPassalution	00.4751	1	0.4420	10 6110	0.0204

 Click on the adjacent Gear icon to display the ProbeSet Summary Table Settings window. (Figure 18) Click on a column's toggle to turn its visibility on or off. Note: ProbeSet ID cannot be toggled off.

Note: Columns Visibility preferences must be set for each workflow.

Figure 18ProbeSet SetTable Settings window	ummary
ProbeSet Summary Table	Settings
Columns Visibility	
Probeset ID	
Affy SNP ID	
Conversion Type	
Call Rate	
BestProbeset	
Minor Allele Frequency	
FLD	
H.W. p-Value	

 Table 3
 ProbeSet Summary Table

Column	Description
ProbeSet ID	The Axiom unique identifier for the set of probes used to detect a particular Single Nucleotide Polymorphism (SNP probe sets only).
Affy SNP ID	The Axiom unique identifier for the set of probes used to detect a particular Single Nucleotide Polymorphism (SNP).
Conversion Type	ProbeSet classification
Call Rate (CR)	The percentage of samples with a genotype call other than "No Call" for the SNP.
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.
Minor Allele Frequency	The allele frequency for the A allele is calculated as: $PA = \frac{(\# AA \ Calls + 0.5 * AB \ Calls)}{Total \# Calls}$
	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 3 ProbeSet Summary Table

Column	Description
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.
H.W. p-Value	<ul> <li>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.</li> <li>There are two statistical tests used for HWE. When AA, AB, and BB counts are all &gt;=10, a Chi-squared test is used. When one or more of the AA, AB, and BB counts are &lt;10, an Exact test is used. An Exact test means that the p-value is calculated exactly and not approximated from a population distribution.</li> </ul>

### Filtering the ProbeSet Summary Table

1. Enter a word or value in the **Search** field (upper right).

If a match is found, only your search results are displayed.

To Clear (return the table to its original state), delete your search entry.

### Sorting the ProbeSet Summary Table

1. Click on a column header to sort it either in ascending or descending order.

A small arrow appears on the header indicting the new sorting order.

2. Click the header again to change the sorting order.

### Copying the table

Note: Up to 15 records displayed in the table can be copied.

- 1. Click the button to copy the table.
- 2. Open Word or an Excel file.
- 3. Press Ctrl v to paste the table data.

# Batch analysis parameters

The batch analysis and extension parameters can be found on the far right pane of the Batches window tab. (Figure 19)

- 1. Click the down arrow button to view the batch's analysis or extension details.
- 2. Use the pane's scroll bar to view all the reported analyses or click the **left arrow** button to view it in a full window.
- 3. Click the **right arrow** button to return to the pane's (far right pane) default view.

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	17	~	-	
- 80				

	<b>gure 19</b> E arameters	Batch analysis	
	Best Practic		
		•	
	Inbred File   Value (optional)	-	
	Analysis File	Axiom_BovMDv3_96orMore_Step1.r 2.apt-genotype-axiom.AxiomGT1.apt 2.xml	
	Prior Model File	Axiom_BovMDv3.r2.generic_prior.txt	
	SNP List File (recommended)	Axiom_BovMDv3.r2.step1.ps	
	DQC ≥	0.82	
	QC call_rate ≥	97	
	Average call rate for passing samples ≥	98.5	
	Percent of passing samples ≥	95	
3	Multiplate Normalization (recommended)	true	
	Analysis File	Axiom_BovMDv3_96orMore_Step2.r 2.apt-genotype-axiom.mm.SnpSpecifi cPriors.AxiomGT1.apt2.xml	
	Prior Model File	Axiom_BovMDv3.r2.AxiomGT1.mode ls	
	Multi-allele Background Prior Model File	Axiom_BovMDv3.r2.generic_prior.m mb.multimodels_background	
	Multi-allele Pairwise Prior Model File	Axiom_BovMDv3.r2.generic_prior.m mp.multimodels_pairwise	
	Multi-allele Prior Model File	Axiom_BovMDv3.r2.generic_prior.m m.multimodels	
	SNP List File (recommended)	Axiom_BovMDv3.r2.step2.ps	
	ps2snp File (recommended)	Axiom_BovMDv3.r2.ps2snp_map.ps	
	species-type	Diploid	
_	or outoff >	70	1

# **Workflow Setup**



The **Workflow Setup** window tab (Figure 20) contains three different customizable workflows and supported extensions for the Best Practices workflows.

- "Sample QC"
- "Best Practices" on page 27
- "Best Practices with Dual Workflow (Agbio Arrays only)" on page 32

**Note:** The *BloodGenomiX Reporter* and *BloodGenomiX Reporter XY* workflows are enabled for Blood Transfusion arrays. For more information, see Appendix A.

## Sample QC

1. Click the Workflow Setup tab. (Figure 20)



#### The Workflow Setup page appears. (Figure 21)

appliedbiosystems	Automated Axiom	Analysis
tches Workflow Setup M	ultiplate	
xiom_BovMDv3.r2 $\times \vee$	✓ Sample QC × ∨	Select type ~
	Analysis	Extension
	Default 🔹 🕀 Create 🖓 Reset to Default	to Octate to show
	General Analysis Sample QC	
	Analysis File Axiom_BovMDv3_96orMore_Step1.r2.apt-genotype-axi	Note: This Extension pane is not available with Sample QC workflows.
	Prior Model File	
	Axiom_BovMDv3.r2.generic_prior.txt  SNP List File (recommended)	
	Axiom_BovMDv3.r2.step1.ps X V	
	DQC ≥ 0.82	
	QC calL_rate ≥	
	97	



## Customizing an analysis

**Selecting an array** 1. Click the **Array Type** drop-down (Figure 22) to select the array you want to use. Use the search field to locate a different array.

Figure 22 Array	Туре
Axiom_BovMDv3.r2	× ~
	Q
Axiom_BovMDv3.r2	î
Axiom_PMDRAv2.r3	
Axiom_PMRA.r3	-

Note: The Workflow drop-down displays Sample QC by default. (Figure 23)

Figure 23	Jure 23 Workflow drop-down		
✓ Sample QC	>	<	

# Using the Analysis pane

The Analysis pane features a **Sample QC** tab and a **General Analysis** tab that features **Inbred File** options.

**Note:** Make sure the **Default** toggle is on (Figure 24) if you want to keep the factory default settings and you are not going to modify them.

Figure 24 Analysis options	
General Analysis Sample QC	
Analysis File	Î
Axiom_BovMDv3_96orMore_Step1.r2.apt-genotype-axi	
Prior Model File	
Axiom_BovMDv3.r2.generic_prior.txt ~	
SNP List File (recommended)	
Axiom_BovMDv3.r2.step1.ps × ~	
DQC ≥	
0.82	
QC call_rate ≥	
97 💿	~

- Analysis File: Displays the array you selected.
- Prior Model File: Displays the default Prior Model File associated with your selected array. Click the drop-down to select a different file or add a new file.
- SNP List File (recommended): Displays the default *SNP List File* associated with your selected array. Click the drop-down to select a different file or add a new file.
- **DQC** ≥: Displays the default *DQC* value associated with your selected array. To enter a new value, click inside the text box.
- QC call\_rate ≥: Scroll down to display. Displays the default QC\_call\_rate value associated with your selected array. To enter a new value, click inside the text box.
- Average call rate for passing samples ≥: Scroll down to display. Displays the default Average call rate for passing samples value associated with your selected array. To enter a new value, click inside the text box.
- Percent of passing samples ≥: Scroll down to display. Displays the Percent of passing samples value associated with your selected array. To enter a new value, click inside the text box.
- To add a file to one of the above entries, see "Adding a file not listed" on page 36.
- To change a value, see "Changing a value" on page 36.

### General Analysis options (Optional)

#### Inbred File value for all samples

1. Click the check box, then enter a value. (Figure 25)

Figure	<b>25</b> Ge	neral Analysis pa	ane - Inbred File
Default	-	🕒 Save	C Reset to Saved
Genera	l Analysis	Sample QC	
	ile   Value (opt value for all sa		
0		Default: -, Min: 0, Max:	16
		$\otimes$	

To add an Inbred file not listed in the drop-down (Figure 26), click **Add File**, then see "Adding a file not listed" on page 36 for detailed instructions.

