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Axiom™ Long Format Export Tool v1.3

Publication Number 703455 **Revision** 4

IMPORTANT! Axiom Analysis Suite must be installed on your system before installing and using the Axiom Long Format Export 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 Axiom genotype data 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 prior research.

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: Multi-allelic markers are reported as no calls.

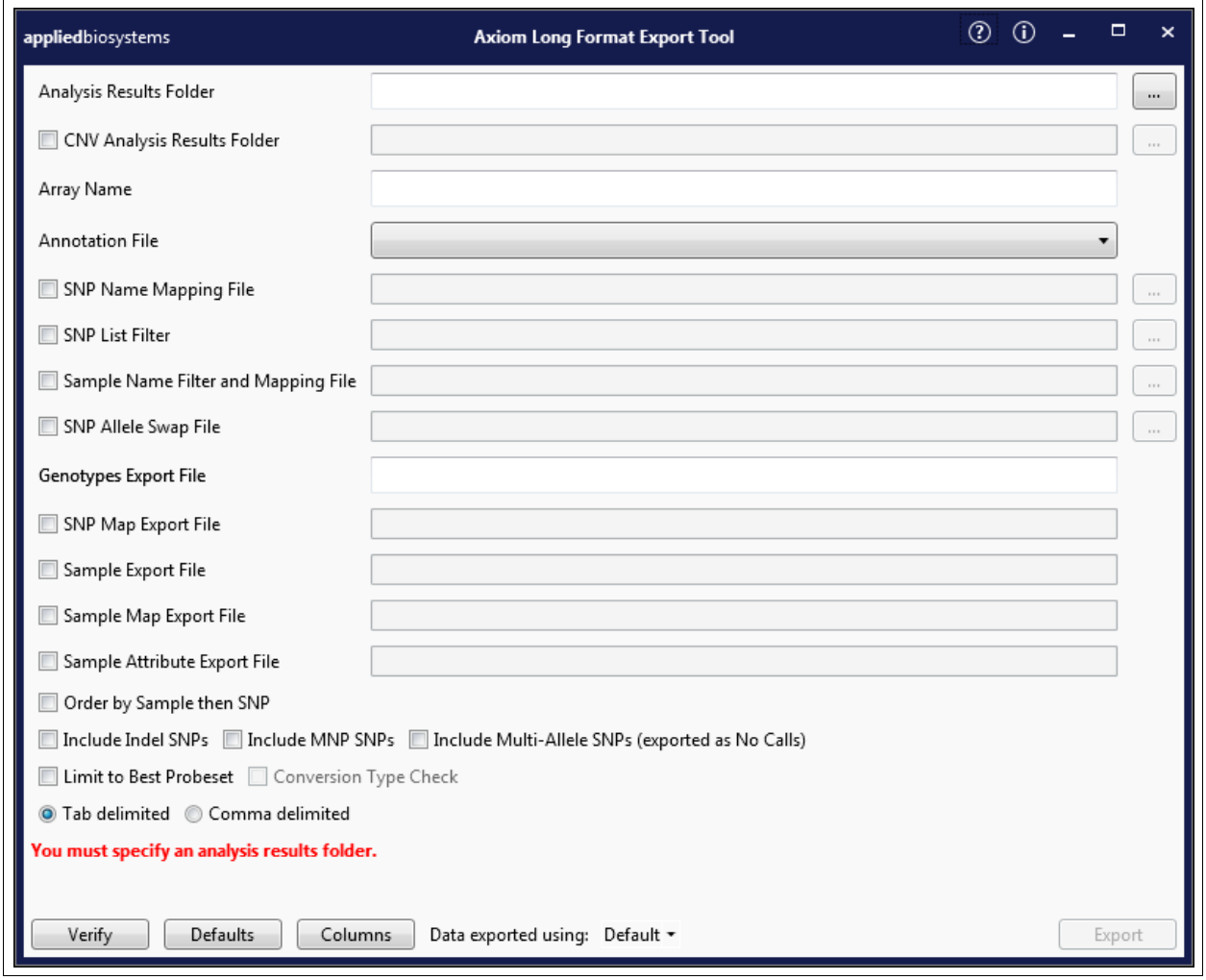
Launching the tool

1. Click **Start** → **All Programs** → **Thermo Fisher Scientific** → **Axiom Analysis Suite** → **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)

Figure 1 Axiom Long Format Export Tool window



Using the tool

IMPORTANT! All data to be exported and used with this tool must reside on a local drive.

