

# Axiom™ Long Format Export Tool v1.5

Publication Number 703455 Revision 6

**IMPORTANT!** Axiom Analysis Suite must be installed on your system BEFORE installing and using this tool.

The Axiom Long Format Export Tool is a companion application to the Applied Biosystems™ Axiom Analysis Suite software and has been designed to format genotype data from the Applied Biosystems Axiom platform using the top (TOP) and bottom (BOT) designations based on the polymorphism itself, or the contextual surrounding sequence. It also designates the A/B allele to enable easy correlation of genotype calls made today to legacy data.

This tool exports genotypes for multi nucleotide polymorphisms (MNPs) and indels using an internally developed TOP/BOT assignment and AB naming convention, while enabling AB swaps (with the addition of an input file).

**Note:** Multiallelic markers are reported as no calls.

# Launching the tool

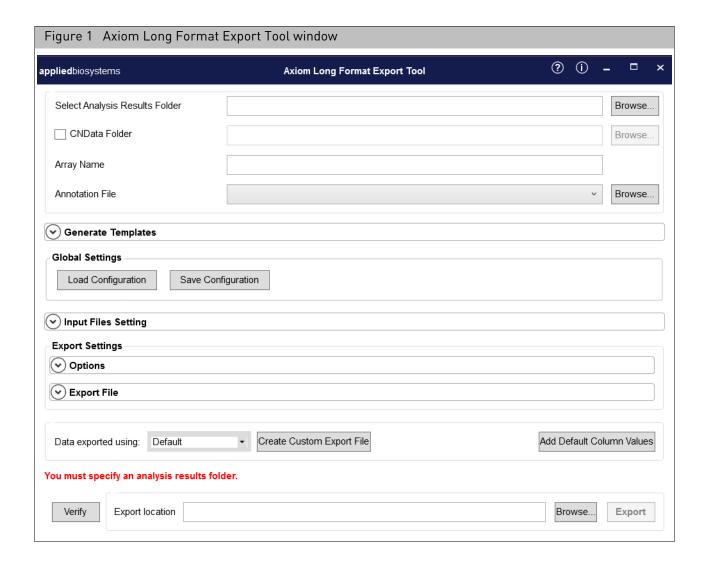
**IMPORTANT!** You must close the batch you want to export from the Axiom Analysis Suite BEFORE launching this tool.

1. Click Start → All Programs → Thermo Fisher Scientific → Axiom Long Format Export Tool.

Alternatively, from the Axiom Analysis Suite application, click the **External Tools** tab, then click on the **Long Format Export Tool** button.

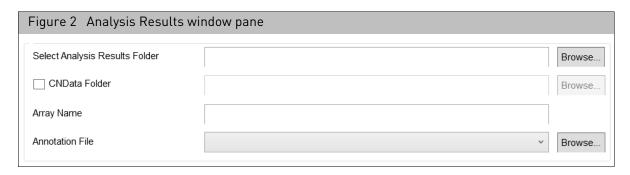
The Axiom Long Format Export Tool window opens. (Figure 1)





## **Analysis information**

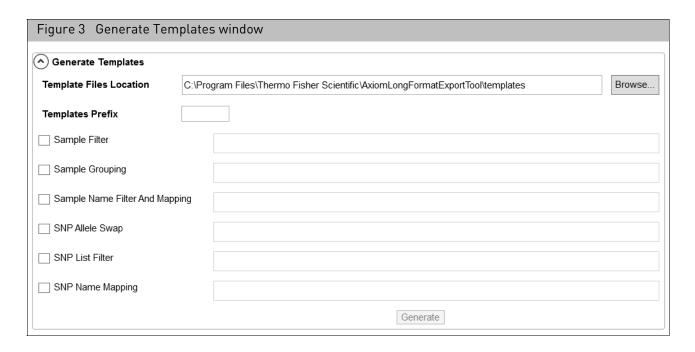
Use this window pane (Figure 2) to set and assign the following:



- Select Analysis Results Folder: Click the Browse... button, then navigate to the Axiom Analysis Suite Results folder. Click Select Folder to populate the Analysis Results window pane.
- CNData Folder: (Optional) Click the check box, then click the Browse... button to
  select the location of the CNData folder. Navigate to your copy number batch,
  then click Select Folder to assign it as your CNV Results Folder. Note: The tool
  performs a check to make sure the same CEL files are used for both genotyping
  and CNV analysis.
- **Array Name**: The Array Name field is auto-detected and populates after selecting the Analysis Results folder.
- Annotation File: The Annotation File field is auto-detected and populates after selecting the Analysis Results folder. Click the drop-down to select a different annotation file (if available). To select an annotation file that is not listed, click on the **Browse...** button, then navigate to your annotation folder.

## Generating templates

1. Click the Generate Templates drop-down arrow to reveal its options. (Figure 3)

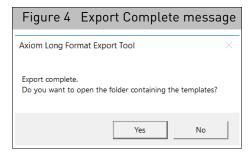


- **Template Files Location:** By default, template files are saved in a sub-folder inside the batch folder. If you want to change the displayed **Template Files Location**, click **Browse...** then navigate to another location.
- Templates Prefix: The default prefix used for template files is the batch name.
   Click inside the Templates Prefix text field to enter a different prefix. Note: If you want to change the auto-populated names, click inside the text field, then enter a different name.
- Sample Filter: Click this check box to place this file is a single column text file with the CEL files in the batch. Use this option if you want to export the CEL files listed in the Sample Filter file.
- Check boxes: Click the check box(es) that correspond with the template(s) you want to generate. For details on each of these options, see "File options" on page 5.

**IMPORTANT!** Sample Grouping cannot be selected and generated at same time as Sample Filter or Sample Name Filter and Mapping.

2. Click the **Generate** button.

After a few moments, a dialog box appears. (Figure 4)



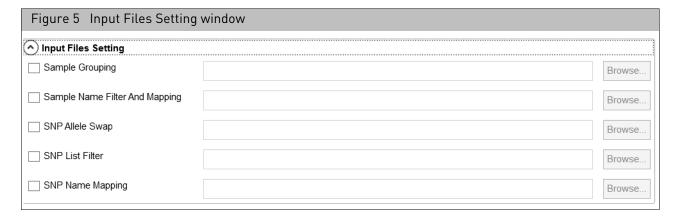
Click Yes.

The generated templates appear.

4. Double-click on the template file you want to open.

# Assigning input files

1. Click the **Input Files Setting** pane's drop-down arrow to reveal its options, as shown in Figure 5.



## File options

- Sample Grouping: This is a two column file with the headers group and cel\_files. This option detects the samples listed in the group column and places them into separate folders and files. CEL files are displayed in the cel\_files column.
- Sample Name Filter and Mapping File: Click this check box to restrict the output to a list of samples and to map them to user-defined Sample Names contained in a single file. The Sample Filter file is also included in this file.
- SNP Allele Swap: Click this check box to specify a file of SNP Allele to user-defined SNP Alleles. Use this option to address discrepancies with legacy data.
   Note: Before using this as an input file, you must know the allele information of the probesets to be swapped.
- SNP List Filter: Click this check box to restrict the output to a list of SNPs (probeset\_id) contained in a file.
- SNP Name Mapping: Click this check box to specify a file of probeset ids to user-defined SNP names.

2. Click a check box.

The **Browse...** button is enabled.

3. Click Browse...

An Explorer window appears.

4. Navigate to the Input file's location, then click **Open**.

The path is now displayed.

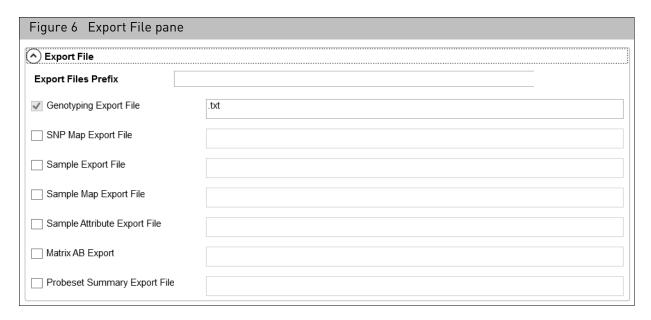
5. Repeat steps 2-4 to add more Input files.