Figure 26 General Analysis options
Analysis
Default 🕖 🕀 Create 📿 Reset to Defaults
General Analysis Sample QC
Inbred File   Value (optional)
analysis_files/5511664410139010422903.txt $\times \vee$

### Saving your custom configuration

1. Click **Create**. (If modifying an existing custom workflow, click **Save**). After clicking **Create**, a message window appears. (Figure 27)

Figure 27 Message window				
Create new custom configuration of the create a custom workflow, e		eate Custom		
Enter Name Name is required!				
Name is required:		Cancel		

2. Enter a workflow name, then click Create Custom.

Your new value is displayed.

### Saving an existing custom configuration

Once you modify an existing custom configuration, the **Save** button is enabled.

1. Click Save.

Your custom configuration is now saved.

## **Best Practices**

1. From the **Workflow Setup** tab, click the **Workflow** drop-down, then select **Best Practices**. (Figure 28)

atches Workflow Setup Multiplate	
xiom_BovMDv3.r2 X V	AxLE Export × ~
Analysis	Extension
Default	Create Z Reset to Defaults     Default     Default     Default
General Analysis Sample QC Genotyping	Input Output
Analysis File	^ Sample Name Mapping and Filter File
Axiom_BovMDv3_96orMore_Step1.r2.apt-genotype	Select a file v
Prior Model File	SNP List Filter
Axiom_BovMDv3.r2.generic_prior.txt	Select a file v
SNP List File (recommended)	SNP Mapping File
Axiom_BovMDv3.r2.step1.ps	Select a file v
DQC ≥	SNP Allele Swap
0.82	Select a file v
QC call_rate ≥	
97	
Average call rate for passing samples ≥	
98.5	

## Customizing an analysis

**Selecting an array** 1. Click the **Array Type** drop-down (Figure 22) to select the array you want to use. Use the search field to locate a different array.



Figure 29 Array	Туре
Axiom_BovMDv3.r2	× ~
	Q
Axiom_BovMDv3.r2	Î
Axiom_PMDRAv2.r3	
Axiom_PMRA.r3	-

2. Click the Workflow drop-down to select Best Practices.

# Using the Analysis pane

The Analysis pane features **Sample QC**, **Genotyping**, and **CN QC** tabs. The **General Analysis** tab features **Inbred File** options.

**Note:** Make sure the **Default** toggle is on (Figure 30) if you want to keep the factory default settings and you are not going to modify them.

Jure 30 Analysis opt	ions	
Analysis		
Default		C Reset to Defaults
General Analysis Sample QC	Genotyping	CN QC
Analysis File		ĺ
Axiom_BloodGenomiX_96orMore_	_Step1.r1.apt-ger	noty
Prior Model File		
Axiom_BloodGenomiX.r1.generic_	prior.txt	~
SNP List File (recommended)		
Axiom_BloodGenomiX.r1.step1.ps	3	× ~
DQC ≥		
0.88		
QC call_rate ≥		
98	0	
	0	

### Sample QC options

- Analysis File: Displays the array you selected.
- Prior Model File: Displays the default Prior Model File associated with your selected array. Click the drop-down to select a different file or add a new file.



- SNP List File (recommended): Displays the default *SNP List File* associated with your selected array. Click the drop-down to select a different file or add a new file.
- **DQC** ≥: Displays the default *DQC* value associated with your selected array. To enter a new value, click inside the text box.
- QC call\_rate ≥: Scroll down to display. Displays the default QC\_call\_rate value associated with your selected array. To enter a new value, click inside the text box.
- Average call rate for passing samples ≥: Scroll down to display. Displays the default Average call rate for passing samples value associated with your selected array. To enter a new value, click inside the text box.
- Percent of passing samples ≥: Scroll down to display. Displays the Percent of passing samples value associated with your selected array. To enter a new value, click inside the text box.
- To add a file to one of the above entries, see "Adding a file not listed" on page 36.
- To change a value, see "Changing a value" on page 36.

### **Genotyping options**

- 1. Click on the **Genotyping** tab to display the options listed below:
- Multiplate Normalization (recommended) becomes enabled for batches using more than one plate.
- **CN Reference Model File**: Contains reference information for CN analysis step.
- CN Priors File: Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
- Analysis File: This field displays the array you selected in the Array Type dropdown.
- Prior Model File: This field displays the default Prior Model File associated with your selected array. Click the drop-down to select a different file or add a new file.
- SNP List File (recommended): This field displays the default SNP List File associated with your selected array. Click the drop-down to select a different file or add a new file.
- **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.
- 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.
- species-type: Species type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.
- **cr-cutoff**: Minimum acceptable call rate.
- fld-cutoff: For autosomal probesets, minimum acceptable FLD value for cluster separation.

- het-so-cutoff: Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.
- hom-ro1-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.
- hom-ro2-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.
- 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.
- recommended: ProbeSets having ConversionTypes in this recommended set will be included in the recommended probeset list.

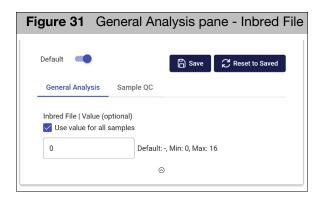
### **CN QC options**

- 1. Click on the CN QC tab to display the options listed below:
- 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.
- 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.

### **General Analysis options (Optional)**

### Inbred File value for all samples

1. Click the check box, then enter a value. (Figure 31)



To add an Inbred file not listed in the drop-down (Figure 32), click **Add File**, then see "Adding a file not listed" on page 36 for detailed instructions.

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			iC.	24	
		P		в	10
		æ	2	-	

Figure 32 General Analysis opt	ions
Analysis	
Default 🕖 Create	C Reset to Defaults
General Analysis Sample QC	
Inbred File   Value (optional)	
analysis_files/5511664410139010422903.txt	× ~
0	

### Saving your custom configuration

 Click Create. (If modifying an existing custom workflow, click Save). After clicking Create, a message window appears. (Figure 33)

Figure 33 Message window				
Create new custom configura	tion?			
To create a custom workflow, er	nter name and press Cre	eate Custom		
Enter Name Name is required!				
		Cancel		

2. Enter a workflow name, then click Create Custom.

Your new value is displayed.

### Saving an existing custom configuration

Once you modify an existing custom configuration, the Save button is enabled.

1. Click Save.

Your custom configuration is now saved.

## **Extension pane**

The **Extension** pane (right) manages additional features and settings available for export or analysis within the Best Practices Workflow.

Extensions are enabled when available, as they vary depending on the array design. **Note:** Not every extension is available for every array design.

To use the Extension pane, see "Using the extension pane" on page 61.

# Best Practices with Dual Workflow (Agbio Arrays only)

For selected arrays, Best Practices with Dual Workflow is an additional analysis option. For samples that pass DQC and do not make the QCCR, Automated Axiom will re-analyze those samples within the same batch. For more information, see the *Axiom Genotyping Data Solutions Guide* (MAN0018363).

1. From the **Workflow Setup** tab, click the **Workflow** drop-down, then select **Best Practices with Dual Workflow**. (Figure 34)

Figure 34 Workflow Setup window tab - Populated example					
命 appliedbiosystems	Automated Axiom Analysis	5			
Batches Workflow Setup Multiplate					
Axiom_BovMDv3.r2 × ~	$\checkmark$ Best Practices with Dual Workflow $\times$ $\vee$	✓ VCF × ✓			
	Default  Create  Create  Create	Extension Default			
	Central Analysis       Sample QC       Genotyping         Analysis File	Input Output ProbeSet List Filter SNPolisher/Recommended ps  Standard Chromosomes Annotation File Axiom_BovMDv3.na35.r2.a1.annot.db  SNP Identifier probeset_id			

## Customizing an analysis

- Selecting an array
- 1. Click the **Array Type** drop-down (Figure 35) to select the array you want to use. Use the search field to locate a different array.