1. **Analysis Results**: Click the ... button, and navigate to the Axiom Analysis Suite Results folder. Click the folder name to select.
2. **CNV Analysis Results Folder** (Optional): Click the check box, then click the ... button to select the location of the CNV (log2/baf) results. This can either be the folder containing the Axiom CNV Summary Tools results (same folder as the Analysis Results Folder), or the Axiom Analysis Suite 4.0 software's CNV results folder. This tool auto-checks that the same CEL files are used for both genotyping and CNV analysis.
3. **Array Name**: The Array Name field is auto-detected and populates after selecting the Analysis Results folder.
4. **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).
5. **SNP Name Mapping File** (Optional): Specify a file of Axiom SNP names to user-defined SNP names. Click the check box to activate this field, then click the ... button.
   - An Explorer window appears.
   - Navigate to the SNP Filter List location, then click **Open**.
   - The SNP Name Mapping File field is populated.

   Note: The SNP Name Mapping File is used to map an Axiom SNP name to a user defined name. The file is a tab delimited text file with two columns. The first row of the file is the column header where the column labels are "SNP" for the Axiom SNP name and "User" for the user defined name. See Figure 9 on page 12 for a SNP renaming text file example.
6. **SNP List Filter** (Optional): Restrict the output to a list of SNPs (probeset_IDs) contained in a file. Click the check box to activate this field, then click the ... button.
   - An Explorer window appears.
   - Navigate to the SNP Filter List location, then click **Open**.
   - The SNP Filter List field is populated.

   Note: We suggest only exporting the ***recommended*** probesets from the software, as these are the results which are appropriate for use in downstream applications. The recommended probeset list ("Recommended.ps") is found in the SNPolisher subdirectory in your Axiom Analysis Suite results folder. The Recommended.ps file is located in the following Axiom Analysis Suite batch folder:
   C:\Users\Public\Documents\AxiomAnalysisSuite\Output\AnalysisName\SNPPolisher

7. **Sample Name Filter and Mapping File** (Optional): Restrict the output to a list of samples and to map them to user-defined Sample Names contained in a single file. Click the check box to activate this field, then click the ... button.
   - An Explorer window appears.
   - Navigate to the Sample Name and Mapping File location, then click **Open**. See Figure 9 on page 12 for a sample filter renaming text file example.

     The Sample Name Field and Mapping File field is populated.
8. **SNP Allele Swap File** (Optional): Specify a file of SNP Allele to user-defined SNP Alleles. Click the check box to activate this field, then click the ... button.
   - An Explorer window appears.
   - Navigate to the Sample Name and Mapping File location, then click **Open**. See Figure 10 on page 12 for a swap text file example.
   - The SNP Allele Swap File field is populated.

   Note: If you want to change the auto-populated names (generated during steps 9-13), click inside the text field, then enter a different name.
9. **Genotypes Export File**: A default file name (using the array name's date and time stamp) auto-populates.
10. **SNP Map Export** (Optional): Click the check box. The field auto-populates.
11. **Sample Export File** (Optional): Click the check box. The field auto-populates.
12. **Sample Map Export File** (Optional): Click the check box. The field auto-populates.
13. **Sample Attribute Export File** (Optional): Click the check box. The field auto-populates.
14. To further customize your output file, click on the appropriate check box(es).
    - **Order by sample then SNP**
    - **Include Indel SNPs**
    - **Include MNP SNPs**
    - **Include Multi-Allele SNPs (export as No Calls)**
    - **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 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
15. **Tab delimited/Comma delimited**: Click the appropriate radio button to select the type of text file you want export.
16. **Data exported using** (Optional): Click the drop-down arrow to select **All Columns**.