**IMPORTANT!** Sample Grouping input selection cannot be used at the same time as Sample Name Filter and Mapping file.

## Export file options

1. Click the **Export File** pane's drop-down arrow to reveal its options, as shown in Figure 6.

**Note:** The Export Files Prefix window is auto-populated with the analysis batch name. Click inside the Export Files Prefix text field to enter a different prefix.

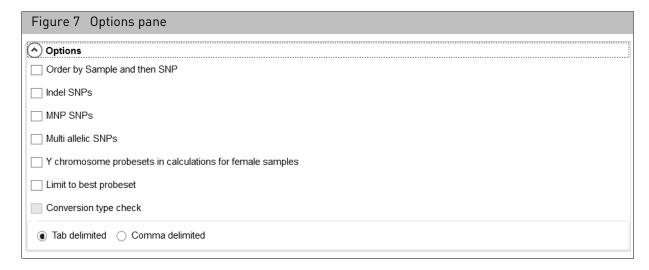


2. Click on the check box(es) adjacent to the Export File type(s) you want to use in your export.

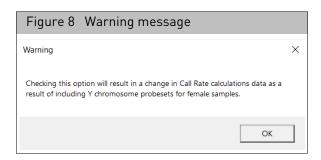
**Note:** The **Genotyping Export File** check box is checked by default and cannot be unchecked.

## **Exporting options**

Click the **Option** pane's drop-down arrow to reveal its options, as shown in Figure 7.



- 1. Click on the appropriate check box(es) to further enhance your Export.
  - Order by sample then SNP
  - Indel SNPs
  - MNP SNPs
  - Multi allellic SNPs: Exported as a No Call (-).
  - Y chromosome probesets in calculations for female samples: Check this
    check box to include probesets on Y chromosome for female samples. A dialog
    box appears. (Figure 8) Click OK to acknowledge it.



- Limit to best probeset: Limits the genotype export results to be only for the best probeset as defined in the SNPolisher\Ps.performance.txt file. Note: If there is no *Best* probeset for a given SNP in the TXT file, then the first probeset of that SNP is exported. This ensures your export is compatible with files exported from the Applied Biosystems™CDCB Export Tool.
- Conversion type check: If checked, then calls are exported as No Calls if the conversion type in the Ps.performance.txt file is not PolyHighResolution,
   NoMinorHom or MonoHighResolution. Note: To enable this check box, the Limit to best probeset check box must be checked.
- Tab delimited/Comma delimited: Click the appropriate radio button to select the type of text file you want export.

## Global settings

Use this pane (Figure 9) to save your new configuration or load a previously saved one.



**Note:** Any Input file(s), Export option(s), Export file option(s), or file selection can be saved using the Global Settings feature.

**Note:** If you want your export file(s) to have a custom name, remove the prefix from the prefix field, rename the file(s), then click **Save Configuration**.

# Saving a new configuration

1. After configuring your Input file(s), Export option(s), and Export file option(s), click **Save Configuration**.

An Explorer window appears.

2. Enter a name for your settings in n the File name text field, then click **Save**.

Your new Settings name is now displayed next to the **Save Configuration** button.

**Note:** The Annotation file is also saved as part of your newly saved configuration.

# Loading a saved configuration

1. Click Load Configuration.

An Explorer window appears.

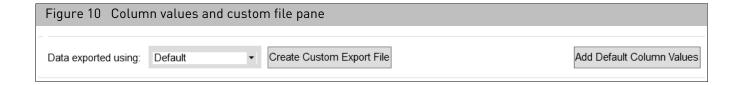
- 2. Navigate to your saved configuration file.
- 3. Single-click on it, then click **Open** or double-click on the file.