Figure 35 Array Type
Axiom_BovMDv3.r2 × ~
Q
Axiom_BovMDv3.r2
Axiom_PMDRAv2.r3

2. Click the Workflow drop-down to select **Best Practices with Dual Workflow.** 

## Using the Analysis pane

The Analysis pane features **Sample QC**, **Genotyping**, and a **General Analysis** tab features **Inbred File** options.

**Note:** Make sure the **Default** toggle is on (Figure 36) if you want to keep the factory default settings and you are not going to modify them.

Figure 36 Analysis options				
Default	+ Create	Reset to Defaults		
General Analysis Sample QC	Genotyping			
Analysis File		Î		
Axiom_BovMDv3_96orMore_Step1	I.r2.apt-genotype	e-axi		
Prior Model File				
Axiom_BovMDv3.r2.generic_prior.1	txt	~		
SNP List File (recommended)				
Axiom_BovMDv3.r2.step1.ps		× ~		
DQC ≥				
0.82				
QC call_rate ≥				
97	-	~		
	$\odot$			

### Sample QC options

- Analysis File: Displays the array you selected.
- Prior Model File: Displays the default Prior Model File associated with your selected array. Click the drop-down to select a different file or add a new file.
- SNP List File (recommended): Displays the default *SNP List File* associated with your selected array. Click the drop-down to select a different file or add a new file.
- **DQC** ≥: Displays the default *DQC* value associated with your selected array. To enter a new value, click inside the text box.
- **QC call\_rate** ≥: Scroll down to display. Displays the default QC\_call\_rate value associated with your selected array. To enter a new value, click inside the text box.
- Average call rate for passing samples ≥: Scroll down to display. Displays the default Average call rate for passing samples value associated with your selected array. To enter a new value, click inside the text box.
- Percent of passing samples ≥: Scroll down to display. Displays the Percent of passing samples value associated with your selected array. To enter a new value, click inside the text box.
- To add a file to one of the above entries, see "Adding a file not listed" on page 36.
- To change a value, see "Changing a value" on page 36.

### **Genotyping options**

- 1. Click on the **Genotyping** tab to display the options listed below:
- Multiplate Normalization (recommended) becomes enabled for batches using more than one plate.
- **CN Reference Model File**: Contains reference information for CN analysis step.
- **CN Priors File**: Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
- Analysis File: This field displays the array you selected in the Array Type dropdown.
- Prior Model File: This field displays the default Prior Model File associated with your selected array. Click the drop-down to select a different file or add a new file.
- SNP List File (recommended): This field displays the default SNP List File associated with your selected array. Click the drop-down to select a different file or add a new file.
- **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.
- 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.
- species-type: Species type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.
- **cr-cutoff**: Minimum acceptable call rate.
- fld-cutoff: For autosomal probesets, minimum acceptable FLD value for cluster separation.
- het-so-cutoff: Minimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.
- hom-ro1-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.
- hom-ro2-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.
- 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.
- recommended: ProbeSets having ConversionTypes in this recommended set will be included in the recommended probeset list.

### General Analysis options (Optional)

#### Inbred File value for all samples

1. Click the check box, then enter a value. (Figure 37)

gure 37	7 Gen	eral An	alysis pa	ne - Inbred File
Default 🗨	•		🛱 Save	C Reset to Saved
General An	alysis S	Sample QC		
		,		
0		Default:	-, Min: 0, Max: 1	6
		6	)	
	Default General An Inbred File   Use valu	Default  General Analysis  Inbred File   Value (optic Use value for all sam	Default General Analysis Sample QC Inbred File   Value (optional) Use value for all samples 0 Default:	General Analysis Sample QC Inbred File   Value (optional) Use value for all samples

To add an Inbred file not listed in the drop-down (Figure 38), click **Add File**, then see "Adding a file not listed" on page 36 for detailed instructions.

Figure 38 General Analysis options
Analysis
Default 🕖 🕀 Create 📿 Reset to Defaults
General Analysis Sample QC
Inbred File   Value (optional)
analysis_files/5511664410139010422903.txt $\times$ $\vee$
0

### Saving your custom configuration

 Click Create. (If modifying an existing custom workflow, click Save). After clicking Create, a message window appears. (Figure 39)

Figure 39 Message window				
Create new custom config	uration?			
To create a custom workflow Enter Name Name is required!	v, enter name and press Cr	eate Custom		
Hame lo required.		Cancel		

2. Enter a workflow name, then click Create Custom.

Your new value is displayed.

### Saving an existing custom configuration

Once you modify an existing custom configuration, the **Save** button is enabled.

- 1. Click Save.
  - Your custom configuration is now saved.

## **Extension pane**

The Extension pane (right) manages additional features and settings available for export or analysis within the Best Practices with Dual Workflow.

Extensions are enabled when available, as types vary depending on the array design. **Note:** Not every extension is available for every array design.

To use the Extension pane, see "Using the extension pane" on page 61.

## Adding a file not listed

- To select a file not listed on the drop-down menu, click Add File. An Explorer window appears.
- 2. Select the file you want, then click Open.

If you are modifying an existing custom configuration the **Save** button is now enabled. If you are modifying a factory default workflow the **Create** button is enabled.

## Changing a value

### New value

- 1. To change a default value, click inside its text box.
- 2. Enter a new value.

If you are modifying an existing custom configuration the **Save** button is now enabled. If you are modifying a factory default workflow the **Create** button is enabled.

# **Multiplate**



The Multiplate function can be used to manually analyze batches made of 1 to 50 plates. Batches can be set up to include any extensions available for the selected array type.

1. Click the Multiplate tab. (Figure 40)

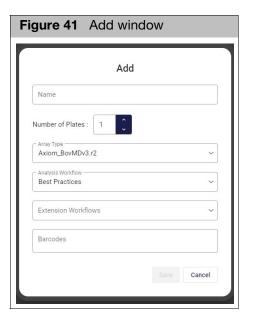
Figure 40	Workflow Setup tab	
Batches Wo	rkflow Setup Multiplate	

The Multiplate window appears.

## Using the Multiplate window tab

## Adding a new multiplate

 Click the Add button (upper left). The Add window appears. (Figure 41)



2. In the Name field, enter the name of your batch.



- 3. **Number of Plates:** Click the up and down arrows to select the number of plates (1-50). Alternatively, type in the number of plates.
- 4. **Array Type:** Click the drop-down to select the array type. The Array Type defaults to the first alphabetical array in the library.
- 5. Analysis Workflow: Click the drop-down to select an analysis workflow.
- 6. **Extension Workflows:** (Optional and if available) Click the drop-down to display the available extensions (Figure 42), then click on the extension's check box.

Figure 42 Extension Workflows	
Extension Workflows	~
Allele Translation	<b>^</b>
AxLE Export	
Axiom HLA	
UCF	-

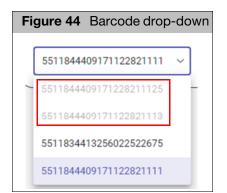
7. Barcodes: Use this field to paste in the associated barcode(s), then click Save.

If all plates have been uploaded, the analysis starts immediately. If there are any plates within your multiplate that are still in process on the GeneTitan, a multiplate card appears. (Figure 43)

Figure 43 New multiplate card
PFocus 3
Process Now Available: 2 / 4 Last updated: 2023-12-20 13:51:58.842
5511834413256022522675 V Edit

## Running a multiplate

8. Click the Barcode field's drop-down to view the associated barcodes. **Note:** Grayed out barcodes (Figure 44) are still being uploaded to the server.



9. If you want to run plates (not grayed out), click the Multiplate plate's **Process Now** button.

A Confirmation pop up window appears.

- 10. Click **OK** to start processing the available plates.
- 1. Click the **Edit** button.

The Edit window appears. (Figure 45)

gure 45 Edit	window
E	dit
Name PFocus 3	
Number of Plates : 4	
Array Type PharmacoFocus.r5	~
Analysis Workflow	~
Extension Workflows	~
Barcodes	5511844409171122821113 ()
	5511844409171122821111 ③
	Save Cancel

Use the Edit window to change:

- Number of plates
- Analysis Workflow
- Extension Workflow
- Barcodes: To add a plate, paste a new barcode. To remove a barcode, click on its X.
- Click Cancel to cancel your edits.
- Click **Save** to save your changes.