    Note: If any input errors are detected a red warning message is displayed in the lower section of the window. Example: **You must specify an analysis results folder**.

After fixing any issues, click Verify to recheck. If you want copy number variation data included in the output file, 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) and the Analysis Results folder will contain the Axiom GT1.cnv.txt file.

17. (Optional) To change a column's default value, click Defaults. See page 6 for detailed instructions.

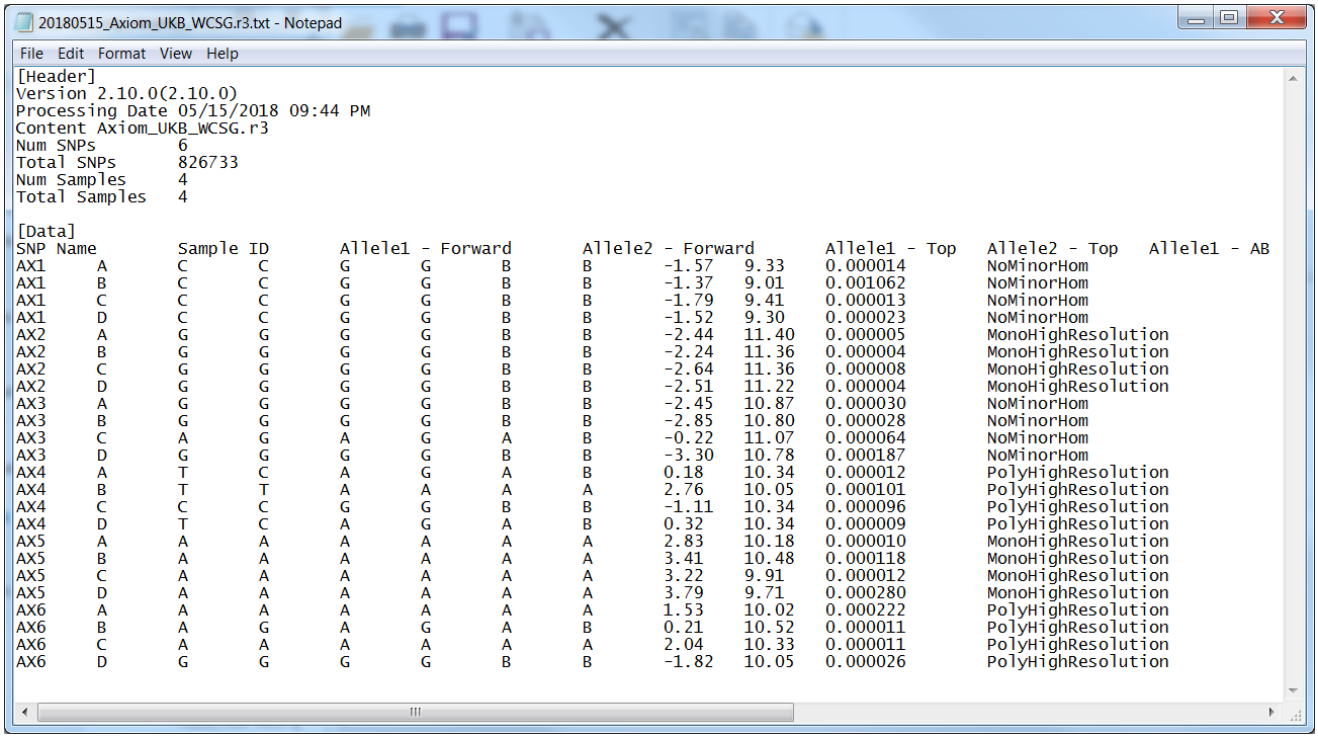
18. After all required fields are populated, click Export.

    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 2.

    The Header contains: Axiom Analysis Suite version, date of export, array type (content), #SNPs, and #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. If the Analysis Results folder contains the AxiomGT1.cnv.txt file generated from the Axiom CNV Summary Tool, the data will also contain the Log2Ratio and BAF values.