The Settings name is now displayed next to the **Save Configuration** button.

**Note:** An analysis batch must be loaded first prior to loading your saved configuration.

# Adding columns and creating custom files (optional)

Use this pane to add columns, create a custom file, and export data using the default columns or all available columns. (Figure 10)



# Changing the number of columns (optional)

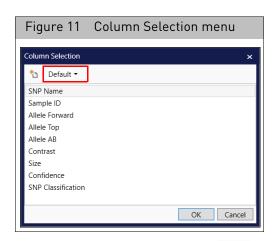
1. By default, data is exported using factory default columns. To use all available columns in your exported data, click the **Data exported using** drop-down arrow, then select **All Columns**.

# Creating a custom column set (optional)

1. Click Create Custom Export File

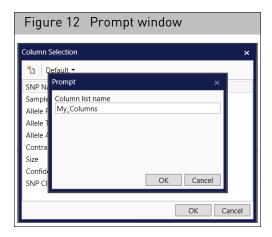
The Column Selection window (Figure 11) appears displaying the default columns. See "Default columns" on page 14 for their definitions.

**Note:** To view all columns, click the **Default** drop-down, then select **All Columns**. To view Log2Ratio and BAF output, make sure to add the columns in your newly created export.

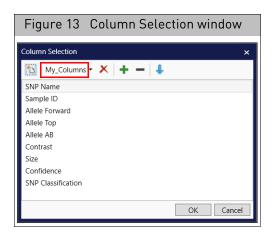


2. Click the create new column icon

A dialog box appears. (Figure 12)



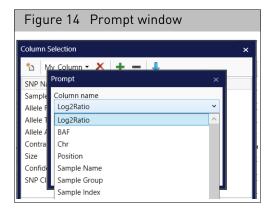
Use the text field to enter a column name, then click OK.Your newly entered column name now appears. (Figure 13)



## Adding columns

1. Click the **+** button.

A dialog box appears. (Figure 14)



2. Click the Column name drop-down menu, then click to select the column you want to add to your column set.

The selected column is now added to your custom set list.

3. Click **OK**, or repeat steps 1-2 to add more columns.

# Rearranging columns

- From your Column Selection window, click to highlight the column you want to move.
- 2. Click the appropriate arrow † button.

The column is now moved to its new position.

Repeat steps 1-2 to move additional columns.

## Removing columns

- From your Column Selection window, click to highlight the column you want to remove.
- 2. Click the **b**utton.

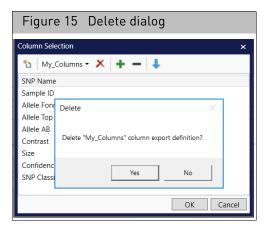
The column is now removed.

Repeat steps 1-2 to remove additional columns from your custom set list.

# Deleting a custom column set

1. From the custom column set's Column Selection window, click the button.

A dialog box appears. (Figure 15)



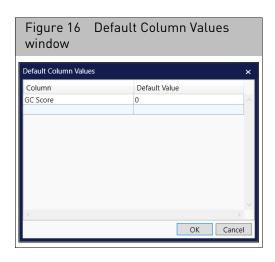
2. Click Yes.

Your custom column set is now removed.

# Adding default column values

1. Click Add Default Column Values .

The Default Column Values window appears. (Figure 16) **Note:** You can assign a value for any of the designated column listed.



- 2. Enter a permissible column name, then enter a new default value or leave this value field blank.
- 3. Click inside the empty row (below your latest entry) to enter another column and new default value. Repeat this step, as needed.

Only the following default column values can be changed:

GC Score, GT Score, Cluster Sep, Theta, R, X Raw, Y Raw, CNV Value, CNV Confidence, 0/1, NormID, and GenTrain Score.

All other default columns have set values and cannot be changed.

### 4. Click OK.

Your generated export file will now contains the values you assigned.

**Note:** An error message appears if the software does not recognize a column name you entered or if it is not one of the permissible column names listed above.