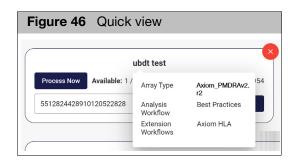
multiplate group

Editing a

Z

## Multiplate quick view

1. Mouse over a plate name to view its information. (Figure 46)



2. Click the X button to remove a plate card from the Multiplate window tab.

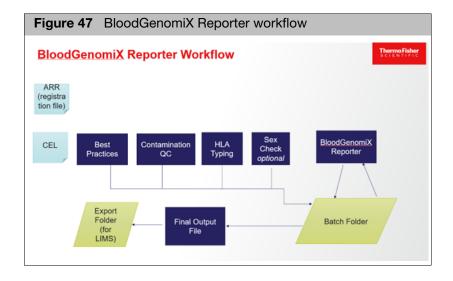




**IMPORTANT!** You must pre-purchase reports before using the BloodGenomiX Reporter feature. See "Purchasing reports" on page 51.

### Overview and workflow

The BloodGenomiX Reporter workflow consists of the following analyses and quality controls steps:



- Best Practices
- ContaminationQC
- Axiom HLA
- Sex Check (Optional)
- Blood typing for research

Best Practices is the standard Axiom Best Practices workflow in which only the samples that pass the preset QC thresholds are genotyped and progressed further in the workflow. Best Practices includes copy number aware genotyping. For more information, see the Axiom Genotyping Data Solutions Guide (P/N MAN0018363).

The next step in the workflow is ContaminationQC. Contamination is estimated by the slope of a linear regression of intensity ratio of A or B allele for homozygous BB or AA calls for a given sample versus the plate frequency of the A or B allele. Based on predefined threshold values, samples are given a Yes or No result for contamination.



Samples that pass Best Practices and ContaminationQC are then Axiom HLA and blood typed. The Axiom HLA output is limited to the Class I loci (A, B, C). Extended blood phenotyping is based on the array and library file package.

If the BloodGenomiX Reporter XY workflow is selected, computed sex is compared to the supplied sex, then either a PASS or FAIL will be reported. To generate a report *without* Sex Check, select **BloodGenomiX Reporter**.

For more information about other available Outputs and their settings, see Chapter 3.

### Setting up a BloodGenomiX analysis

The Analysis window contains a row of tabs. (Figure 48) Each tab is an analysis type. Click on an analysis tab to select it and view its available fields and values.

Figure 48 Analysis window Reporter XY (includes Sex ch				ιiX
loodGenomiX Reporter XY × ~				
Analysis				
Default 💭				
Sample QC Genotyping CN QC Contan	inationQC	HLA	XY	BGX
	~ × ~			
DQC ≥ 0.82				
QC call_rate ≥ 97				
Average call rate for passing samples ≥ 98.5				

**Note:** Once a factory default workflow parameter is changed, a **Create** button appears. If changing a parameter within an existing custom workflow, a **Save** button appears.

A

After clicking Create, a message window appears. (Figure 49)

Figure 49 Mes	sage window	
Create new custom configu	uration?	
To create a custom workflow Enter Name Name is required!	, enter name and press Cr -	eate Custom
	Q Create Custom	Cancel

 Enter a workflow name, then click Create Custom. Your new value is displayed.

Sample QC (fields and descriptions)

BloodGenomiX Analysis window tabs

Analysis File	Displays the selected array.
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 (recommended)	A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.
DQC	A sample's dish QC value must be greater than or equal to DQC to pass 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.
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.
Percent of passing samples	Displays the Percent of passing samples value associated with your selected array. <b>Note:</b> If a plate's percent of passing samples is smaller than this number, all samples on the plate will display a warning in the Sample Table.

#### Genotyping (fields and descriptions)

Normalizationmultiple plate genotyping analysis when individual plates are expected to have similar allele frequencies, i.e. a similar mix of genetic ancestry with no (or the same) enrichment for specific blood types.Analysis FileDisplays the selected array.Prior Model FileDefines 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 (recommended)A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.CN Reference Model FileSpecifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.ps2snp FileIf 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.Genotype Frequency FileIf 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.species-typeSpecies type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid. cr-cutoff		
Prior Model FileDefines 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 genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.SNP List File (recommended)A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.CN Reference Model FileContains reference information for CN analysis step.File CN Priors FileSpecifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.ps2snp FileIf 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.Genotype Frequency FileIf 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.species-typeSpecies type for the array, which affects some SNP QC checks. Species type include: Diploid, Human, and Polyploid.cr-cutoffMinimum acceptable call rate.fid-cutoffFor autosomal probesets, minimum acceptable FLD value for cluster separation.het-so-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous cluster. <tr< th=""><th>Multiplate Normalization</th><th>multiple plate genotyping analysis when individual plates are expected to have similar allele frequencies, i.e. a similar mix of genetic ancestry with no (or the same) enrichment for specific blood</th></tr<>	Multiplate Normalization	multiple plate genotyping analysis when individual plates are expected to have similar allele frequencies, i.e. a similar mix of genetic ancestry with no (or the same) enrichment for specific blood
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 (recommended)A file of probeset IDs to genotype. For Sample QC it defines the probesets used to calculate QC Call Rate.CN Reference Model FileContains reference information for CN analysis step.SP2 snp FileIf multiple probeset los do genotype defining each state.ps2snp FileIf multiple probeset designs exist on the array for a given SNP (for example, one forward and one reverse strand design), then the 	Analysis File	Displays the selected array.
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FileCN Priors FileSpecifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.ps2snp FileIf 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.Genotype FrequencyIf 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.species-typeSpecies type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.cr-cutoffMinimum acceptable call rate.fld-cutoffFor autosomal probesets, minimum acceptable FLD value for cluster separation.het-so-cutoffMinimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.hom-ro1-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 1 genotype clusters.hom-ro3-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.hom-ro-3-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.hom-ro-3-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.	SNP List File (recommended)	
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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.Genotype Frequency FileIf 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.species-typeSpecies type for the array, which affects some SNP QC checks. Species types include: Diploid, Human, and Polyploid.cr-cutoffMinimum acceptable call rate.fld-cutoffFor autosomal probesets, minimum acceptable FLD value for cluster separation.het-so-cutoffMinimum acceptable value for the correctness of the Size (Y position) offset of the heterozygous cluster.hom-ro1-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 1 genotype cluster.hom-ro2-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.hom-ro-3-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.hom-ro-3-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.recommendedProbeSets having ConversionTypes in this recommended set will be	CN Priors File	
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position) of the homozygous clusters (Ratio Offset) when a probeset has 2 genotype clusters.hom-ro-3-cutoffMinimum acceptable value for the correctness of the Contrast (X position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.recommendedProbeSets having ConversionTypes in this recommended set will be	hom-ro1-cutoff	position) of the homozygous clusters (Ratio Offset) when a probeset
position) of the homozygous clusters (Ratio Offset) when a probeset has 3 genotype clusters.recommendedProbeSets having ConversionTypes in this recommended set will be	hom-ro2-cutoff	position) of the homozygous clusters (Ratio Offset) when a probeset
	hom-ro-3-cutoff	position) of the homozygous clusters (Ratio Offset) when a probeset
	recommended	



#### CN QC (fields and descriptions)

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.
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.

#### ContaminationQC (fields and descriptions)

ProbeSet ID	ProbeSet in this list will be used to determination contamination.
Genotype Clusters	Clusters that will be used to generate the linear model used to determine cross contamination.
Threshold p-value	Threshold for adjusted p-value, samples with adjusted p-value greater than threshold will have contaminated set to <b>No</b> in the output.
Threshold estimated contamination	Samples with a value less than threshold will have contaminated set to <b>No</b> in the output.

