applied biosystems

# Automated Axiom™ 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.<br>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. |

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# 1 Getting started

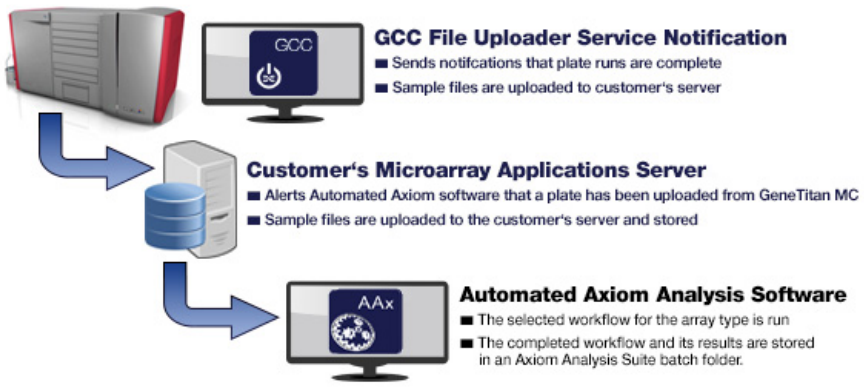
## Introduction

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.

**Figure 1** The Automated Axiom Analysis workflow




**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.

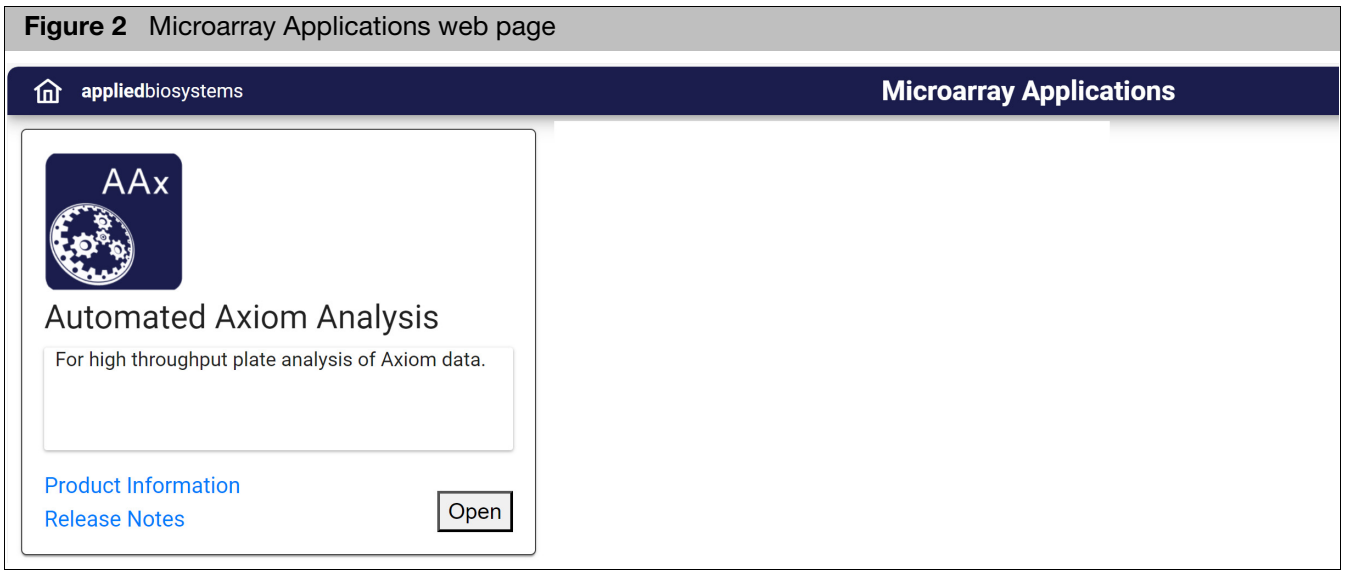
# Launching AutoAx

**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

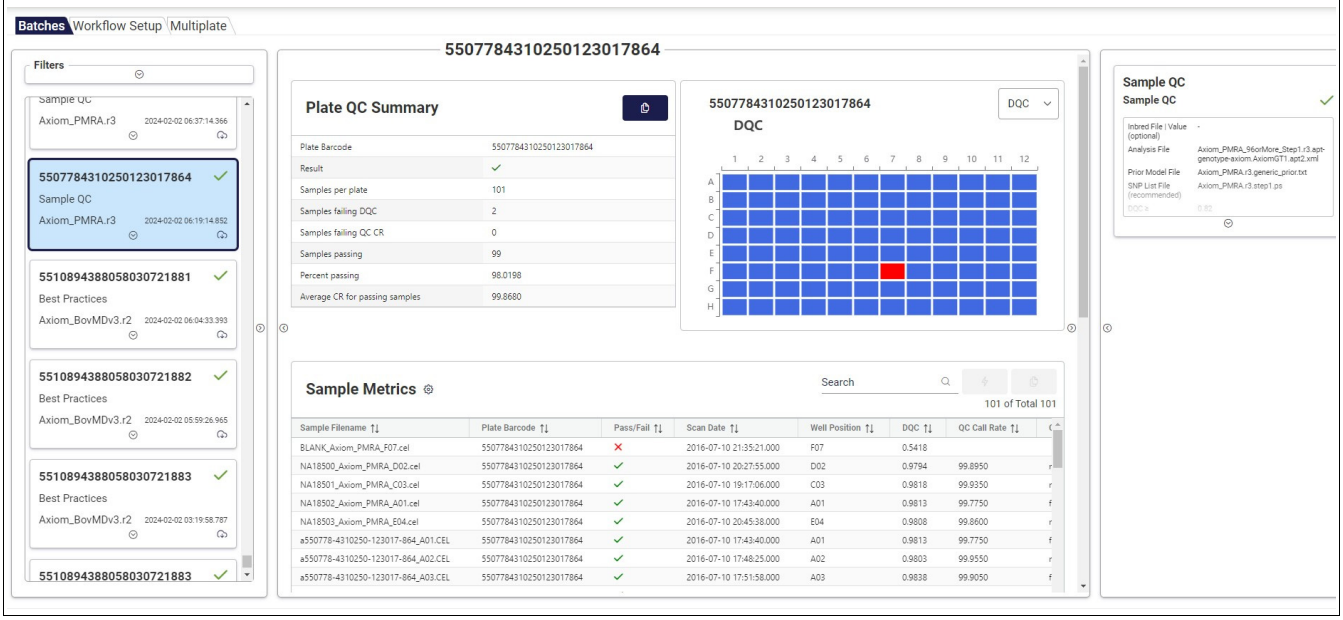



- Click the Product Information link to learn more about the software.
- Click the Release Notes link for information about this v2.0 release.

2. Click **Open**.

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

**Figure 3** Batches window tab - Populated example




# 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

1. 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**.

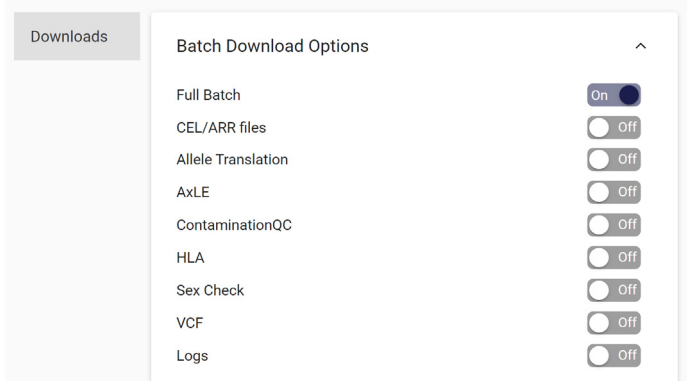
   The Batch Download Options window appears. (Figure 4)

## 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

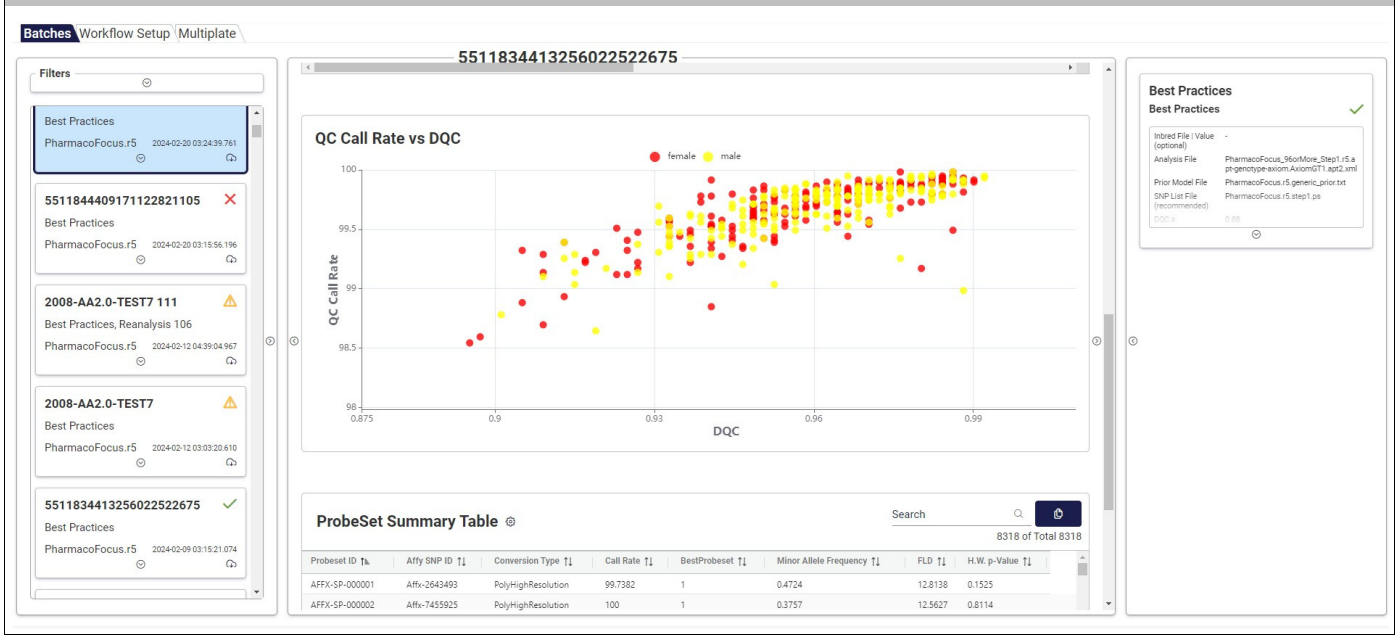



2. Click on the **Batches** tab to close the window.

# 2 Batches

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

**Figure 5** Batches window tab - Populated example

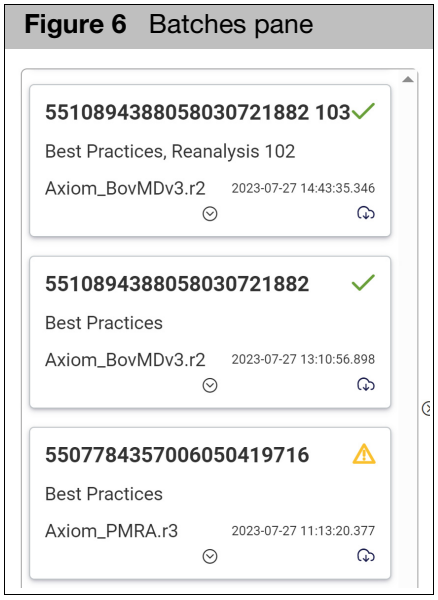



## Using the Batches window tab

### Batches pane

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

**Figure 6** Batches pane




- Card symbols:

  = Completed

  = Error

  = Warning

  = Download

  = Running

  = Pending

- 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.