Figure 2 Example output file



20180515_Axiom_UKB_WCSG.r3.txt - Notepad

File Edit Format View Help

```
[Header]
Version 2.10.0(2.10.0)
Processing Date 05/15/2018 09:44 PM
Content Axiom_UKB_WCSG.r3
Num SNPs        6
Total SNPs      826733
Num Samples     4
Total Samples   4

[Data]
SNP Name        Sample ID       Allele1 - Forward       Allele2 - Forward       Allele1 - Top   Allele2 - Top   Allele1 - AB
AX1     A       C       C       G       G       B       B       -1.57   9.33    0.000014        NoMinorHom
AX1     B       C       C       G       G       B       B       -1.37   9.01    0.001062        NoMinorHom
AX1     C       C       C       G       G       B       B       -1.79   9.41    0.000013        NoMinorHom
AX1     D       C       C       G       G       B       B       -1.52   9.30    0.000023        NoMinorHom
AX2     A       G       G       G       G       B       B       -2.44   11.40   0.000005        MonoHighResolution
AX2     B       G       G       G       G       B       B       -2.24   11.36   0.000004        MonoHighResolution
AX2     C       G       G       G       G       B       B       -2.64   11.36   0.000008        MonoHighResolution
AX2     D       G       G       G       G       B       B       -2.51   11.22   0.000004        MonoHighResolution
AX3     A       G       G       G       G       B       B       -2.45   10.87   0.000030        NoMinorHom
AX3     B       G       G       G       G       B       B       -2.85   10.80   0.000028        NoMinorHom
AX3     C       A       G       A       G       A       B       -0.22   11.07   0.000064        NoMinorHom
AX3     D       G       G       G       G       B       B       -3.30   10.78   0.000187        NoMinorHom
AX4     A       T       C       A       G       A       B       0.18    10.34   0.000012        PolyHighResolution
AX4     B       T       T       A       A       A       A       2.76    10.05   0.000101        PolyHighResolution
AX4     C       C       C       G       G       B       B       -1.11   10.34   0.000096        PolyHighResolution
AX4     D       T       C       A       G       A       B       0.32    10.34   0.000009        PolyHighResolution
AX5     A       A       A       A       A       A       A       2.83    10.18   0.000010        MonoHighResolution
AX5     B       A       A       A       A       A       A       3.41    10.48   0.000118        MonoHighResolution
AX5     C       A       A       A       A       A       A       3.22    9.91    0.000012        MonoHighResolution
AX5     D       A       A       A       A       A       A       3.79    9.71    0.000280        MonoHighResolution
AX6     A       A       A       A       A       A       A       1.53    10.02   0.000222        PolyHighResolution
AX6     B       A       G       A       G       A       B       0.21    10.52   0.000011        PolyHighResolution
AX6     C       A       A       A       A       A       A       2.04    10.33   0.000011        PolyHighResolution
AX6     D       G       G       G       G       B       B       -1.82   10.05   0.000026        PolyHighResolution
```

Default column values

You can change a column's default value, but only to the columns listed below:

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

Note: All other columns have preset values and cannot be changed.

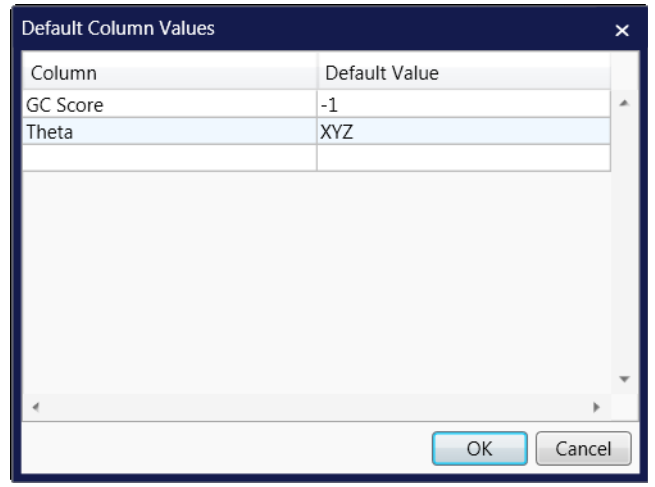
Changing a default column value

1. Click Defaults.

   The Default Column Values window appears. (Figure 3)

2. Enter a permissible column name, then enter a new default value or leave this value field blank.

Figure 3 Default Column Values window



3. Click inside the empty row (below your latest entry) to enter another column and new default value. Repeat this step, as needed.
4. Click **OK**.