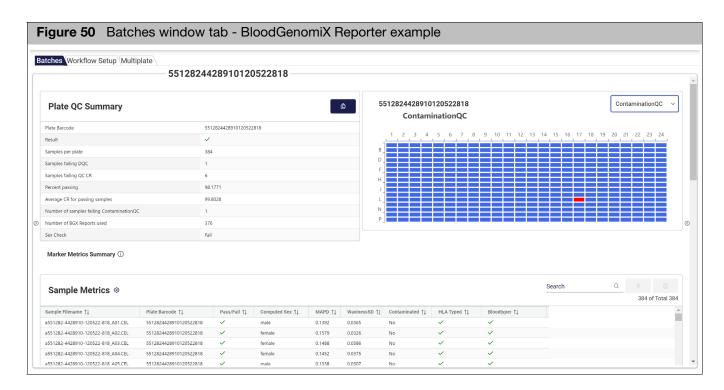
#### HLA (fields and descriptions)

Loci Selection (default All)	Loci A, B, C are options for HLA typing.
Output Folder	Location and name of output folder.
Output Name	Name of the output which includes 2 or 4 digit as well as loci. Example: ( <b>AxiomHLA_2_A_Results</b> )

BGX (fields and descriptions)

Antigen list	List of antigens and applicable alleles that will be typed. A two column text file with the headers: test_group_name and test_name. Test group name is Blood Group System and test name is antigen/ allele name.
Output Folder	Name of the Results folder.

## Viewing a BloodGenomiX batch



The BloodGenomiX Reporter creates two output folders:

- The BGXPrimary folder contains the initial files and folders that are generated from the BloodGenomiX workflow. This output is then merged into the BloodGenomix Report.
- The BGX folder contains the report file in text format. The results from the primary output files along with the QC results from the Best Practices analysis are collated into a BGX Report file.

The BGXPrimary folder contains the following files:

#### apt2-bloodtyper

This text file contains samples and corresponding blood types.

cel_file	Sample file
test_group_name	Defines the Blood group system: HEA (Human Erythrocyte Antigen, HPA (Human Platelet Antigen) or, HNA (Human Neutrophil Antigen).
test_name	Blood type that is dependent on array and antigen group file that was used for analysis.
test_description	Antigen or Allele

### BGXPrimary folder contents

result_value	Positive: Selected test_name is detected in the sample,
	Negative: Selected test_name is not detected in the sample,
	NT: Not Typed, selected test_name antigen was not analyzed
	Not_present: Selected test_name allele is not detected in the sample.
	<b>HEM</b> : Test_name allele is present as hemizygous
	HET: Test_name allele is present as heterozygous
	HOM: Test_name allele is present as homozygous
	PARTIAL: Phenotype is partially expressed
	WEAK: Reduced phenotype expression
result_status	<b>PASS</b> : Sample is found to be either positive or negative for th corresponding test_name.
	FAIL: Sample was not typed, no result was found

AxiomBT.bloodtyper\_summary

This text file contains the frequency of antigen or alleles in the batch.

Test type	Defines test; antigen or allele
Antigen or Allele Name	Test_name
Pos/Hom Samples	Number of samples that are positive and homozygous for antigen or allele.
Neg/Not Present Samples	Number of samples that are negative and not present for antigen or allele.
Weak/Het Samples	Number of samples that are weak and heterozygous for antigen or allele.
Partial/Hem Samples	Number of samples that are partial and hemizygous for antigen or allele.
NT Samples	Number of samples that are not typed for antigen.
Percent Pass	Percentage of samples that pass for given antigen or allele.

#### AxiomBT.marker\_calls

This text file contains the test\_name and the associated genotype or copy number call in cname format.

cel_file	Sample file
test_group_name	Blood group system: HEA, HPA or HNA, and antigen or allele type.
test_name	HGVS cname
result_value	Genotype calls in forward strand format or copy number call.
result_status	PASS or FAIL

blood-sample-list

This single column text file contains the samples that were typed.



#### collected-attributes

This text file contains the collated attributes of the samples in the batch.

#### BGXPrimary Contamination

subfolders

This folder contains the text file that shows contamination status of the samples in the batch.

cel_file	Sample file
alpha	Estimated contamination threshold, proportional to the level of contamination, value is expected to be centered on 0.0, default: 0.02.
p-value	The p-value of the linear regression fit. Null hypothesis is that the slope (alpha) is zero. The default p-value threshold is 0.005.
contaminated	Yes or No, depending on the alpha and p-value of the sample.

#### HLA

The analysis sub-folder within this HLA folder contains the intermediary and results files for the A, B, and C loci.

**BGX folder contents** The BGX folder contains the Report file that is in a text (TXT) format. The results from the primary output files along with the QC results from the Best Practices analysis are collated into the BGX Report. The Report file displays each sample result in a single row.

Each BGX Report displays these same default column headers:

sample_identifier	Sample name; derived from the ARR file
	<b>Note:</b> For more information on ARR, see the GeneChip Command Console (GCC) User Guide (P/N MAN0027771).
cel_file	.cel file
array_barcode	Array barcode
run_identifier	Automated Axiom batch ID
method	Array name
result_date	ISO8601 date time stamp of the analysis

The remaining columns: **software**, **test\_group\_name**, **test\_name**, **test\_description**, **result\_value**, **result\_unit**, and **result\_status** are specific for each primary BGX report file. These columns are detailed in the following filename tables:

#### Collected-attribute

The BloodGenomiX Reporter requires a customized batch registration file. To obtain one or for more information, contact your local field support person.



software	test_group_name	test_name	test_description	result_value	result_unit	result_status
		Well location	,	well	string	PASS/FAIL
		Source plate barcode		barcode	string	PASS/FAIL
		Source plate well location	Sample Attributes	well	string	PASS/FAIL
		sample type		source of DNA	string	PASS/FAIL
ARR	Attributes	Customer supplied sex		Female or Male	string	PASS/FAIL
		Date received		date	datetime	PASS/FAIL
		Reagent Lot		Lot number	string	PASS/FAIL
		Project name		Name	string	PASS/FAIL

#### Output from the AxiomGT1.contamination

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
		p-value		Example: 0.476659	integer	PASS/FAIL
apt2- contamination- qc version #	ContaminationQC	Estimated contamination	ContaminationQC	Example: 0.00242784	integer	PASS/FAIL
		Contaminated	]	No	string	PASS/FAIL



#### Output from HLA

The HLA section of the BGX Report contains the output from Loci A, B and C. X and Y are designations for allele 1 and allele 2.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
	apt2-HLA HLA Locus Genotype	HLA-locus-x	type	HLA-four- field	four digit	PASS/FAIL
apt2-HLA		HLA-locus-y	type	HLA-four- field	four digit	PASS/FAIL
		HLA-locus-p1	probability	probability to 2 decimals	unit interval	PASS/FAIL
		HLA-locus-p2	probability	probability to 2 decimals	unit interval	PASS/FAIL

#### **QC** Metrics

The sample QC metrics are ported from the Best Practices analysis.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
		DQC		example: 0.98617	float	PASS
AutoAx 2.0.0	AutoAx 2.0.0 QC	QC Call Rate		example: 99.84586	float	PASS
AutoAx 2.0.0		40	MAPD		example: 0.136106	float
		WavinessSD	]	example: 0.036856	float	PASS/FAIL

#### Antigen Genotype

From the **AxiomBT.marker\_calls** file, the BGX Report collates the batch samples' genotype or copy number calls per test name in HGVS (cname) nomenclature.

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
AutoAx 2.0.0	Blood group name	HGVS cname or CN Region	0 11	Forward strand or Copy Number Call	genotype	PASS/FAIL

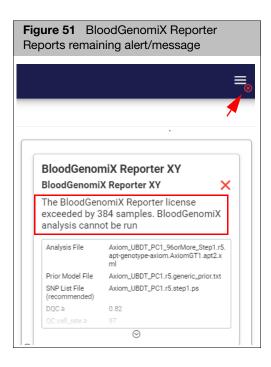


#### Extended Blood Typing

software	test_group_name	test_name	test_description	result_value	result_unit	result_status
apt2-bloodtyper, version #	Blood group name	Blood Type	Antigen or Allele	POS NEG	antigen type	PASS/FAIL
				NT NOT_PRESENT		
				HEM HET		
				HOM		
				PARTIAL WEAK		

### Purchasing reports

If you see this alert (Figure 51), you will need to purchase reports to unlock and use AutoAx's BloodGenomiX reporting feature.