**Figure 7** Expanded (All) cards view

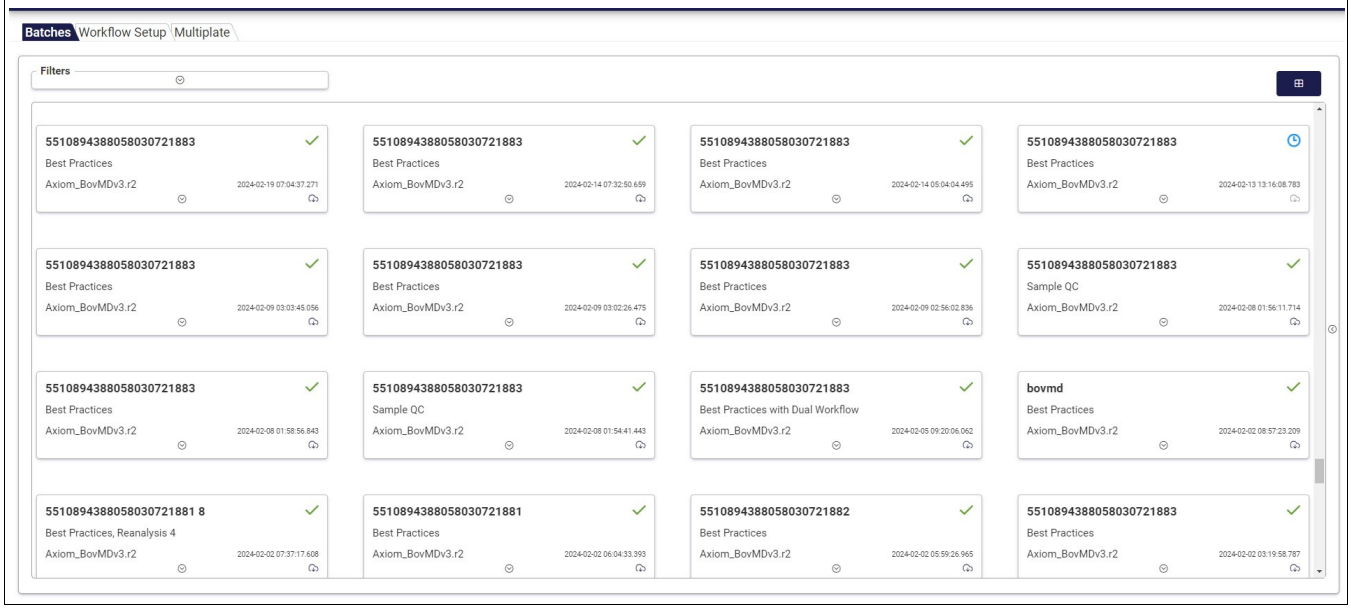
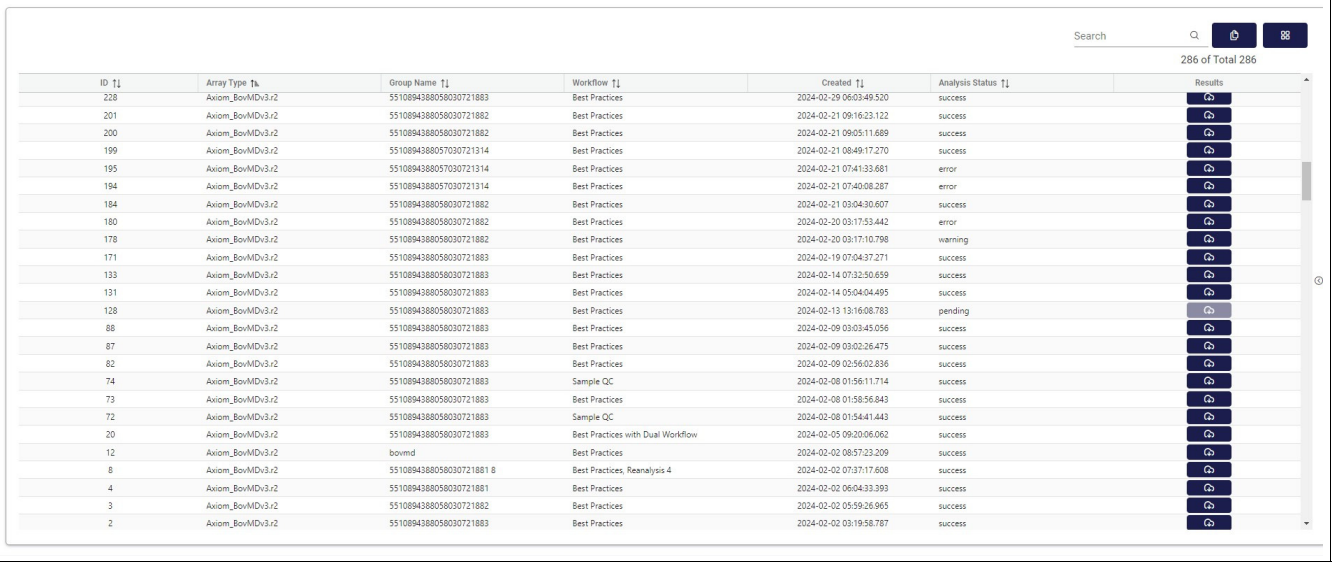


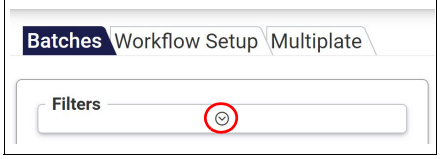

**Figure 8** Expanded (All) Table view



## Filtering batches

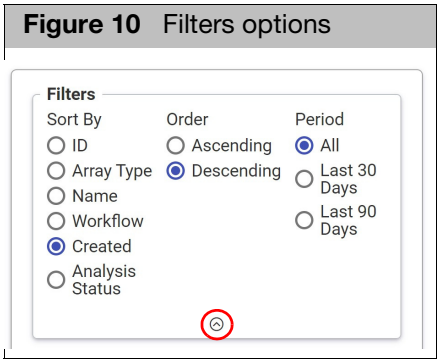
1. At the main Batches window tab, click the **down arrow** button under **Filters** (Figure 9) to display the filters.

**Figure 9** Filters button




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 options

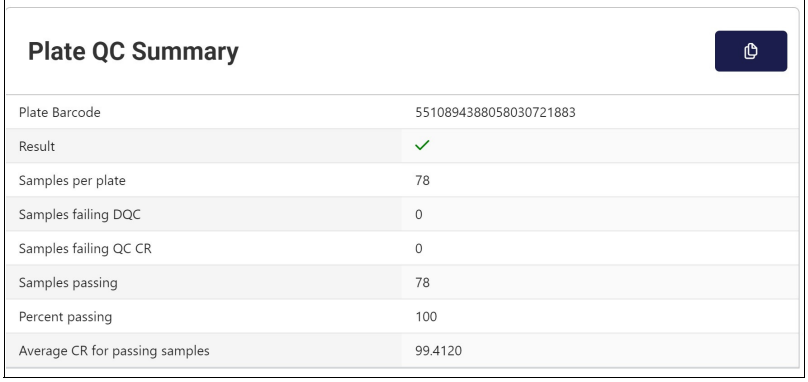



# Viewing a selected batch

## 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 Barcode | 5510894388058030721883 |
| Result | ✓ |
| 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.

**Table 1** Plate QC Summary table

| Column | Description |
|---|---|
| **Plate Barcode** | Barcode of the plate that was analyzed in the batch. |
| **Result** | Based on QC thresholds set in analysis configuration template. **Note:** 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.<br>* BloodGenomiX Reporter analyses only. |
| **Number of BGX Reports used *** | Number of BGX reports used per plate.<br>* BloodGenomiX Reporter analyses only. |
| **Sex Check *** | Number of samples with confirmed computed sex.<br>* BloodGenomiX Reporter analyses only. |

### 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.

**Figure 12** Plots drop-down


- Each sample value is shown in its well location.

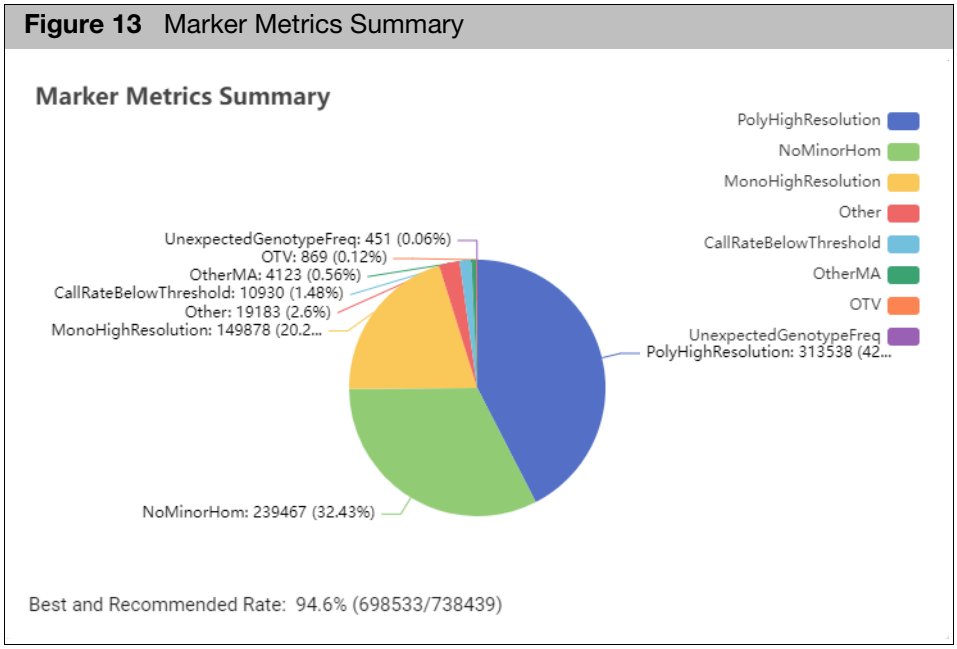
- Wells with failed samples are shown in **red**.
- A white well denotes an empty well or one that was not calculated.
- Mouse over a well to view its details.

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

## Marker metrics summary

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

**Figure 13** Marker Metrics Summary




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

## Sample metrics table

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

**Figure 14** Sample Metrics table

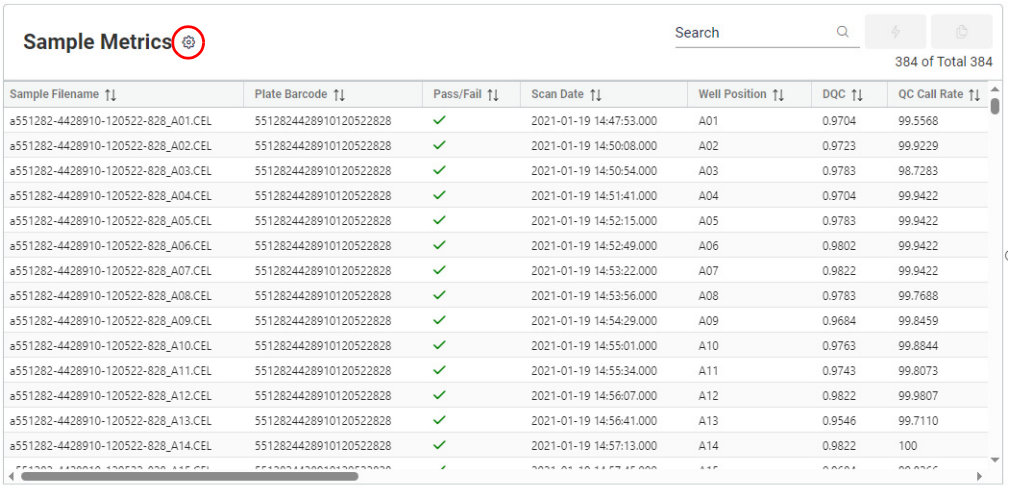



**Table 2** Sample Metrics table *

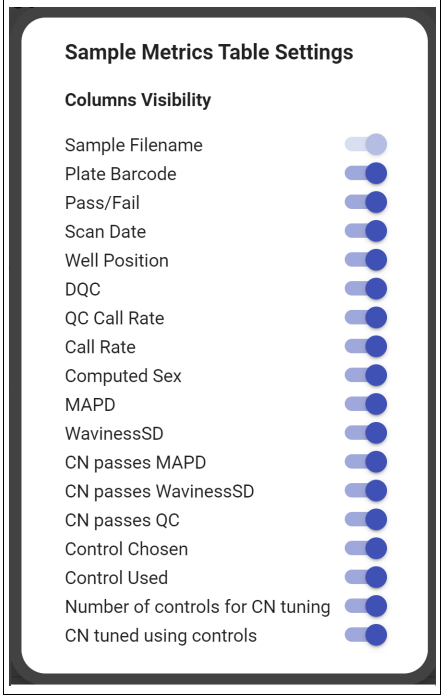
| Column | Description |
|---|---|
| **Sample Filename** | Name of CEL file. |
| **Pass/File** | Sample quality control status.<br>✓ = The Sample QC step deems it meets the minimum thresholds for DQC, QC call_rate, and average call rate for passing samples.<br>✗ = The sample did not pass Sample QC. |
| **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. **Note:** 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. **Note:** 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. **Note:** 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.<br>**Note:** 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** icon.
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 Table Settings window




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

1. 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.