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

   To remove a row from this window, completely delete both its text fields, then click **OK**.

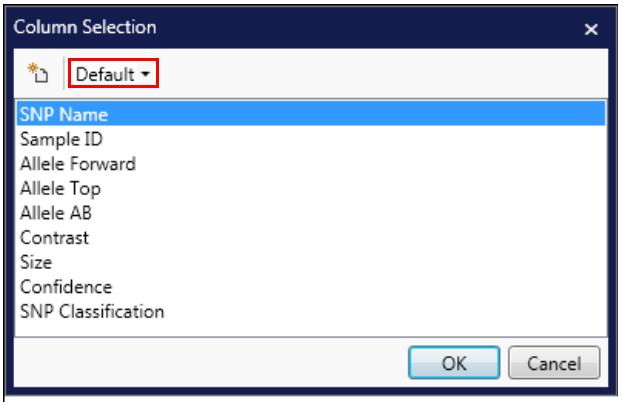
Creating a custom column set

1. Click Columns .

   The Column Selection window (Figure 4) appears displaying the default columns. See "Default columns" on page 10 for their definitions.

   Note: To view all columns, click the **Default** drop-down, then select **All Columns**.

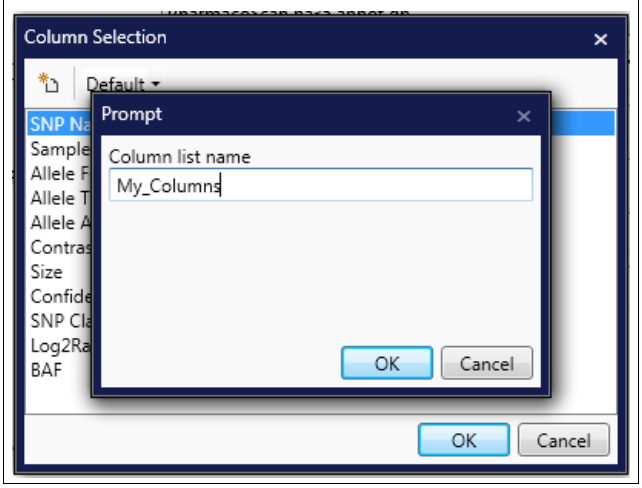
Figure 4 Column Selection menu



2. Click the create new column icon .

   The Prompt window appears. (Figure 5)

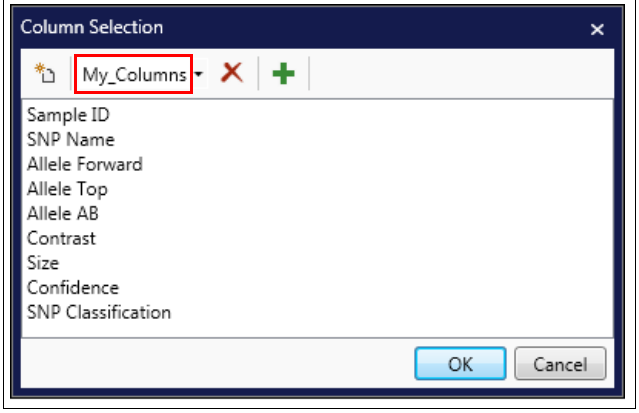
Figure 5 Prompt window



3. Use the text field to enter a column name, then click **OK**.

   Your newly entered column name now appears. (Figure 6)