- 1. Contact your Thermo Fisher Sales Representative for a BloodGenomiX Reporter quote.
- 2. Click on  $\blacksquare$  (far right), then click **Configurations**.

The Batch Download Options window appears.

3. Click Licensing.

The BloodGenomiX Reporter window appears. (Figure 52)



Figure 52	BloodGenom	niX Reporter v	vindow
Downloads			
Licensing	BloodGenomiX Rep Reports Remaining	^	
	Thresholds	Warning Critical	50 % 10 %
	Server ID	56544020-0bf1-44fa-993	88-d02345d0fd0c
	License Key		
			Apply

- 4. Click on the Server ID field's Copy to Clipboard button.
- 5. Open a new email, then paste the Server ID into it.
- 6. Add your Purchase Order number to the email, then send it your Thermo Fisher Sales Representative.

Allow 48 hours for your order to process. After your order is processed, you will receive an email containing the report license.

7. Copy the license string from the email, then paste it into the License Key field, as shown in Figure 53.

Figure 53 License Ke		niX Reporter w	indow wit	h
Downloads Licensing	BloodGenomiX Re Reports Remaining Thresholds	0 Warning Critical	50 % 10 %	^
	Server ID License Key	56544020-0bf1-44fa-993 U2FsdGVkX1+kV8bTEI zhuYmo+43b9JepbMLM 6p3rGguINJKhI5ObtJtu luugdb0/2621v+3JJS+ /SnjIBA5vuUNaB4/J8Xs kjbsuCfYgHoq3dGhaC ez4XrEc808DTAuC17Bk eNufRY89PP8ADFY4P0 +Ruk8Q	_gNklt40GZyhxu04 /11vCDl8bqsPsca/Y ldbOK/eTXOUgMHI YIFXw4MmJBWhW alz8Mij34+po/+iNK zB/6Eu3z0zebo5V 2LQ7IPFaUTasBBhf (vyDPJ9oH2CCnKQ	Ym bh V1 Lj J2 Da JJ Vu

8. Click Apply.

#### The **Reports Remaining** count updates.

Customizing thresholds (Optional)

After clicking **Apply**, you can change these two default **Threshold** values:

 Warning: This threshold's default is 50%. Meaning: When 50% of the reports have been used, a yellow triangle icon will appear on the hamburger-shaped icon (top right), as shown below.



Critical: This threshold's default is 10%. Meaning: When 10% of the reports are remaining, a red circle with X icon will appear on the hamburger-shaped icon (top right), as shown below.



To change these default values, click inside each percentage field to enter a different value, then click the **Apply** button. Click on the **Batches** window tab to close the Licensing window.

**IMPORTANT!** The number of reports must be <u>equal to or greater</u> than the number of samples in the batch that are to be processed. If not, BloodGenomiX Reporter will not run.

## Archiving



## GCC Uploader information

The **GCC File Uploader**, uses APIs to upload CEL and ARR files to the server from a GeneTitan MC Workstation.

The GCC Uploader uses APIs to notify the AutoAx server when a plate is complete. A plate complete notification triggers a batch to be created, its workflow steps queued, and when able, it is processed.

## Archiving in AutoAx

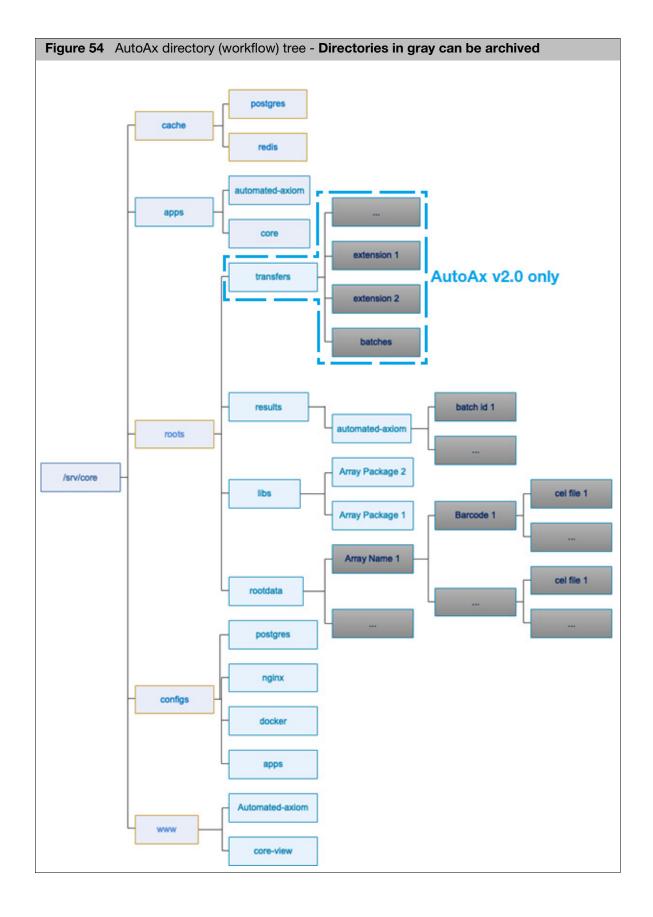
- AutoAx v2.0 users see "Archiving with AutoAx v2.0" on page 56.
- AutoAx v1.2 users see "Archiving with AutoAx v1.2" on page 60.

### AutoAx directory (workflow) tree

Plate data is stored by its array type name, then by sub-directories named using the plate's barcode. This plate data location is at: /srv/core/roots/rootdata

Example: /srv/core/roots/rootdata/Axiom\_BovMDv3/ 5510894388058030721881

- Results are here: /srv/core/roots/results/automated-axiom
- Each batch is stored in a sub-directory by its ID as seen on the AutoAx application's dashboard. Note: Boxes shown in gray (Figure 54) can be archived.



## Archiving with AutoAx v2.0

**IMPORTANT!** All archiving procedures listed below are intended for script writers, command line users, and those with HTTP and Linux experience only.

**Note:** All APIs in AutoAx v2.0 for results (batch information) use **RESTful api** endpoints. To make API endpoint calls, tools such as **curl**, **mget**, or **postman** are required. Specific endpoints are required to be called to help ensure the system stays in sync with any data that may have been archived.

Archiving of **roots/rootdata** does not require synchronization of the system, however *Results* (such as batches), do require the system to be synchronized.

**Retrieving and** sorting batches Before you can view the **batchld**, the batch information must be retrieved from the system. The screen examples that follow (Figure 55) show how to retrieve the batchld based on the ID and the display name, which is at the top of the batch information card.

**Note:** Change or remove **sortby** and/or **sortorder** based on your needs. The barcode ending in **2828** is used in the screen examples that follow.

If searching for the ID, replace 2828 with the value of the ID. The results are a collection of batches that match the filter value in any field of the batch. It is very important to review the results before using the **batchId** command. Doing so will help ensure the batch you want to remove is the correct one.

The following example screens within Figure 55 show how to retrieve the batchld based on the ID and the display name, which is at the top of the batch information card.