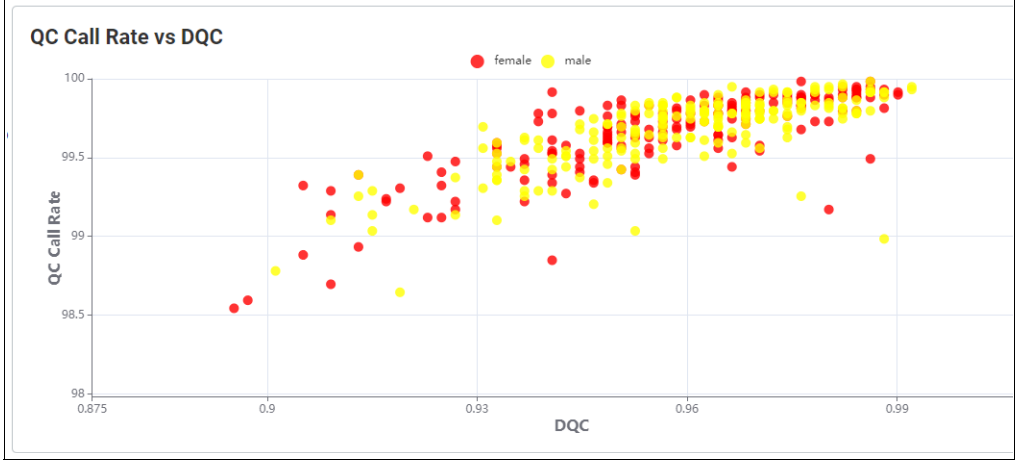
### Copying the table

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

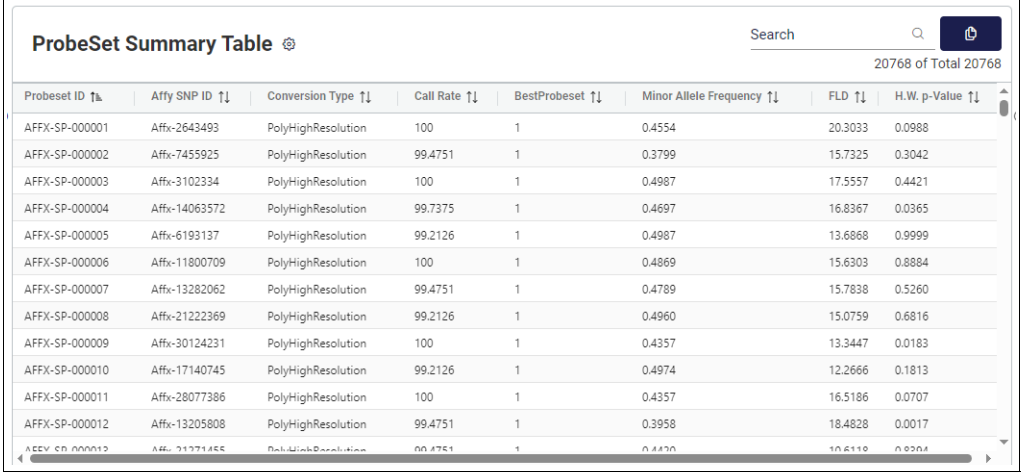
**Figure 16** Scatter plot - QC Call Rate vs DQC




## ProbeSet Summary table

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

**Figure 17** ProbeSet Summary Table




| 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 |

1. 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 18** ProbeSet Summary Table Settings window

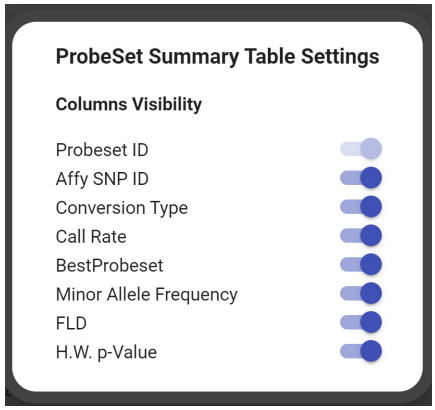



**Table 3** ProbeSet Summary Table

| 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: $P_A = \frac{(\#\ AA\ Calls + 0.5 * AB\ Calls)}{Total\ \#\ Calls}$ Where the Total # Calls does not include the No Calls. The B allele frequency is . $P_B = 1 - P_A$ The minor allele frequency is the Min($P_A$, $P_B$). |

**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** | 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.<br>There are two statistical tests used for HWE. When AA, AB, and BB counts are all >=10, a Chi-squared test is used. When one or more of the AA, AB, and BB counts are <10, an Exact test is used. An Exact test means that the p-value is calculated exactly and not approximated from a population distribution. |

### 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.

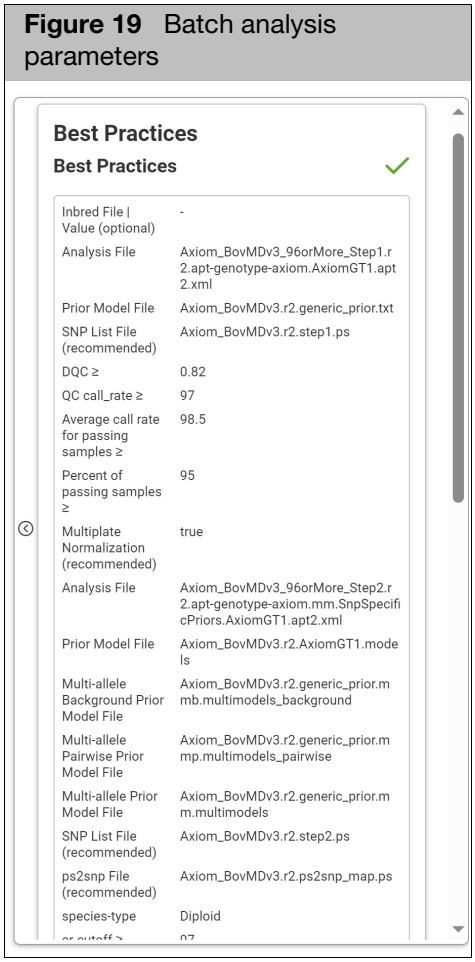
1. Click the  button to copy the table.
2. Open Word or an Excel file.
3. Press **Ctrl v** to paste the table data.

## 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.

**Figure 19** Batch analysis parameters



# 3 Workflow Setup

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

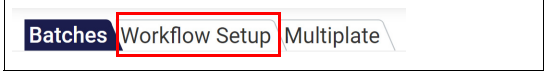
- "Sample QC"
- "Best Practices" on page 27
- "Best Practices with Dual Workflow (Agbio Arrays only)" on page 32

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

## Sample QC

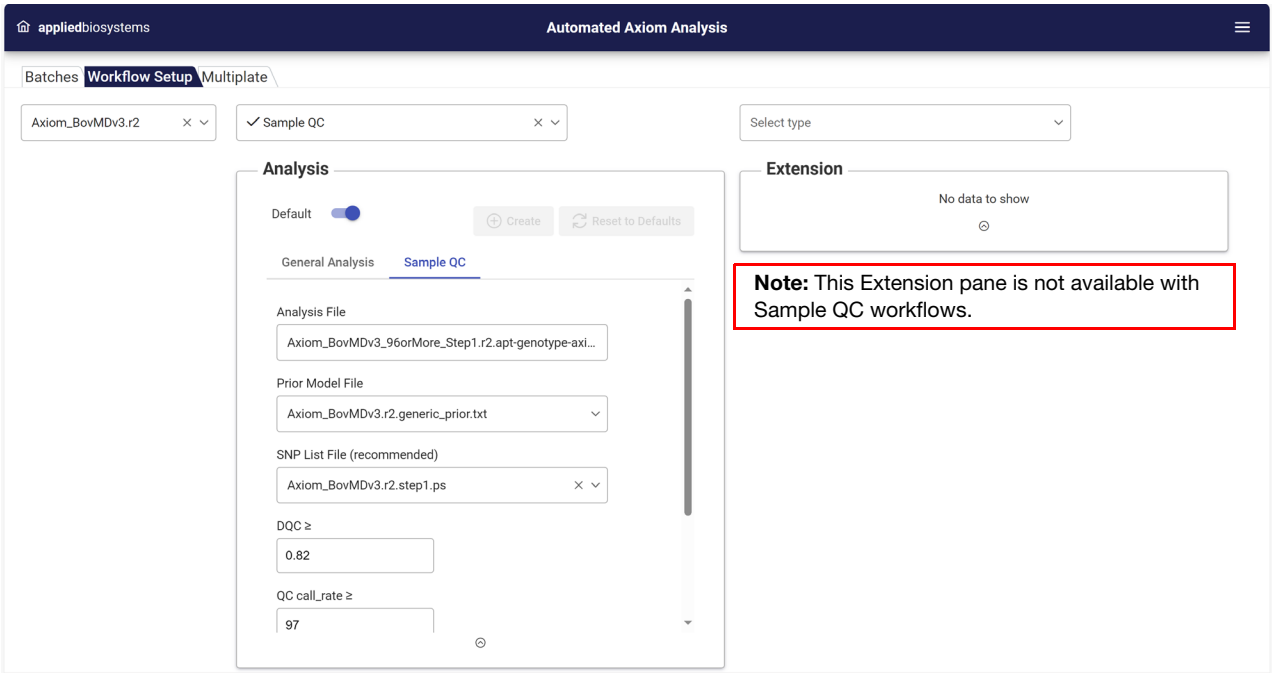
1. Click the **Workflow Setup** tab. (Figure 20)

**Figure 20** Workflow Setup tab




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

**Figure 21** Workflow Setup window tab - Populated example

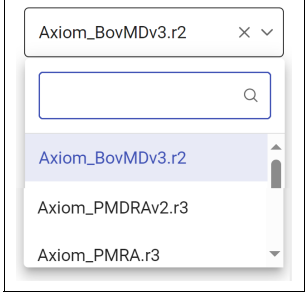


# Customizing an analysis

## 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 Type




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

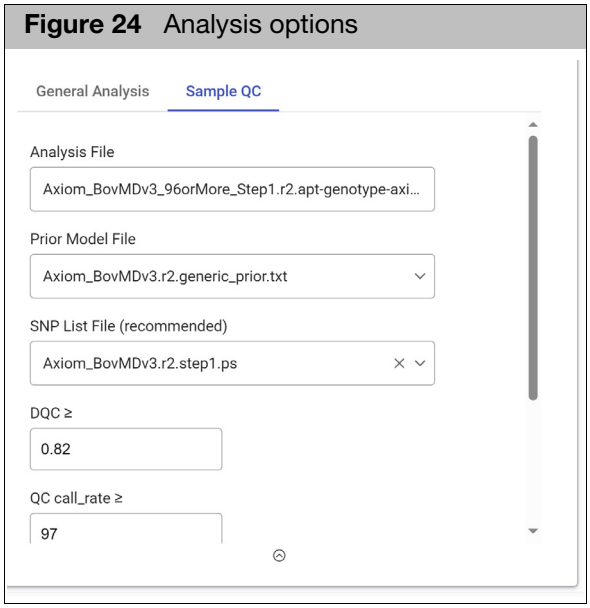
**Figure 23** Workflow drop-down


## 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



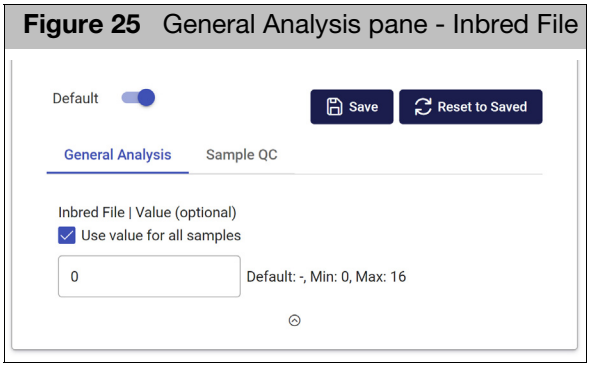

- **Analysis File**: Displays the array you selected.
- **Prior Model File**: Displays the default *Prior Model File* associated with your selected array. Click the drop-down to select a different file or add a new file.
- **SNP List File (recommended)**: Displays the default *SNP List File* associated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