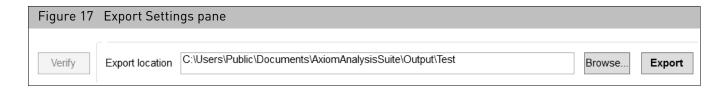
# Removing default column values

1. To remove a row from the Default Column Value window (Figure 16), completely empty/delete both the row's text fields, then click **OK**.

## **Exporting**

**Note:** If you want copy number variation data included in the output file and have NOT run a Discovery or Fixed Regions workflow, use the Axiom CNV Summary Tool prior to the Axiom Long Format Export Tool. The Axiom CNV Summary Tool will calculate Log2Ratio and B allele frequencies (BAF).

By default, the Export location is set to your Analysis Results Folder location (path). (Figure 17) If you want to change this path, go to "Changing the default export location".



If the software detects any error(s), a red warning message will appear in the lower section of the window. Example: **You must specify an analysis results folder**.

Before you can perform an export, you must address the detected error(s), then click the **Verify** button to perform a final check.

- If there are no reported error(s), click Export.
   After several minutes, the message Export complete appears.
- 2. Click **OK**, then navigate to the export location.
- 3. Locate the report you want to view, then double-click on it to open it.

Axiom results are exported to the Genotypes Export File found in the Analysis Results folder. The output file is a tab delimited text file that contains a Header and Data section, as shown in Figure 18. Command or comma delimited line scripts can be used for export.

The Header contains: Axiom Analysis Suite version, date of export, array type (content), Num SNPs, Total SNPs, Num Samples, and Total Samples.

**Note:** Number of SNPs is equal to the number of probesets on the array. The default columns for the Data section are SNP Name, Sample ID, Forward, TOP and AB allele calls, Contrast, Size, Confidence, and SNP Classification.

# Changing the default export location

1. Click the **Browse...** button.

An Explorer window appears.

2. Navigate to another location, the click the **Select Folder** button.

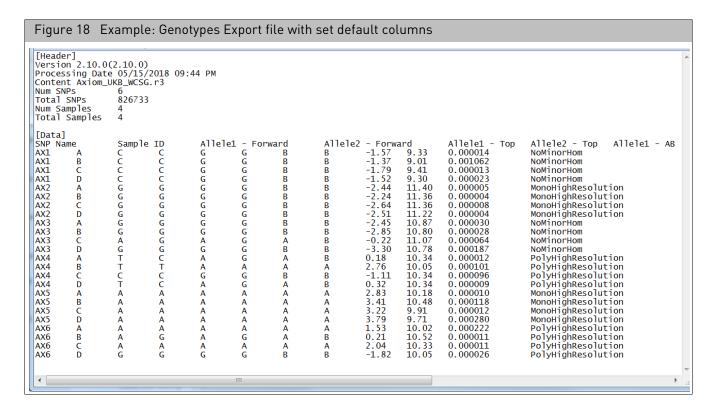
Your new location (path) is now displayed.

3. Click Export.

After several minutes, a Export complete message appears.

- 4. Click **OK**, then navigate to the export location you reassigned earlier.
- 5. Locate the report you want to view, the double-click on it to open it.

# Example report



## Default columns

Column name	Description	
SNP Name	ProbeSet ID	
Sample ID	Sample name (Either the name of the CEL file or user supplied)	
Allele Forward	Genotype call on the forward strand	
Allele Top	Genotype call on the top strand	
Allele AB	The AB call with respect to the strand designation. <b>Note:</b> This may be different than the AB call from Axiom Analysis Suite.	
Contrast	Contrast value (X axis of the Axiom Analysis Suite cluster plot)	
Size	Size value (Y axis of the Axiom Analysis Suite cluster plot)	
Confidence	Confidence value from the AxiomGT1.confidence.txt file	
SNP Classification	SNP conversion type	