	40.00400440/		
GET	✓ 10.80.100.142/api/axiom	n-results-svc/batches?sortby=id&sortorder=	desc&filter=2828&fiel
Params	• Auth Headers (6) Body Pr	e-req. Tests Settings	
Query P	arams		
	Кеу	Value	Description
	sortby	id	
	sortorder	desc	
	filter	2828	
# 🗸	fields	id,batchld,name	
	11 • • • • 1 = 1		
Powersh 1. \$resp svc/batc \$headers 2. \$resp		100.142/api/axiom-results- ter=2828&fields= <u>id,batchId</u> ,name' -Metho	od 'GET' -Headers
1. \$resp svc/batc \$headers 2. \$resp 3.	oonse = Invoke-RestMethod '10.80.: hes?sortby=id&sortorder=desc&fil s		od 'GET' -Headers
Powersh 1. \$resp svc/batc \$headers 2. \$resp 3. Python F 1. impor 2. url = svc/batc 3. paylo 4. heade 5. respo	<pre>conse = Invoke-RestMethod '10.80.: ches?sortby=id&amp;sortorder=desc&amp;fil: conse   ConvertTo-Json Request Method rt requests = "10.80.100.142/api/axiom-result: ches?sortby=id&amp;sortorder=desc&amp;fil: ches?sortby=id&amp;sortby=id&amp;sortorder=desc&amp;fil: ches?sortby=id&amp;sortb</pre>	ter=2828&fields= <u>id,batchId</u> ,name' -Metho s-	od 'GET' -Headers
Powersh 1. \$resp svc/batc \$headers 2. \$resp 3. Python F 1. impor 2. url = svc/batc 3. paylo 4. heade 5. respo 6. print	<pre>conse = Invoke-RestMethod '10.80.: ches?sortby=id&amp;sortorder=desc&amp;file conse   ConvertTo-Json Request Method rt requests = "10.80.100.142/api/axiom-result: ches?sortby=id&amp;sortorder=desc&amp;file conse = {} conse = {} cnse = {} cnse = cequests.request("GET", units c(response.text)</pre>	ter=2828&fields= <u>id,batchId</u> ,name' -Metho s- ter=2828&fields= <u>id,batchId</u> ,name"	od 'GET' -Headers

Removing Batches using API

- Internally, all batches are identified and referenced by the **batchid**, not the ID.
- The **batchId** is a <u>Universal</u> <u>Unique</u> <u>Identifier</u> (UUID)
- If you want to remove a batch from the system without having to ssh, delete, and refresh the cache, you can use the remove api. This is a two-step process.

To know the batchld for any batch, the batch information must be retrieved from the system.

After the batchld you want has been retrieved, use the remove api call using one of the methods shown in Figure 56.

DELI	ETE ~   10	.80.100.142/api/a	axiom-results-svc/batches/:batchlo	l
Param	s 🍨 Auth Head	ders (6) Body	Pre-req. Tests Settings	
Query	Params			
	Кеу		Value	Description
	Кеу		Value	Description
Path V	′ariables			
	Кеу		Value	Description
	batchld		a088b340-3279-4e98-8522	Description
. curl - 522- <u>8c0</u> e	locationreques <u>=19a46486</u> '	t DELETE '10.80.1	100.142/api/axiom-results-svc/batches	:/a088b340-3279-4
curl 3522- <u>8c0e</u> PowerSho \$respo 5522-8c0e	locationreques <u>e19a46486</u> ' ell Method pnse = Invoke-RestM e19a46486' -Method	ethod '10.80.100 'DELETE' -Headers	.142/api/axiom-results-svc/batches/a0	
curl - 522- <u>8c0e</u> 20werSho \$respo 522-8c0e 2. \$respo	locationreques <u>el9a46486</u> ' ell Method pose = Invoke-RestM	ethod '10.80.100 'DELETE' -Headers	.142/api/axiom-results-svc/batches/a0	
curl - 522- <u>8c0e</u> <i>CowerSha</i> \$respond 522-8c0e \$respond	-locationreques <u>el9a46486</u> ' ell Method ponse = Invoke-RestM <u>el9a46486' -Method</u> onse   ConvertTo-Js equest Method	ethod '10.80.100 'DELETE' -Headers	.142/api/axiom-results-svc/batches/a0	
curl 522- <u>8c@</u> 20werSha \$respond 522-8c@ 20ython R mport <u>re</u> url =  payloa	-locationreques <u>el9a46486</u> ' ell Method ponse = Invoke-RestM 19a46486' -Method ponse   ConvertTo-Js equest Method equests "10.80.100.142/api ad = {}	ethod '10.80.100 'DELETE' -Headers on	.142/api/axiom-results-svc/batches/a0	988b340-3279-4e98
curl - 522- <u>8c@</u> 20werSha \$respond \$respond \$respond url = paylog paylog respond	locationreques <u>el9a46486</u> ' ell Method ponse = Invoke-RestM <u>e19a46486' -Method</u> <u>onse   ConvertTo-Js</u> <u>equest Method</u> <u>equests</u> "10.80.100.142/api ad = {} rs = {}	ethod '10.80.100 'DELETE' -Headers on /axiom-results-sv	.142/api/axiom-results-svc/batches/a@ s \$headers	988b340-3279-4e98
8522-8c06 PowerShi . \$respondent \$522-8c00 2. \$respondent Python R mport respondent python R mport respondent . url = 2. 3. paylog 1. header 5.	locationreques <u>el9a46486</u> ' ell Method ponse = Invoke-RestM <u>e19a46486' -Method</u> <u>onse   ConvertTo-Js</u> <u>equest Method</u> <u>equests</u> "10.80.100.142/api ad = {} rs = {}	ethod '10.80.100 'DELETE' -Headers on /axiom-results-sv	.142/api/axiom-results-svc/batches/a0 \$ \$headers /c/batches/a088b340-3279-4e98-8522-8c	)88b340-3279-4e

## Re-syncing all<br/>batchesAfter the selected batches have been archived off the server, they can be re-synced.<br/>Below, examples for each tool will be provided for how to synchronize the system.

There are many tools/options for performing HTTP endpoint calling. The screen examples in Figure 57 show the most common.

The time to synchronize is based on the number of batches and the speed of the server and/or network connection.

Note: In all cases, you must change the currently displayed IP address to your server's IP address.

Figure 57	Different methods to	re-sync batches	
Postman Me	thod		
POST	✓ http://10.80.9	99.104/api/axiom-results-svc/ba	tches/cache/sync
Params	Auth Headers (7) B	ody Pre-req. Tests Settin	gs
Query Pa	ams		
	(ey	Value	Description
	<еу	Value	Description
Curl Methoa			
1. curllo 2.	cationrequest POST '10.	80.99.104/api/axiom-results-svc/ba	atches/cache/sync'
<pre>'POST' -Head 2. \$response 3. Python Meth 1. import re 2.</pre>	ers \$headers   ConvertTo-Json od quests	0.99.104/api/axiom-results-svc/ba	tches/cache/sync' -Method
4. 5. payload = 6. headers = 7.	0 0	ts-svc/batches/cache/sync" url, headers=headers, data=paylo	ad)
print(respor	se.text)		
wget Metho	1		
2metho 3timeo 4heade			

## Archiving with AutoAx v1.2

<b>ANT!</b> All archiving procedures listed below are intended for script writers, command line d those with HTTP and Linux experience only.
Use care when archiving <b>/srv/core/roots/rootdata</b> . Root data is used as input to the analysis workflow processing when creating batches and for the <b>Reanalysis</b> feature.
If a batch is queued/status as <b>Pending</b> or <b>Running</b> and the data is removed, the Reanalysis for that batch will FAIL.
All APIs in v1.2 for results (batch information) uses a GraphQL calling system. Graphql is provided as part of AutoAx. To access this sub-system, enter the following in your browser's address bar:
<ip address="" domain="" name="" of="" server=""  ="">/api/axiom-results-svc/gql/graphql</ip>
Internally, all batches are identified and referenced by the <b>batchId</b> , not the ID.
The batchId is a <u>Universal</u> <u>Unique</u> Identifier (UUID)
If you want to remove a batch from the system without having to ssh, delete, or refresh the cache, use the remove api. To do this enter:
mutation { remove(batchId: "") }
Replace the with the batchId (this is the UUID not the displayed ID)
To view the UUIDs for each batch and to find the ones you want enter:
query { batches { id batchId
}
Use the browser's Find feature (ctrl+f) to search for the ID you are looking for in the lower left corner.
From the <b>remove</b> command line, use the <b>batchId</b> from each batch you want to remove. Repeat this process for <u>each</u> batch. This will remove the batch from the server and cache memory.
<ol> <li>After the batches have been archived off the server, replace the IP address http://10.80.100.145/api/axiom-results-svc/gql/graphql with your server's IP address.</li> </ol>
2. In the tab that appears, enter:
mutation{ reCacheBatchinfo All caches are refreshed.





## Using the extension pane

The Extension pane (right) manages features or settings available for export or analysis options beyond the Best Practices and Best Practices with Dual Workflows.

Extensions are enabled when available, as types vary depending on the array design. **Note:** Not every extension is available for every array design.