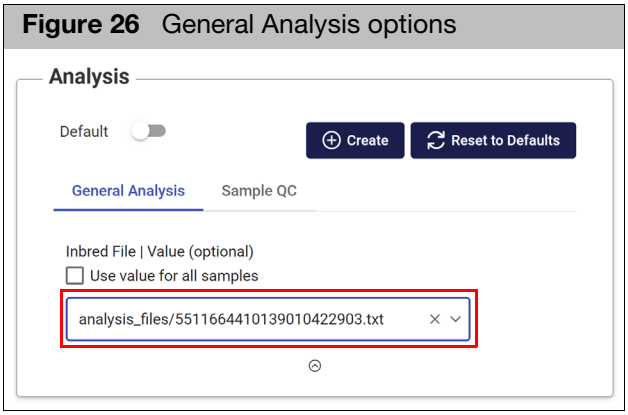
1. Click the check box, then enter a value. (Figure 25)

**Figure 25** General Analysis pane - Inbred File




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

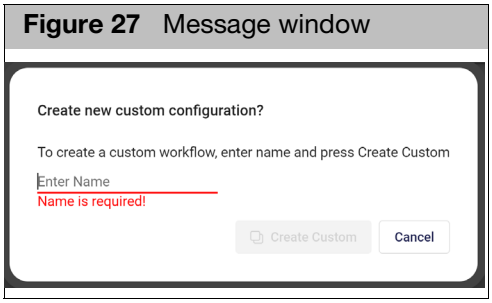



## Saving your custom configuration

1. Click **Create**. (If modifying an existing custom workflow, click **Save**).

   After clicking **Create**, a message window appears. (Figure 27)

**Figure 27** Message window




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.

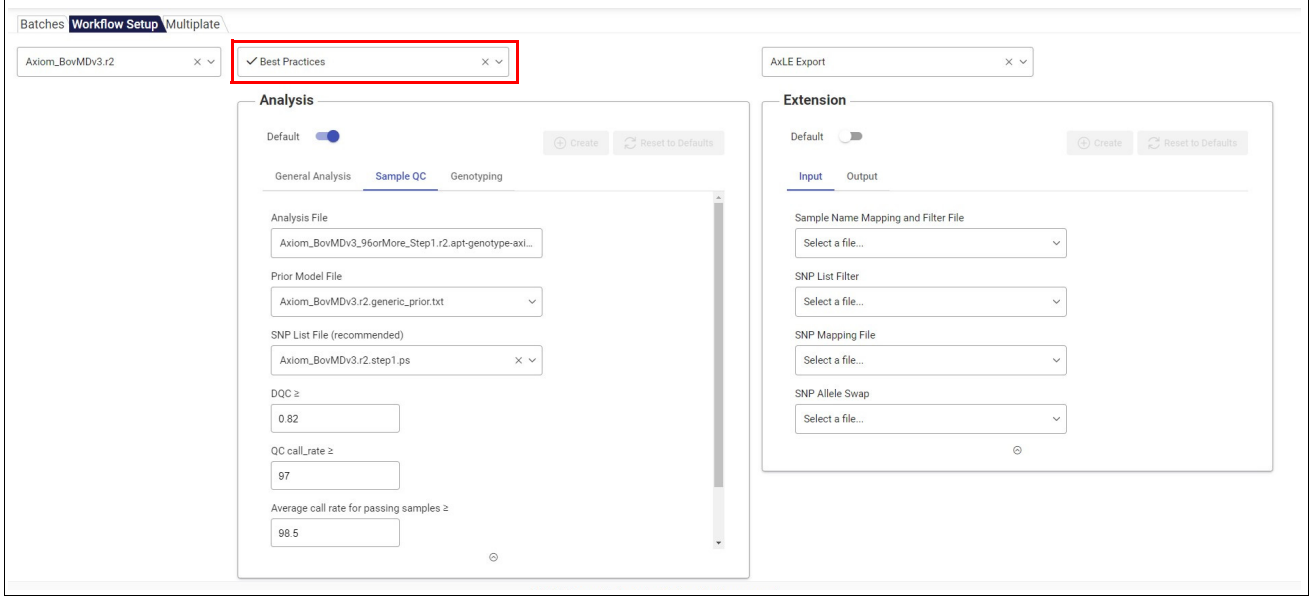
1. Click **Save**.

   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)

**Figure 28** Workflow Setup window tab - Populated example

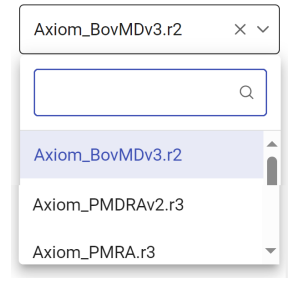



## Customizing an analysis

### 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 Type



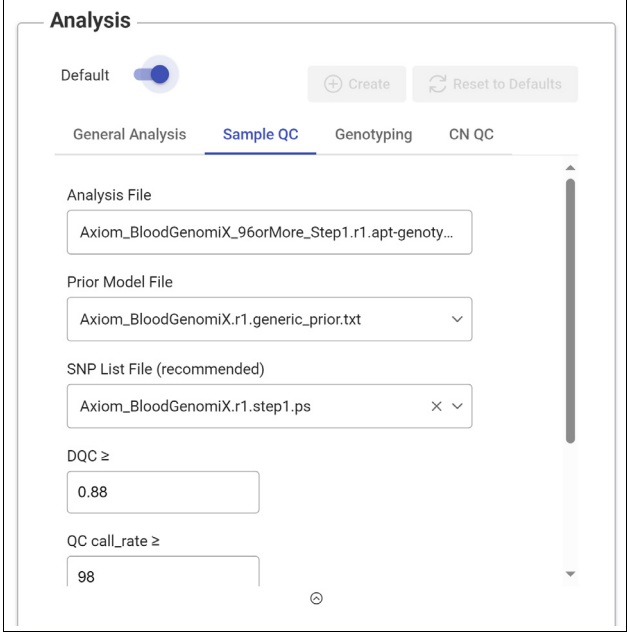

2. Click the Workflow drop-down to select **Best Practices.**

## 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.

**Figure 30** Analysis options




### 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 drop-down.
- **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:

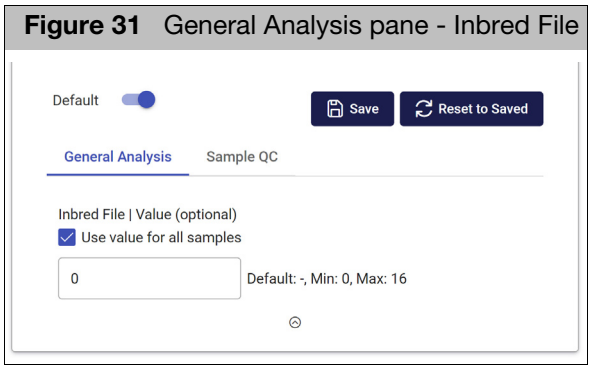
- **MAPD**: For arrays that support copy number (CN) analysis, the Median Absolute Pairwise Difference of log2ratio signals of adjacent copy number (CN) probesets must be less than this value to make CN calls.
- **Waviness SD**: For arrays that support copy number (CN) analysis, the Waviness Standard 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

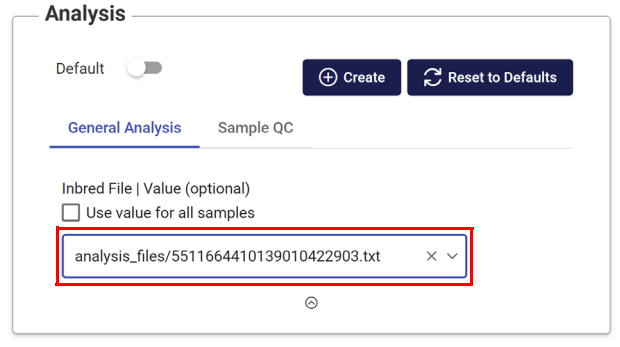
1. Click the check box, then enter a value. (Figure 31)

**Figure 31** General Analysis pane - Inbred File




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.

**Figure 32** General Analysis options

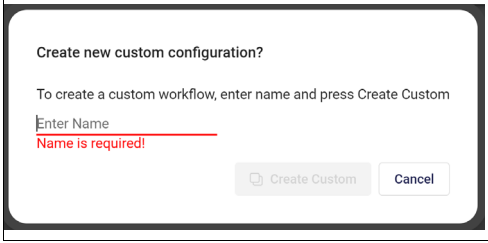



### Saving your custom configuration

1. Click **Create**. (If modifying an existing custom workflow, click **Save**).

   After clicking **Create**, a message window appears. (Figure 33)

**Figure 33** Message window




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.

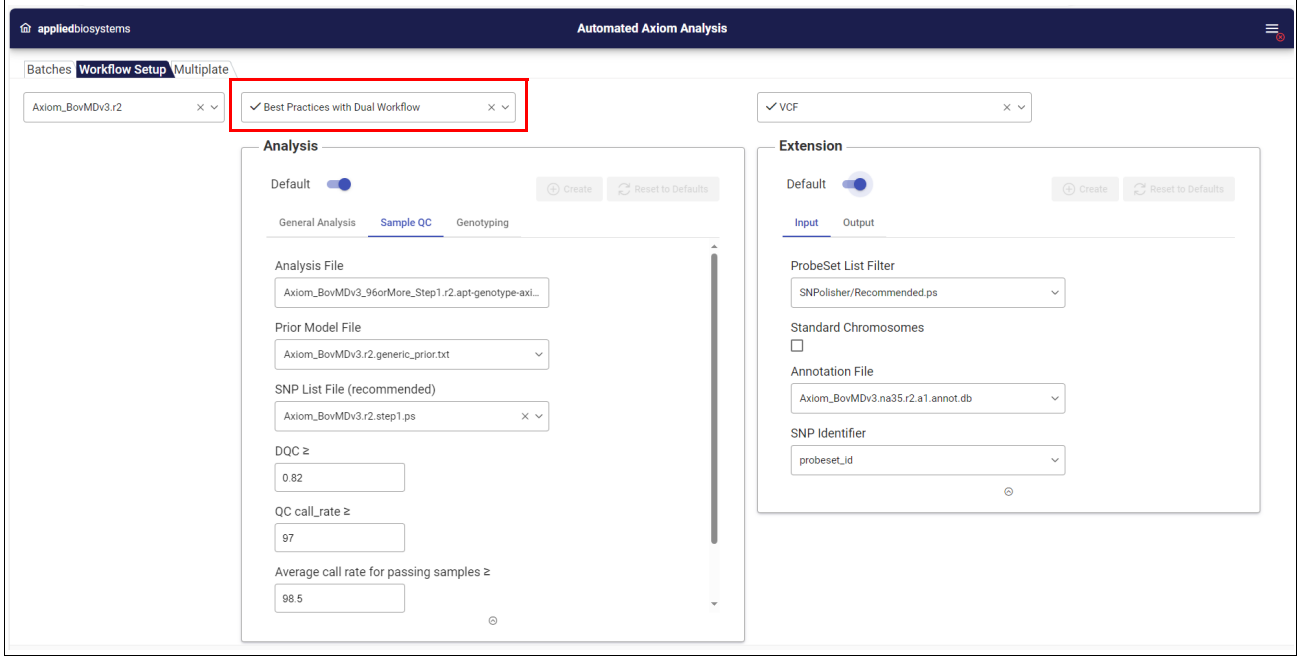
To use the Extension pane, see "Using the extension pane" on page 61.