# Optional columns

Column name	Description	
Log2Ratio	The log 2 ratio from the Axiom CNV Summary Tools results or Axiom Analysis Suite CNData folder. (Only available when CNV data exists)	
BAF	The BAF value from the Axiom CNV Summary Tools results or Axiom Analysis Suite CNData folder. (Only available when CNV data exists)	
Chr	Chromosome from the annotation file	
Position	Genomic position from the annotation file	
Sample Name	Name of the sample	
Sample Group	Name of the sample group	
Sample Index	Name of the sample index	
SNP	SNP alleles in the form [Allele1/Allele2]	
Customer Strand	Customer defined strand	
X	Normalized A allele intensity from AxiomGT1summary.txt file	
Υ	Normalized B allele intensity from AxiomGT1summary.txt file	

# SNP map file columns

Column name	Description	
Index	Row number	
Name	Name of the ProbeSet	
Chromosome	Number of the chromosome (Example: 14)	
Position	SNP position	
SNP	SNP alleles in the form [Allele1/Allele2]	

# Sample export file columns

Column name	Description	
DNA_ID	Sample name	
#No_Calls	Number of no calls <sup>1</sup>	
#Calls	Number of calls (AA/AB/BB) <sup>1</sup>	
Call_Rate	[#AA+#AB+#BB] / [#AA+#AB+#BB+#NC] <sup>1</sup>	
A/A Freq	#AA / [#AA+#AB+#BB] <sup>1</sup>	

Column name	Description	
A/B Freq	#AB / (#AA+#AB+#BB)¹	
B/B Freq	#BB / (#AA+#AB+#BB)¹	
Minor_Freq	Min of (2*#AA + #AB) / (2*(#AA+#AB+#BB)) or (1 – this value) <sup>1</sup>	
Name	Sample name	
Gender	Gender	
Plate	Plate barcode	
Well	Well position	

<sup>1</sup> The call value is the converted call value from the Top/Bot calculation (multi-alleles are all No Calls as there is no algorithm to convert to Top/Bot). The ProbeSets used are those filtered by this tool. **Note:** Y chromosome probesets for females are not included in the calculations unless the **Y chromosome probesets in calculations for female samples** check box is checked.

# Sample map file columns

Column name	Description	
Index	Row number	
Name	Name of sample (if supplied)	
ID	CEL file name	
Gender	Sex of sample	
Plate	Plate barcode	
Well	Well position	

# Probeset summary (Locus summary) report columns

Column name	Description	
Row	Numerical order	
Locus_Name	AX ID	
Illumicode	No value	
# of no calls	# of no calls for specific probesets	
# of calls	# calls for the specific probeset (AA/AB/BB) <sup>1</sup>	
Call_Freq	The number of calls for the specific probeset/ the total number of calls possible for that probeset (# of calls / # of samples).	
A/A Freq	#AA / (#AA+#AB+#BB) <sup>1</sup>	

Column name	Description	
A/B Freq	#AB / (#AA+#AB+#BB) <sup>1</sup>	
B/B Freq	#BB / (#AA+#AB+#BB) <sup>1</sup>	
Minor_Freq	minor_major_frequency(num_aa, num_ab, num_bb): num_called = num_aa + num_ab + num_bb a = ((2.0 * num_aa) + num_ab) / (2.0 * num_called) b = 1.0 - a min(a, b), max(a, b)*1	
Het Excess	Heterozygote excess frequency, calculated as (Observed - Expected)/Expected for the heterozygote class. If $f_{AB}$ is the heterozygote frequency observed at a locus, and p and q are the major and minor allele frequencies, then het excess calculation is the following: $(f_{AB} - 2pq)/2pq)$	

<sup>1</sup> The call value is the converted call value from the Top/Bot calculation (multi-alleles are all No Calls as there is no algorithm to convert to Top/Bot). The probesets used are those filtered by this tool. **Note:** Y chromosome probesets for females are not included in the calculations unless the **Y chromosome probesets in calculations for female samples** check box is checked.