1. To set a specific Extension as default with no customization, toggle **Default** on. (Figure 58) When on, the selected export file (from the drop-down) is automatically generated and placed in the **Extension** folder of your batch.

VCF extension When set as default, a VCF export file is automatically generated and placed in the **Extension** folder of your batch.

#### Input tab

#### (Figure 58)

Note: Depending on what array you are running, the extension options may differ.

- Click the available drop-down to select a different file or add a new file.
- Optional: Click the **Standard Chromosomes** check box to include only probsets that are assigned to standard chromosomes or other annotated contigs.
- To add a file to the VCF Input tab entries above, see "Adding a file not listed" on page 36.

Figure 58 Extension	pane - VCF Input
✓ VCF	× ~
Extension	
Default 💶	① Create ② Reset to Defaults
Input Output	
ProbeSet List Filter	
SNPolisher/Recommended.ps	~
Standard Chromosomes	
Annotation File	
Axiom_BloodGenomiX.na36.r1.	a1.annot.db 🗸
SNP Identifier	
probeset_id	~
	$\otimes$

#### Output tab

(Figure 59)

gure 59 Extensio	on pane - VCF Output
VCF	× ~
Extension	
Default	$(f)$ Create $\begin{array}{c} \ensuremath{\mathcal{C}}^{2} \end{array}$ Reset to Defaults
Input Output	
Output Folder	
extension/vcf/{datetime}	
Name	
{ARRAY}_{batchname}.vcf	
Single Sample per File	
Include SNP Metrics	
	$\otimes$

• **Output Folder:** Displays the output folder's location.

- C
- **Name:** Displays the selected extension's file naming convention.
- **Single Sample per File:** Click this check box to generate one file per sample.
- Include SNP Metrics: Click this check box to include CR, FLD, HomFLD, HomRO, and HetSo to your analysis.

**Note:** Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

#### AxLE extension Input tab

(Figure 60)

**Note:** To learn more about these AxLE Export inputs, see the *Axiom Long Format Export User Guide* (P/N MAN0027964).

Figure 60 Extension	n pane - Ax	kLE Export
AxLE Export	×	. ~
Extension		
Default		$\mathcal{C}$ Reset to Defaults
Input Output		
Sample Name Mapping and F	ilter File	
Select a file		~
SNP List Filter		
Select a file		~
SNP Mapping File		
Select a file		~
SNP Allele Swap		
Select a file		~
	$\odot$	

Note: Depending on what array you are running, the extension options may differ.

- Click the available drop-down to select a different file or add a new file.
- To add a file to the entries above, see "Adding a file not listed" on page 36.



#### Output tab

(Figure 61)

**Note:** To learn more about these AxLE Export outputs, see the *Axiom Long Format Export User Guide (*P/N MAN0027964).

gure 61 Extension	pane - AxLE Export
AxLE Export	× ~
Extension	
Default	① Create
Input Output	
Genotyping Export File	
{ARRAY}_genotype_{yyyy	MMddHHmmss}.txt
SNP Map	
{ARRAY}_snp_map_{yyyy	MMddHHmmss}.txt
Sample Export File	
{ARRAY}_sample_{yyyyM	MddHHmmss}.txt
Sample Map	
{ARRAY}_sample_map_{y	ryyyMMddHHmmss}.txt
Zip File	

- **Genotyping Export File:** Displays the file's naming convention.
- **SNP Map:** Displays the SNP Map's naming convention.
- **Sample Export File:** Displays the file's naming convention.
- **Sample Map:** Displays the Sample Map's naming convention.
- **Zip File:** Displays the file's naming convention.
- Include SNP Metrics (scroll down to view): Click this check box to include CR, FLD, HomFLD, HomRO, and HetSo to your analysis.

## Allele Translation extension

An **Allele Translation** extension appears as an extension option (Figure 62) if the arrays you are using have been enabled. See the *Axiom Analysis Suite User Guide* (P/N MAN0027928) for more information.

Input tab

Note: Depending on what array you are running, the extension options may differ.

- Click the available drop-down to select a different file or add a new file.
- If you disagree with the phenotype interpretations, you may want to leave the Metabolizer File option blank. If left blank, a Phenotype report will not be created. If you want to include Sample Attributes, click its check box.

Figure 62 Extension pane	- Allele Tr	anslation
Allele Translation	× ~	]
Extension		
Default		Reset to Defaults
Input Output		
Annotation File		
PharmacoFocus.r5.dc_annot.csv		~
Translation File		
PharmacoFocus.r5.translation		~
Metabolizer File		
PharmacoFocus.r5.metabolizer		× ~
Include Sample Attributes		
ProbeSet List Filter		
SNPolisher/Recommended.ps		~
	$\otimes$	

#### Output tab

Click on the Extension pane's Output tab to view the Output Folder location and naming convention. Click the Default toggle on to automatically run Allele Translation for arrays that are enabled. Note: If any options have been modified the Create and Reset to Default buttons appear.

Figure 63 Extension pane	e - Allele T	ranslation
✓ Allele Translation	× ~	•
Extension		
Default		C Reset to Defaults
Input Output		
Output Folder		
extension/translation/{datetime}		
Name		
{batchname}		
	$\odot$	

#### Axiom HLA extension For arrays that are enabled, generating human leukocyte antigen calls is an extension option. Axiom HLA uses a multi-population reference panel and the HLA type imputation model HLA\*IMP:021 to statistically infer the HLA types of human samples from genotype data. The loci include Class I (A, B, C) and Class II (DRB1, DRB3, DRB4, DRB5, DQA1, DQB1, DPA1, DQB1).

Input tab

(Figure 64)

Figure 64 Extension pane - Axiom HLA		
Axiom HLA × v		
Extension		
Default		$\stackrel{ extsf{reset}}{\sim}$ Reset to Defaults
Input Output		
Loci Selection	_	
All	•	
0		

1. Click on the Loci Selection drop-down.

**Note:** Once a new Loci is selected, the **Create** and **Reset to Defaults** buttons become enabled.

2. All Locis are selected by default. (Figure 65) Click on a check box(es) to de-select a Loci from your analysis.

Figure 65 Loci Selection	on drop-down
Loci Selection	
All	~
🗹 (Default) All	
A	<b>^</b>
🛃 В	
C C	
DPA1	
DPB1	-

#### Output tab

(Figure 66)

Figure 66 Extension pane - Axiom HLA		
Axiom HLA × ~		
Extension		
Default		C Reset to Defaults
Input Output		
Output Folder		
extension/hla/{datetime}		
Output Name	2	
AxiomHLA_{digit}_{loci}_Results		
©		

- **Output Folder:** Displays the output folder's location.
- **Output Name:** Displays the selected extension's file naming convention.
- (Optional) Click the **Default** toggle right (on) to apply this extension to your designated Axiom HLA array type.



### Accessing extension outputs

For direct access to the BloodGenomiX Report (and any other final output), AutoAx v2.0 contains this additional folder in its directory tree: /srv/core/roots/transfers

- The **transfers** folder contains sub-folders for each extension.
- The BloodGenomiX Report resides here:

/srv/core/roots/transfers/BGX

Enabling access to final outputs

Direct access to final outputs is set to **on** by default. If this feature needs to be restored, go to path:

/srv/core/configs/apps/automated-axtiom/settings/autoXfer.settings.json

- 1. nano into the file.
- 2. Change line #30 *transfer* property to **true** (Figure 67), then press **Enter** to save the .json file.

Figure 67	Editing the transfer property example
27	t and a second terrain of the second s
28 29	<pre>"name": "best_practices_for_blood",</pre>
	"internalBatchPath": "BGX",
30	"transfer": true,
31	"destination": "transfers/BGX",
32	"deleteSrc": false,
33	"type": "xfer",
34	<pre>"imageTag": "core/core-sh:latest",</pre>
34 35	"taskType": "lite"
36	},

**Note:** If you modify the **.json** file with tasks still in the pipeline, those tasks will complete using the previous .json file. Your saved *transfer* property edit will take effect AFTER the task pipeline clears.

For support visit **thermofisher.com/support** or email **techSupport@thermofisher.com** thermofisher.com