# 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

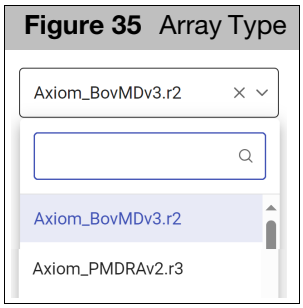



# Customizing an analysis

## 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



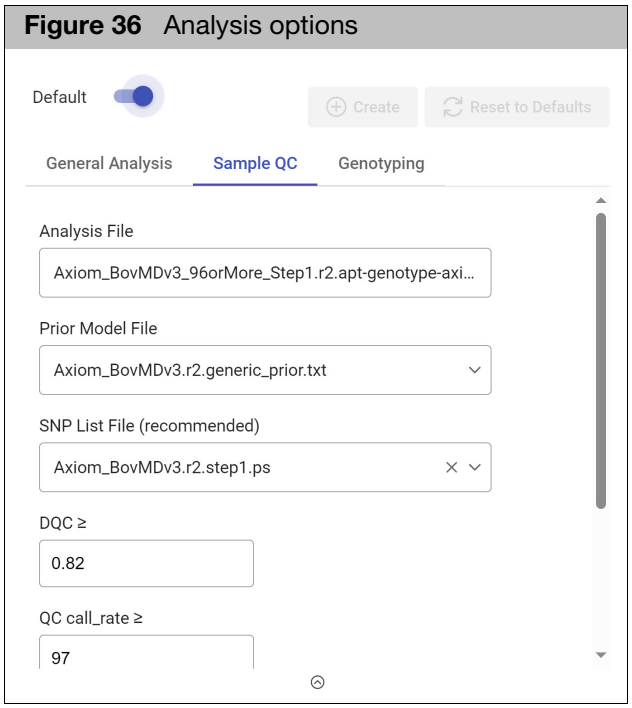

2. Click the Workflow drop-down to select **Best Practices with Dual Workflow.**

## 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




### 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:

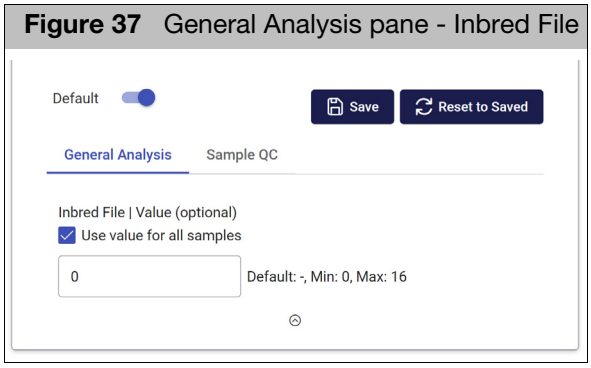
- **Multiplate Normalization (recommended)** becomes enabled for batches using more than one plate.
- **CN Reference Model File**: Contains reference information for CN analysis step.
- **CN Priors File**: Specifies for each predefined CN Region the callable CN states, and the MedianLog2Ratio boundaries defining each state.
- **Analysis File**: This field displays the array you selected in the Array Type drop-down.
- **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

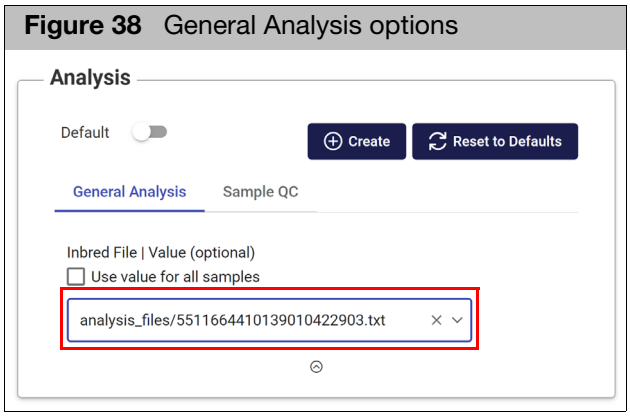
1. Click the check box, then enter a value. (Figure 37)

**Figure 37** General Analysis pane - Inbred File




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

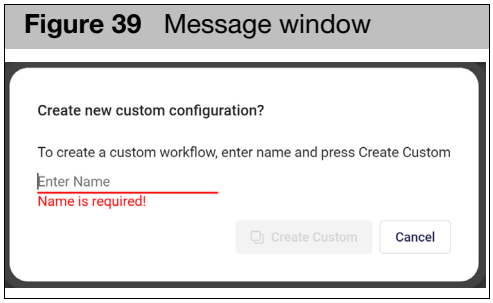



## Saving your custom configuration

1. Click **Create**. (If modifying an existing custom workflow, click **Save**).

   After clicking **Create**, a message window appears. (Figure 39)

**Figure 39** Message window




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

1. 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.

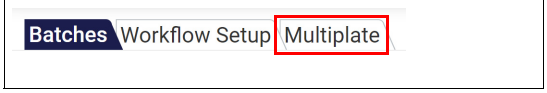
   If you are modifying an existing custom configuration the **Save** button is now enabled. If you are modifying a factory default workflow the **Create** button is enabled.

# 4 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



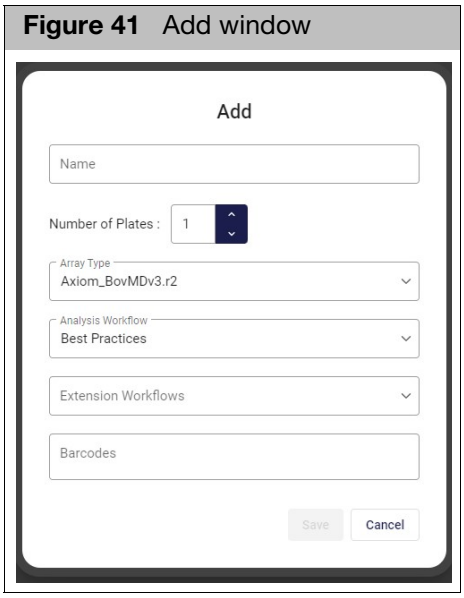

The **Multiplate** window appears.

## Using the Multiplate window tab

### Adding a new multiplate

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

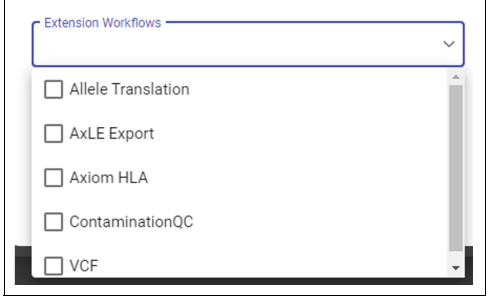
**Figure 41** Add window




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.

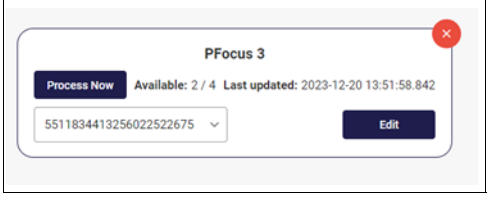
**Figure 42** Extension Workflows




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)

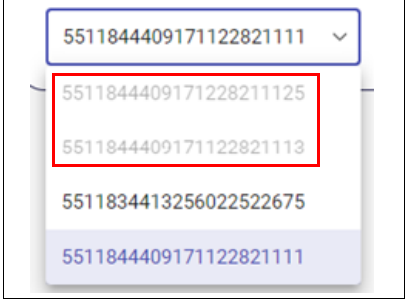
**Figure 43** New multiplate card




## 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.

**Figure 44** Barcode drop-down



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

   A Confirmation pop up window appears.

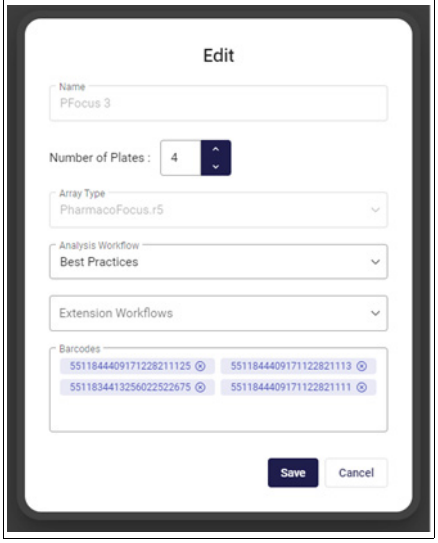
10. Click **OK** to start processing the available plates.

## Editing a multiplate group

1. Click the **Edit** button.

   The Edit window appears. (Figure 45)

**Figure 45** Edit window



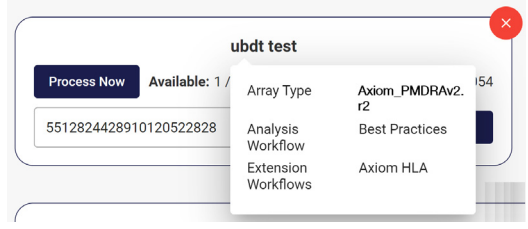

Use the Edit window to change:

- 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 quick view

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

**Figure 46** Quick view




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

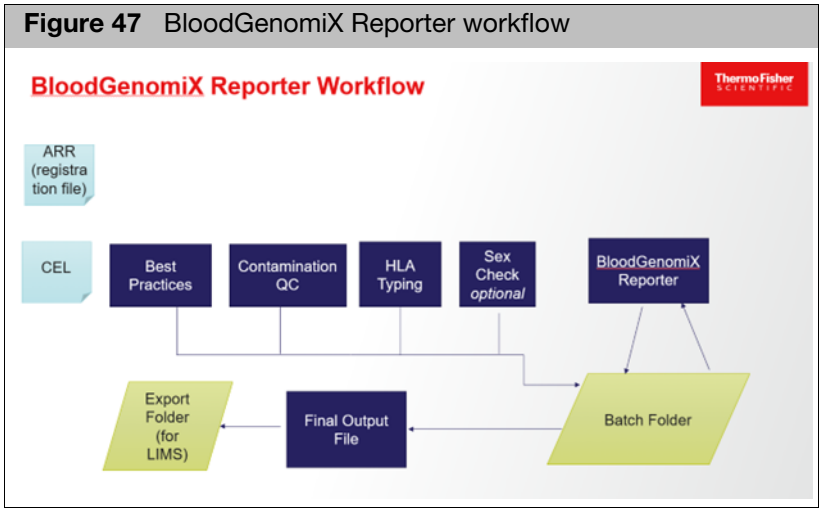
# A BloodGenomiX Reporter

**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:

**Figure 47** BloodGenomiX Reporter workflow




- 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.

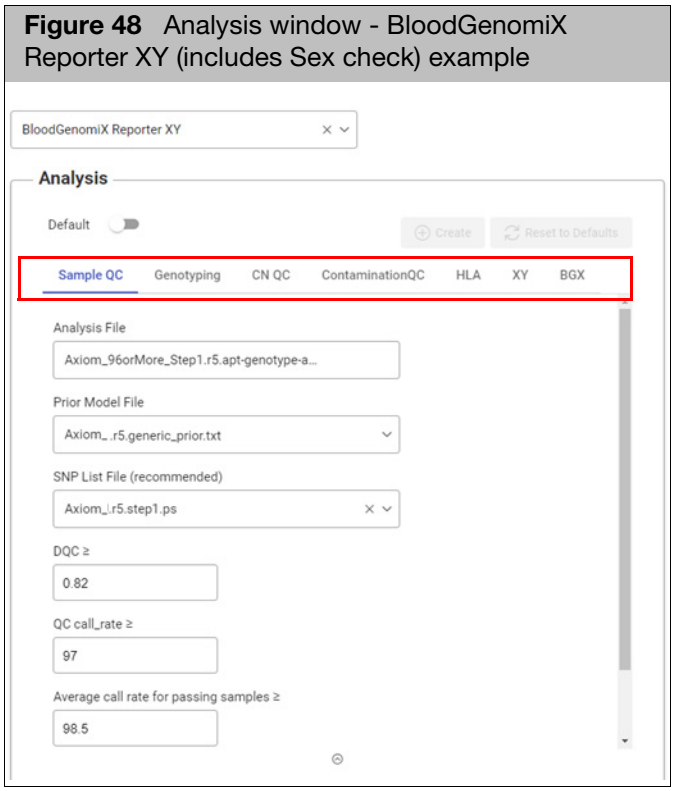
If the BloodGenomiX Reporter XY workflow is selected, computed sex is compared to the supplied sex, then either a PASS or FAIL will be reported. To generate a report *without* Sex Check, select **BloodGenomiX Reporter**.