## Input template examples

- Sample Grouping (Figure 19)
- Sample Name Filter (Figure 20)
- Sample Name Filter and Mapping (Figure 21)
- SNP Allele Swap (Figure 22)
- SNP List Filter (Figure 23)
- SNP Name Mapping (Figure 24)

Figure	19 Sample Grouping template
group	cel_files
san1	LGEV023619_a550431-4179819-022514-394_F06.CEL
san1	LGEV023720_a550431-4179819-022514-394_G12.CEL
san2	LGEV023724_a550431-4179819-022514-392_D01.CEL
san2	LGEV023783_a550431-4179819-022514-392_B06.CEL
san3	LGEV023866_a550431-4179819-022514-392_C10.CEL
san3	LGEV023898_a550431-4179819-022514-402_H01.CEL
	_
<b>Note:</b> When using a Sample Grouping input file for export, the resulting files are auto-separated into individual folders (according to groups). Each gener-	

ated folder is given a group folder name that contain all associated CEL files.

# Figure 20 Sample Name Filter template cel\_files a550995-4395065-072221-717\_A01.CEL a550995-4395065-072221-717\_A02.CEL a550995-4395065-072221-717\_A03.CEL a550995-4395065-072221-717\_A04.CEL a550995-4395065-072221-717\_A06.CEL a550995-4395065-072221-717\_A07.CEL a550995-4395065-072221-717\_A08.CEL a550995-4395065-072221-717\_A09.CEL a550995-4395065-072221-717\_A09.CEL a550995-4395065-072221-717\_A10.CEL

Figure 21 Sample Name	e Filter and Mapping	) template	
cel_files sampl	e_names Index	SentrixPosition LabReference ID	
a550995-4395065-07222	21-717_A01.CEL	a550995-4395065-072221-717_A01.CEL	1
a550995-4395065-07222	21-717_A02.CEL	a550995-4395065-072221-717_A02.CEL	2
a550995-4395065-07222	21-717_A03.CEL	a550995-4395065-072221-717_A03.CEL	3
a550995-4395065-07222	21-717_A04.CEL	a550995-4395065-072221-717_A04.CEL	4
a550995-4395065-07222	21-717_A05.CEL	a550995-4395065-072221-717_A05.CEL	5
a550995-4395065-07222	21-717_A06.CEL	a550995-4395065-072221-717_A06.CEL	6
a550995-4395065-07222	21-717_A07.CEL	a550995-4395065-072221-717_A07.CEL	7
a550995-4395065-07222	21-717_A08.CEL	a550995-4395065-072221-717_A08.CEL	8
a550995-4395065-07222	21-717_A09.CEL	a550995-4395065-072221-717_A09.CEL	9
a550995-4395065-07222	21-717_A10.CEL	a550995-4395065-072221-717_A10.CEL	10

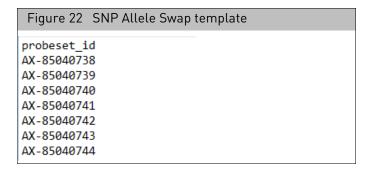


Figure 23 SNP List Filter template
probeset id
AFFX-SP-000001
AFFX-SP-000002
AFFX-SP-000003
AFFX-SP-000004
AFFX-SP-000005
AFFX-SP-000006
AFFX-SP-000007
AFFX-SP-000008
AFFX-SP-000009
AFFX-SP-000010
AFFX-SP-000011
AFFX-SP-000012
AFFX-SP-000013
AFFX-SP-000014
AFFX-SP-000015
AFFX-SP-000016
AFFX-SP-000017
AFFX-SP-000018
AFFX-SP-000019

Figure 24 SNF	Name Mapping template
probeset id	new name
AX-85040738	AX-85040738
AX-85040739	AX-85040739
AX-85040740	AX-85040740
AX-85040741	AX-85040741
AX-85040742	AX-85040742
AX-85040743	AX-85040743
AX-85040744	AX-85040744
AX-85040745	AX-85040745
AX-85040746	AX-85040746
AX-85040747	AX-85040747
AX-85040748	AX-85040748
AX-85040749	AX-85040749
AX-85040750	AX-85040750
AX-85040751	AX-85040751
AX-85040752	AX-85040752
AX-85040753	AX-85040753

Optional: If needed, the columns **Index**, **SentrixPosition**, and **LabReference** can be added to the sample filter renaming template, as shown in Figure 21.