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.

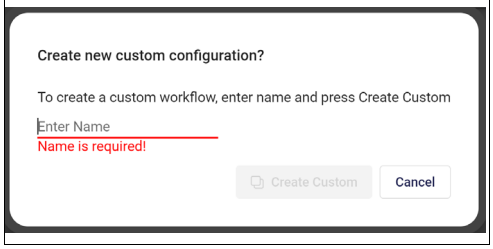
**Figure 48** Analysis window - BloodGenomiX Reporter XY (includes Sex check) example




**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.

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

**Figure 49** Message window




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

## BloodGenomiX Analysis window tabs

### Sample QC (fields and descriptions)

| | |
|---|---|
| **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. **Note:** 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)

| | |
|---|---|
| **Multiplate Normalization** | Multiplate normalization is recommended (checked by default) for 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 types. |
| **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. |
| **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. |
| **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 (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 **No** in the output. |
| **Threshold estimated contamination** | Samples with a value less than threshold will have contaminated set to **No** in the output. |

### HLA (fields and descriptions)

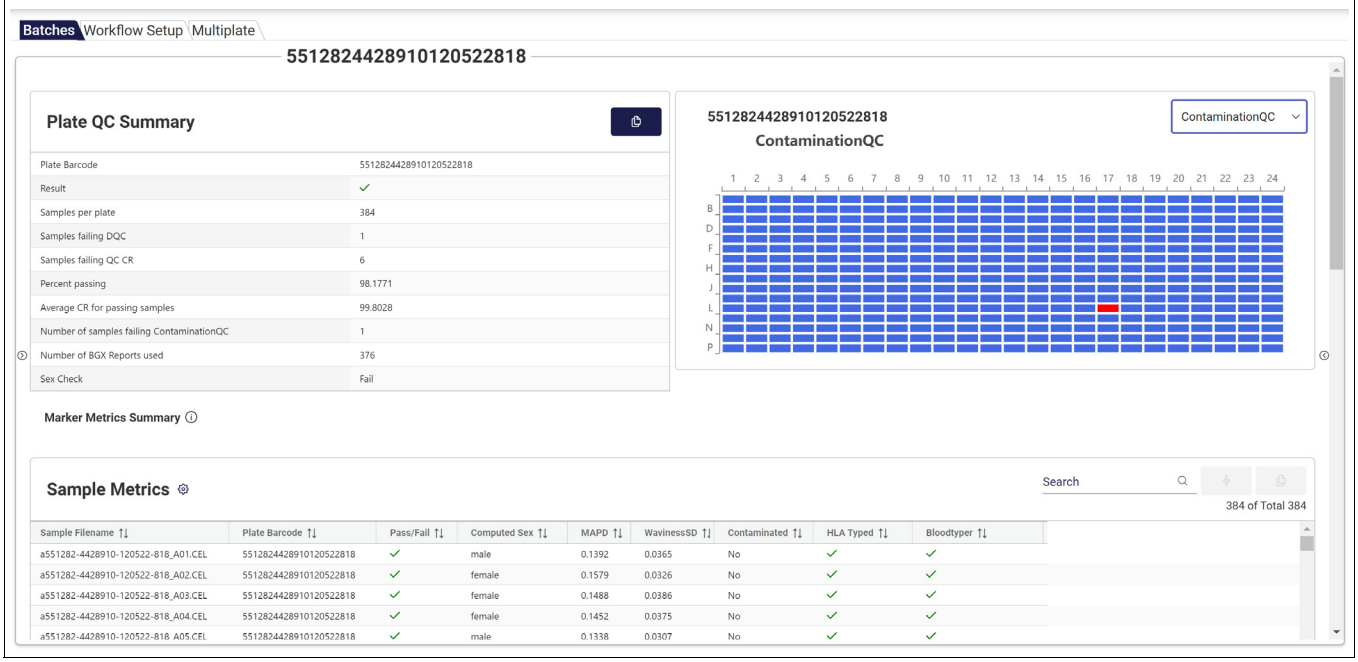
| | |
|---|---|
| **Loci Selection (default All)** | Loci A, B, C are options for HLA typing. |
| **Output Folder** | Location and name of output folder. |
| **Output Name** | Name of the output which includes 2 or 4 digit as well as loci. Example: (**AxiomHLA_2_A_Results**) |

### 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

**Figure 50** Batches window tab - BloodGenomiX Reporter example




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.

## BGXPrimary folder contents

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 |

| | |
|---|---|
| **result_value** | **Positive**: Selected test_name is detected in the sample,<br>**Negative**: Selected test_name is not detected in the sample,<br>**NT**: Not Typed, selected test_name antigen was not analyzed<br>**Not_present**: Selected test_name allele is not detected in the sample.<br>**HEM**: Test_name allele is present as hemizygous<br>**HET**: Test_name allele is present as heterozygous<br>**HOM**: Test_name allele is present as homozygous<br>**PARTIAL**: Phenotype is partially expressed<br>**WEAK**: Reduced phenotype expression |
| **result_status** | **PASS**: Sample is found to be either positive or negative for the corresponding test_name.<br>**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 subfolders

### Contamination

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<br><br>**Note:** 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 |
|---|---|---|---|---|---|---|
| ARR | Attributes | Well location | Sample Attributes | well | string | PASS/FAIL |
| | | Source plate barcode | | barcode | string | PASS/FAIL |
| | | Source plate well location | | well | string | PASS/FAIL |
| | | sample type | | source of DNA | string | PASS/FAIL |
| | | 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 |
|---|---|---|---|---|---|---|
| apt2-contamination-qc version # | ContaminationQC | p-value | ContaminationQC | Example: 0.476659 | integer | PASS/FAIL |
| | | Estimated contamination | | 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 |
| | | 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 |
|---|---|---|---|---|---|---|
| AutoAx 2.0.0 | QC | DQC | Sample QC Metrics | example: 0.98617 | float | PASS |
| | | QC Call Rate | | example: 99.84586 | float | PASS |
| | | MAPD | | example: 0.136106 | float | PASS/FAIL |
| | | 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 | genotype | Forward strand or Copy Number Call | genotype | PASS/FAIL |

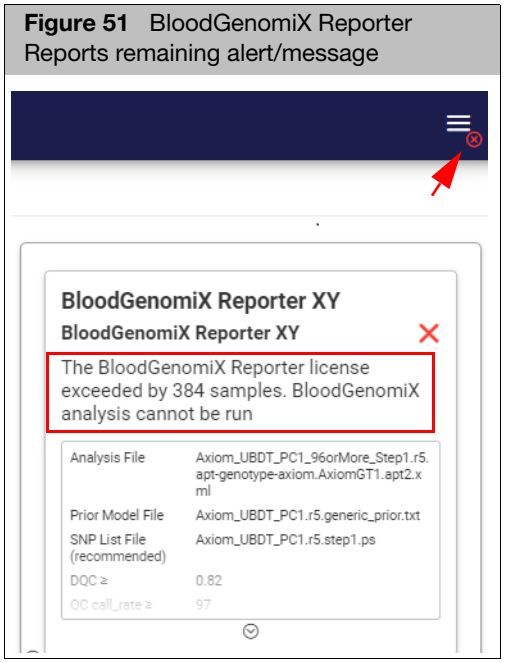
### Extended Blood Typing

| 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<br>NEG<br>NT<br>NOT_PRESENT<br>HEM<br>HET<br>HOM<br>PARTIAL<br>WEAK | antigen type | PASS/FAIL |

## Purchasing reports

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

**Figure 51** BloodGenomiX Reporter Reports remaining alert/message

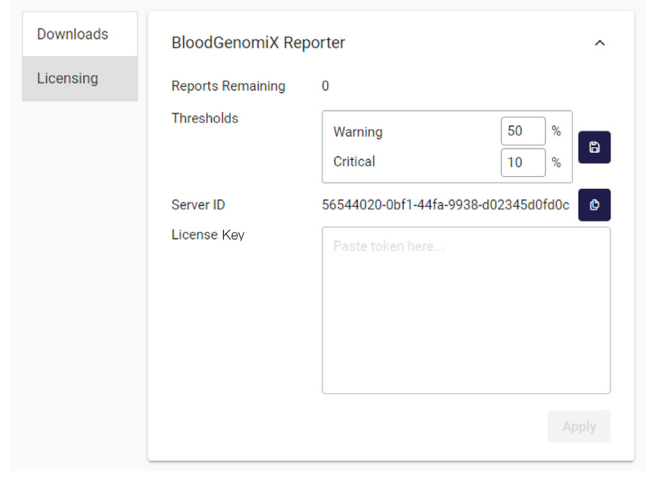



1. Contact your Thermo Fisher Sales Representative for a BloodGenomiX Reporter quote.
2. Click on  (far right), then click **Configurations**.

   The **Batch Download Options** window appears.
3. Click **Licensing**.

   The **BloodGenomiX Reporter** window appears. (Figure 52)

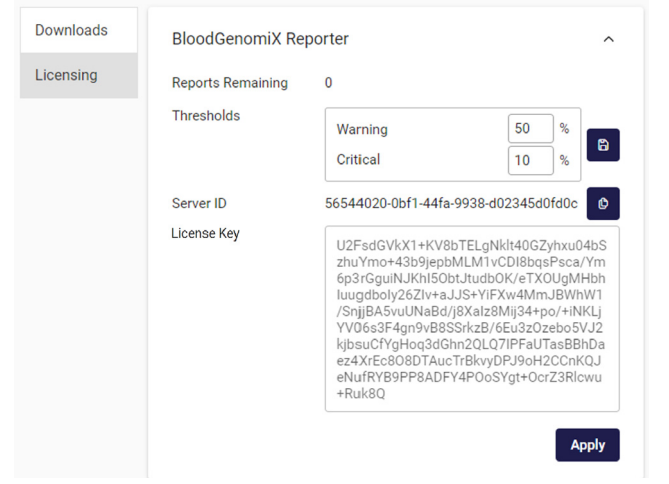
**Figure 52** BloodGenomiX Reporter window




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** BloodGenomiX Reporter window with License Key string




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 equal to or greater than the number of samples in the batch that are to be processed. If not, BloodGenomiX Reporter will not run.

B

# 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

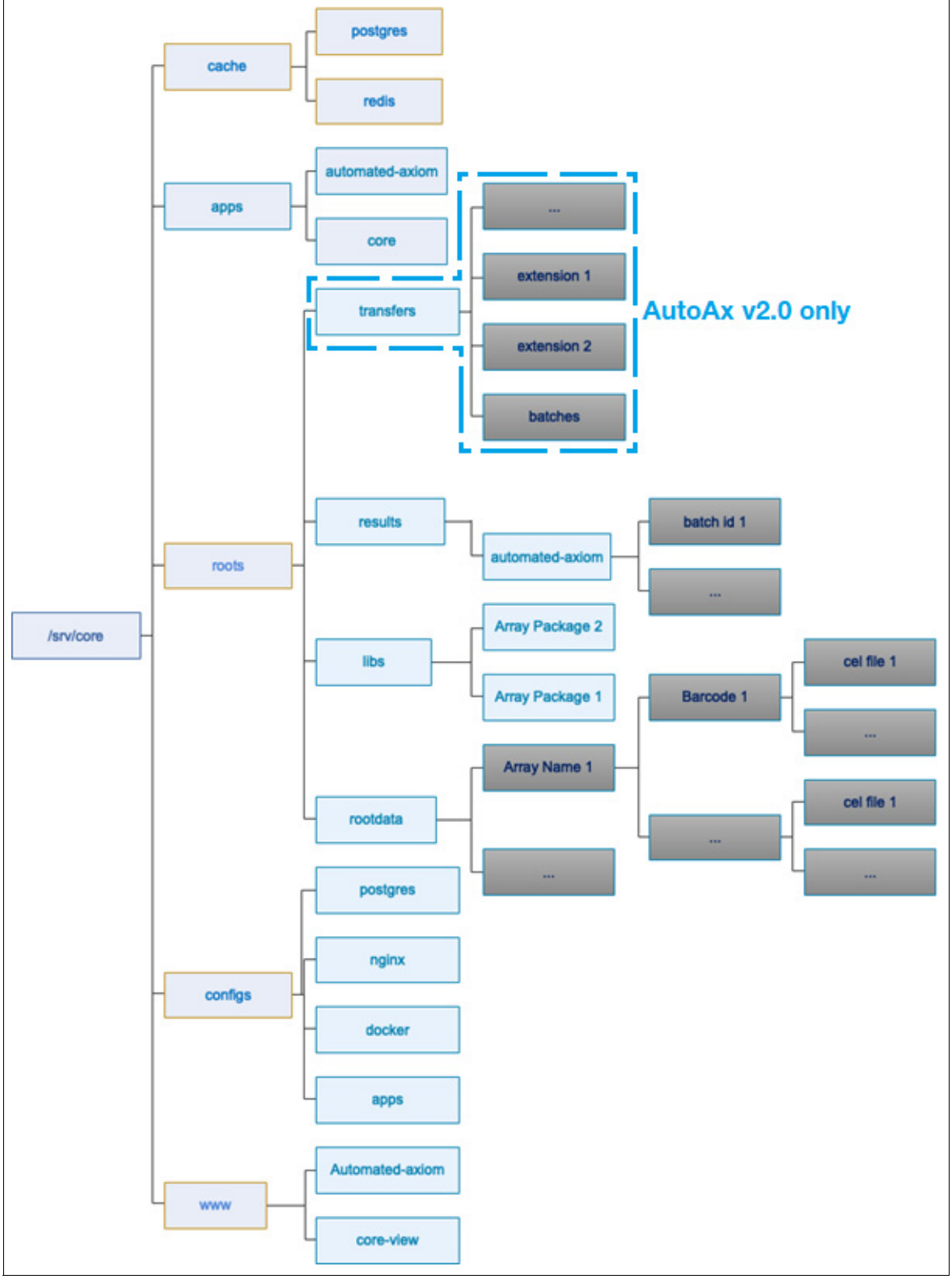
- AutoAx v2.0 users see "Archiving with AutoAx v2.0" on page 56.
- AutoAx v1.2 users see "Archiving with AutoAx v1.2" on page 60.

## 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.

**Figure 54** AutoAx directory (workflow) tree - **Directories in gray 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 **batchId**, the batch information must be retrieved from the system. The screen examples that follow (Figure 55) show how to retrieve the batchId based on the ID and the display name, which is at the top of the batch information card.

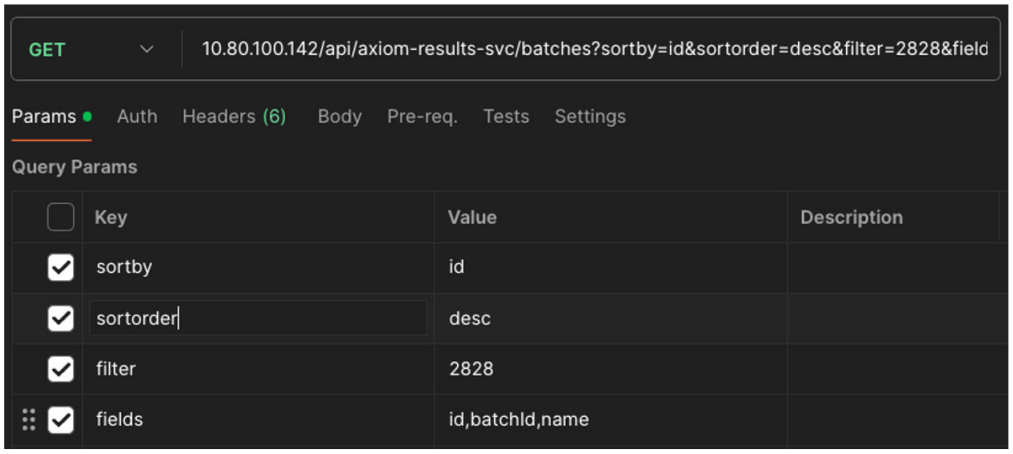
**Note:** Change or remove **sortby** and/or **sortorder** based on your needs. The barcode ending in **2828** is used in the screen examples that follow.

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 batchId based on the ID and the display name, which is at the top of the batch information card.

**Figure 55** Different methods for retrieving, changing, and sorting batches

*Postman Method*




*Curl Method*

```
1. curl --location '10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id%2CbatchId%2Cname'
2.
```

*Powershell Method*

```
1. $response = Invoke-RestMethod '10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id,batchId,name' -Method 'GET' -Headers
$headers
2. $response | ConvertTo-Json
3.
```

*Python Request Method*

```
1. import requests
2. url = "10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id,batchId,name"
3. payload = {}
4. headers = {}
5. response = requests.request("GET", url, headers=headers, data=payload)
6. print(response.text)
7.
```

*wget Method*

```
1. wget --no-check-certificate --quiet \
2.   --method GET \
3.   --timeout=0 \
4.   --header '' \
5.    '10.80.100.142/api/axiom-results-
svc/batches?sortby=id&sortorder=desc&filter=2828&fields=id,batchId,name'
6.
```

## Removing Batches using API

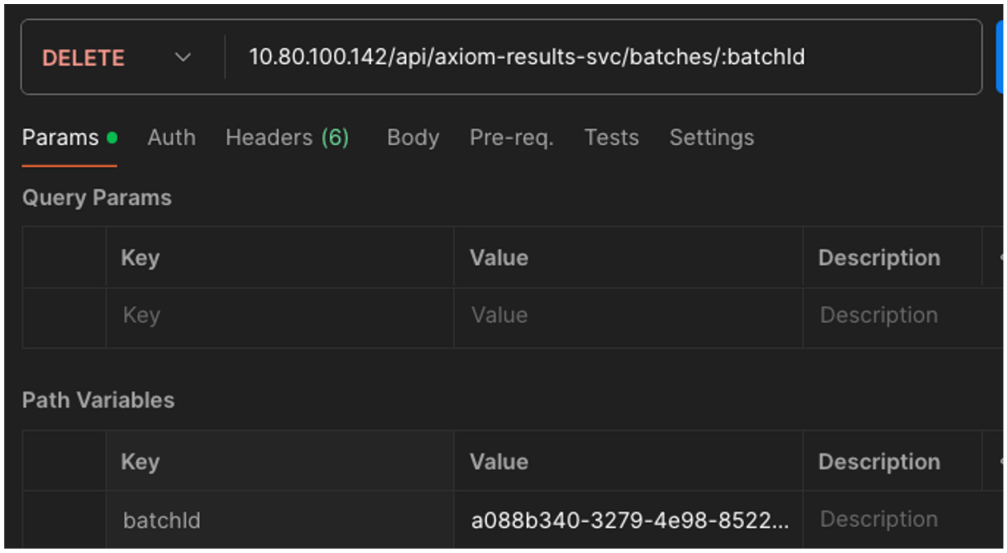
- Internally, all batches are identified and referenced by the **batchId**, not the ID.
- The **batchId** is a **U**niversal **U**nique **I**dentifier (UUID)
- If you want to remove a batch from the system without having to ssh, delete, and refresh the cache, you can use the remove api. This is a two-step process.

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

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

**Figure 56** Different methods for using the remove api call

*Postman Method*




*Curl Method*

```
1. curl --location --request DELETE '10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486'
```

*PowerShell Method*

```
1. $response = Invoke-RestMethod '10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486' -Method 'DELETE' -Headers $headers
2. $response | ConvertTo-Json
3.
```

*Python Request Method*

```
import requests

1. url = "10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486"
2.
3. payload = {}
4. headers = {}
5.
6. response = requests.request("DELETE", url, headers=headers, data=payload)
7.

print(response.text)
```

*wget Method*

```
1. wget --no-check-certificate --quiet \
2.   --method DELETE \
3.   --timeout=0 \
4.   --header '' \
5.    '10.80.100.142/api/axiom-results-svc/batches/a088b340-3279-4e98-8522-8c0e19a46486'
6.
```

## Re-syncing all batches

After the selected batches have been archived off the server, they can be re-synced. Below, examples for each tool will be provided for how to synchronize the system.

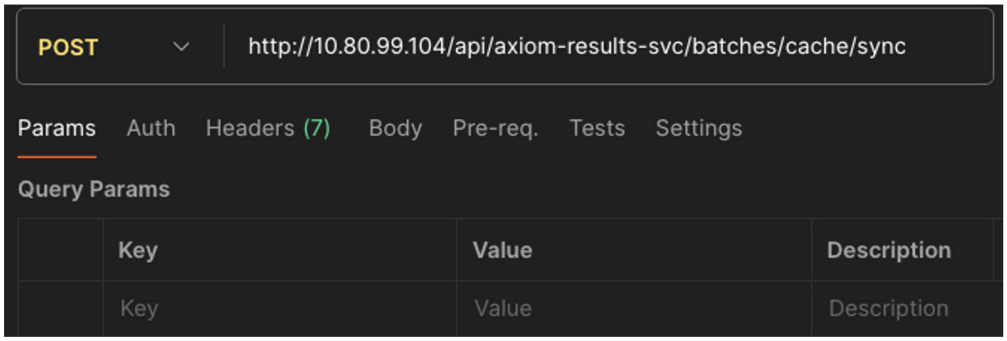
There are many tools/options for performing HTTP endpoint calling. The screen examples in Figure 57 show the most common.

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 Method*




*Curl Method*

```
1. curl --location --request POST '10.80.99.104/api/axiom-results-svc/batches/cache/sync'
2.
```

*Powershell REST method*

```
1. $response = Invoke-RestMethod '10.80.99.104/api/axiom-results-svc/batches/cache/sync' -Method
'POST' -Headers $headers
2. $response | ConvertTo-Json
3.
```

*Python Method*

```
1. import requests
2.
3. url = "10.80.99.104/api/axiom-results-svc/batches/cache/sync"
4.
5. payload = {}
6. headers = {}
7.
8. response = requests.request("POST", url, headers=headers, data=payload)
9.

print(response.text)
```

*wget Method*

```
1. wget --no-check-certificate --quiet \
2.   --method POST \
3.   --timeout=0 \
4.   --header '' \
5.    '10.80.99.104/api/axiom-results-svc/batches/cache/sync'
6.
```

# Archiving with AutoAx v1.2

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

Use care when archiving **/srv/core/roots/rootdata**. Root data is used as input to the analysis workflow processing when creating batches and for the **Reanalysis** feature.

If a batch is queued/status as **Pending** or **Running** 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**

## Removing Batches using API

- Internally, all batches are identified and referenced by the **batchId**, not the ID.
- The **batchId** is a **U**niversal **U**nique **I**dentifier (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 **remove** command line, use the **batchId** from each batch you want to remove. Repeat this process for each batch. This will remove the batch from the server and cache memory.

## Re-syncing all batches

1. 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.
2. In the tab that appears, enter:

   **mutation{**
   **reCacheBatchinfo**
   All caches are refreshed.

# C Extensions

## 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

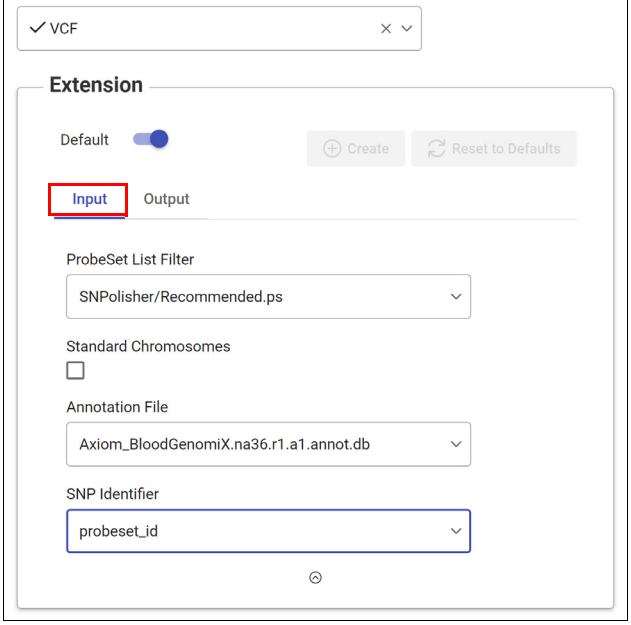
(Figure 58)

**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.

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

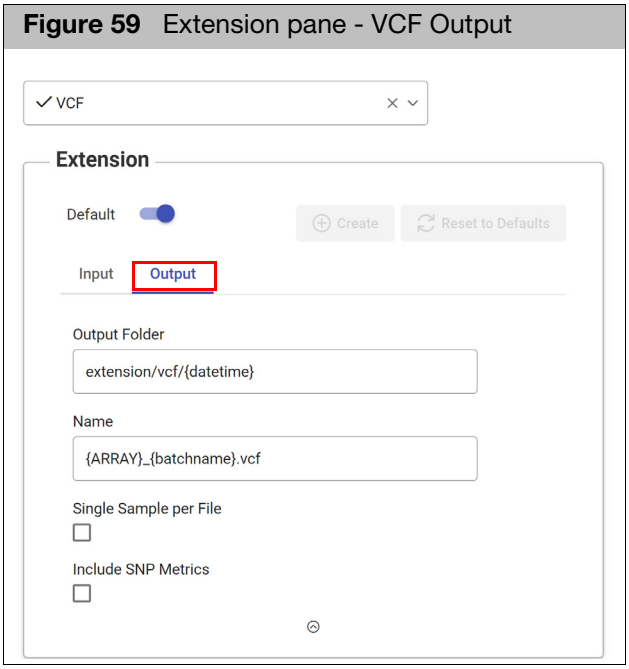
**Figure 58** Extension pane - VCF Input




## Output tab

(Figure 59)

**Figure 59** Extension pane - VCF Output




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

- **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.