**Note:** The LabReference column maps to the **Name** value in the **sample\_map** file.

# Output examples

- Matrix AB (Figure 25)
- Probeset Summary Export (Figure 26)

Figure 25 Ma	atrix AB							
[Header] Version 2.11.6 Processing Dat	e 12/9/		16					
Content Axiom_Buffalo.r2 Num SNPs 123040								
Total SNPs								
Num Samples	6							
Total Samples	6							
[Data]								
Sample_1		Sampl	Sample_2		e_3	Sample_4	Sample_5	Sample_6
AX-85040738	AA	AA	AA	AA	AA	AA		
AX-85040739	BB	BB	BB	BB	BB	BB		
AX-85040740	BB	BB	BB	BB	BB	BB		
AX-85040741	AB	AB	AB	AB	AB	AB		
AX-85040742	BB		AB	AB	AB	AB		
AX-85040743	AA	AB	AB	AA	AA	AB		
AX-85040744	BB	BB			BB			
AX-85040745	AB	AB	AB	AB	AB	AB		
AX-85040746	AB	AB	AB	AB	AB	AB		
AX-85040747	AB	AB	AB	ВВ	AB	BB		

Row	Locus_Name	Illumicode_Name	#No_Calls	#Calls	Call_Freq	A/A_Freq	A/B_Freq
1	AX-85040738		0	6	1	1	0
2	AX-85040739	9	0	6	1	0	0
3	AX-85040740		0	6	1	0	0
4	AX-85040741	(	0	6	1	0	1
5	AX-85040742	113	1	5	0.833333	8 0	0.8
6	AX-85040743	i	0	6	1	0.5	0.5
7	AX-85040744		3	3	0.5	0	0
8	AX-85040745	4	0	6	1	0	1
9	AX-85040746		0	6	1	0	1
10	AX-85040747	a a	а	6	1	0	0.6

## Related documentation

Document	Publication number	Description
Axiom Analysis Suite User Guide	703307	This user guide provides instructions on using Axiom Analysis Suite. A single-source software package to enable complete genotyping analysis of all Axiom arrays.
Axiom Genotyping Solution Data Analysis Guide	702961	This guide provides information and instructions for analyzing Axiom genotyping array data. It includes the use of Axiom Analysis Suite, Applied Biosystems Microarray Power Tools (formerly APT) and SNPolisher R package to perform quality control analysis (QC) for samples and plates, SNP filtering prior to downstream analysis, and advanced genotyping methods.

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**Note:** For SDSs for reagents and chemicals from other manufacturers, contact the manufacturer.



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#### Revision history: Pub No. 703455

Revision	Date	Description
6	September 2020	Version 1.5 release. Redesigned user interface. Improved Template and Export settings functionality. Custom settings can now be saved.
5	December 2019	Version 1.4 release. Output file names are now user definable. The ability to default the Index and Row value of the SNP and Sample files with index values starting at 1 has been enabled. Added a Matrix AB export report that displays your probesets (as rows) and sample names (as columns). Templates are now auto-generated. A probeset summary report can now be generated and exported. An option to select your own annotation file has been added.
4	August 2018	Version 1.3 release. Added A and B alleles to the SNP column.
3	May 2018	Version 1.2 release. The tool now exports genotypes for multi nucleotide polymorphisms (MNPs) and indels using an internally developed TOP/BOT assignment and AB naming convention. The tool now enables AB swaps with the addition of an input file.
2	August 2017	Version 1.1 release. The tool now formats Axiom genotype data using the top (TOP) and bottom (BOT) designations based on the polymorphism itself, or the contextual surrounding sequence. A/B allele is now designated. The tool can now be accessed within the Axiom Analysis Suite software, via the External Tools tab.
1	May 2017	Initial release

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