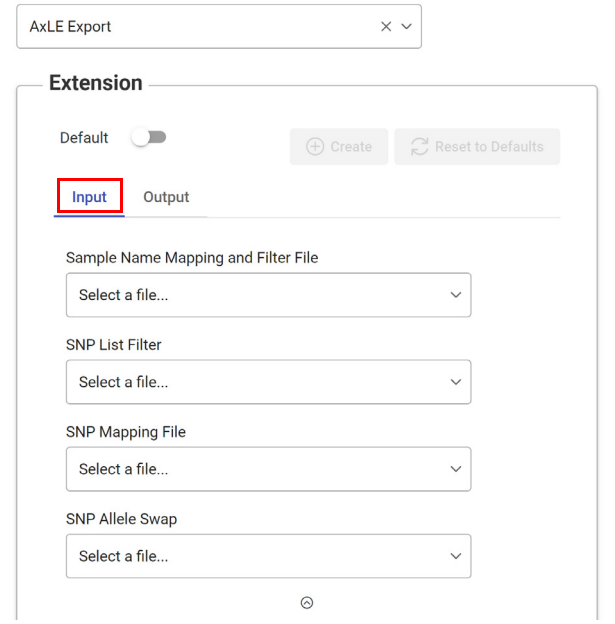
## AxLE extension

### 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 pane - AxLE Export




**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.

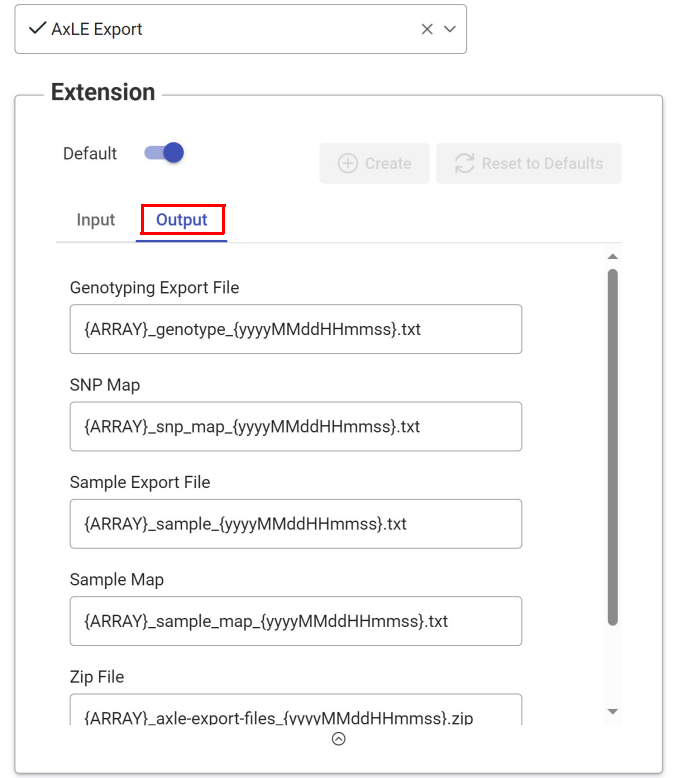
  **Note:** Once a parameter is changed or a file is selected, the **Create** or **Save** and **Reset to Defaults** buttons become enabled.

### Output tab

(Figure 61)

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

**Figure 61** Extension pane - AxLE Export




- **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.

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

## Allele Translation extension

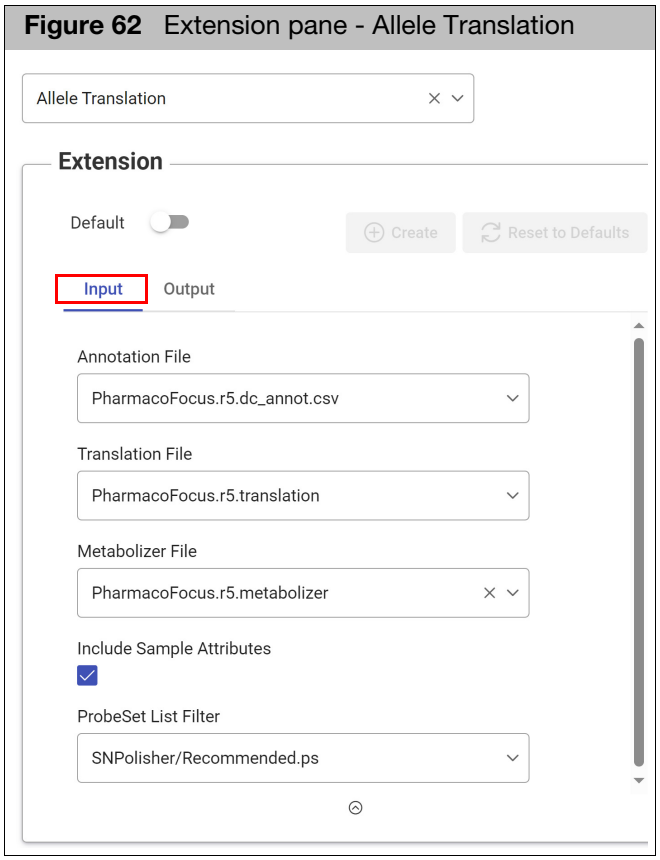
An **Allele Translation** extension appears as an extension option (Figure 62) if the arrays you are using have been enabled. See the *Axiom Analysis Suite User Guide* (P/N MAN0027928) for more information.

### 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 Translation

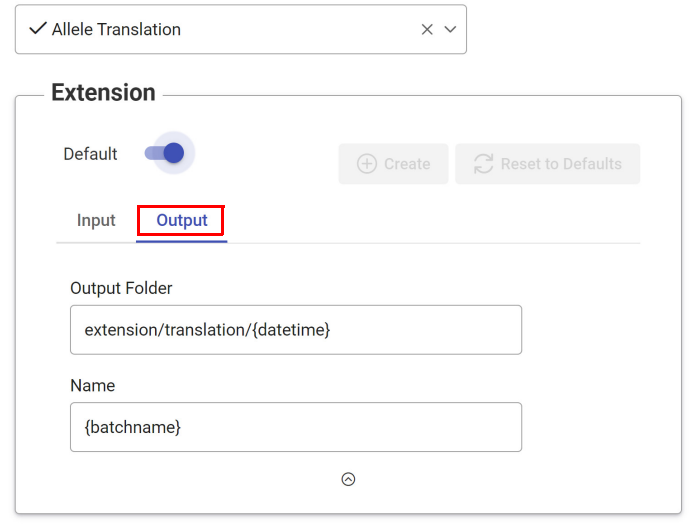



### Output tab

- 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.

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

**Figure 63** Extension pane - Allele Translation



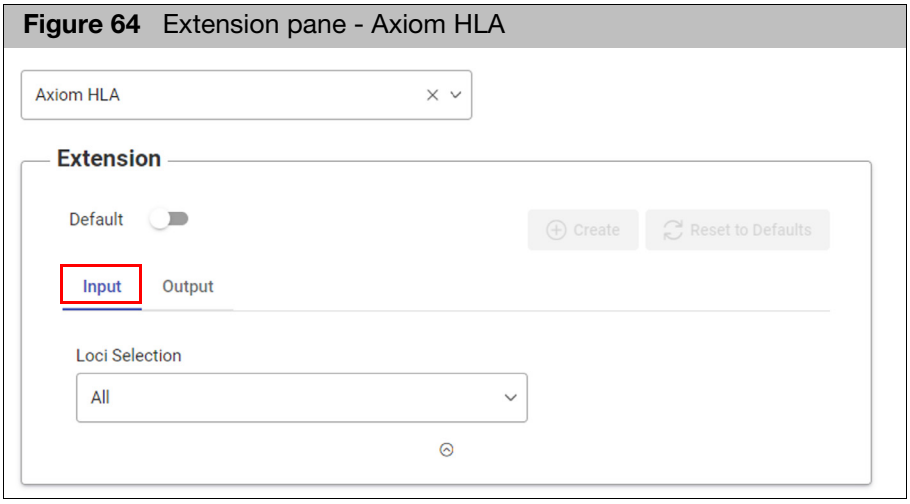

## Axiom HLA extension

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

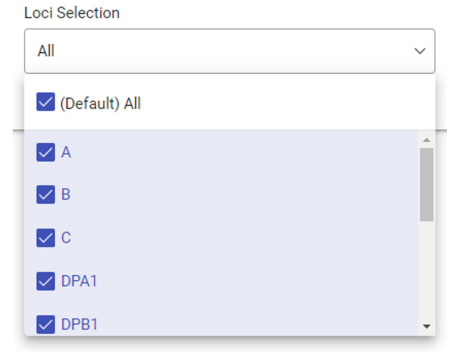



1. Click on the **Loci Selection** drop-down.

   **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.

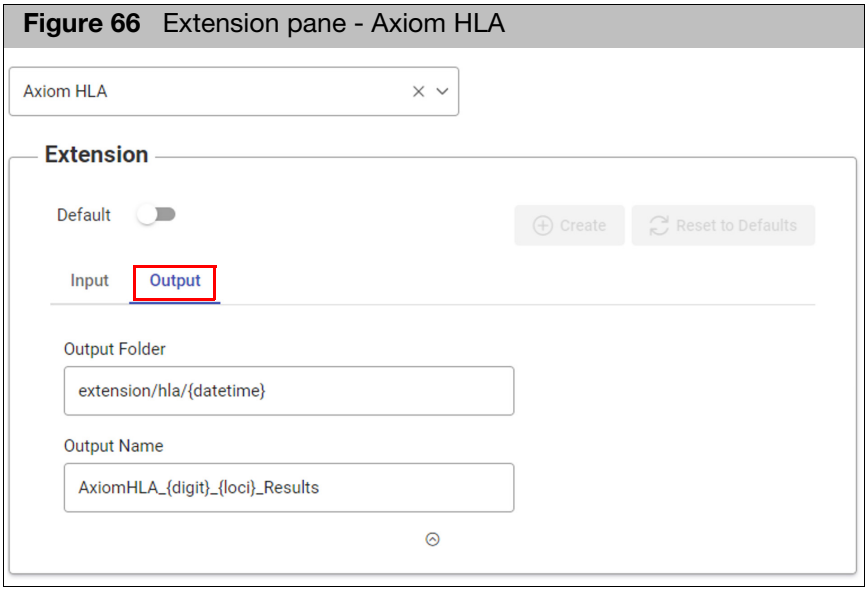
**Figure 65** Loci Selection drop-down




## Output tab

(Figure 66)

**Figure 66** Extension pane - Axiom HLA




- **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.

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

# 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        {
28            "name": "best_practices_for_blood",
29            "internalBatchPath": "BGX",
30            "transfer": true,
31            "destination": "transfers/BGX",
32            "deleteSrc": false,
33            "type": "xfer",
34            "imageTag": "core/core-sh:latest",
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.