Figure 6 Column Selection window

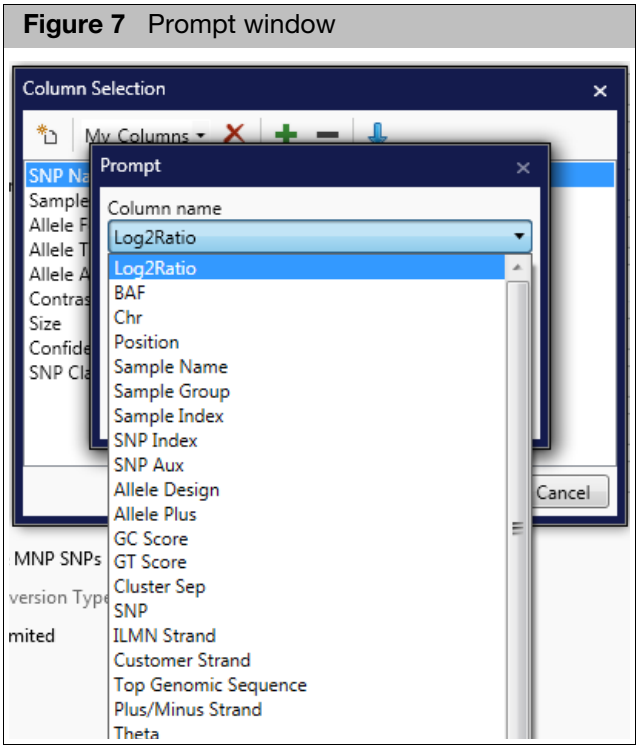


Adding columns

1. Click ✚.

   The Prompt window appears. (Figure 7)

Figure 7 Prompt window



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

1. 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

1. From your Column Selection window, click to highlight the column you want to remove.
2. Click .

   The column is now removed.

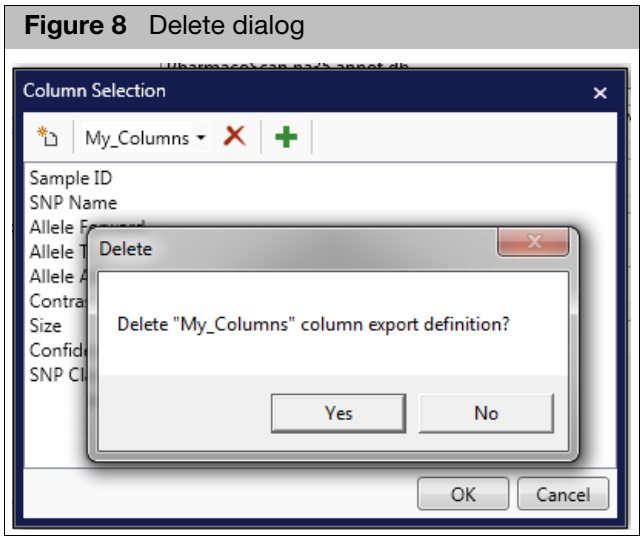
   Repeat steps 1-2 to remove additional columns from your custom set list.

Deleting a custom column set

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

   A Delete dialog window appears.

Figure 8 Delete dialog



2. Click **Yes**.

   Your custom column set is now removed.

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	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 Classification

Optional columns

Column name	Description
Log2Ratio	The log 2 ratio from the Axiom CNV Summary Tools results or Axiom Analysis Suite 4.0 software CNV results folder. (Only available when CNV data exists)
BAF	The BAF value from the Axiom CNV Summary Tools results or Axiom Analysis Suite 4.0 software CNV results 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
Y	Normalized B allele intensity from AxiomGT1summary.txt file

SNP map file columns

Column name	Description
Index	ProbeSet ID
Name	Name of the ProbeSet
Chromosome	Number of the chromosome (Example: 14)
Position	SNP position
SNP	SNP alleles in the form [Allelel1/Allele2]

Sample export file columns

Column name	Description
DNA_ID	Sample name
#No_Calls	Number of no calls[1]
#Calls	Number of calls (AA/AB/BB)[1]
Call_Rate	(#AA+#AB+#BB) / (#AA+#AB+#BB+#NC)[1]
A/A Freq	#AA / (#AA+#AB+#BB)[1]
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)[1]
Name	Sample name
Gender	Gender
Plate	Plate barcode
Well	Well position

[1] 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.

Example input templates

Figure 9 Example SNP renaming template

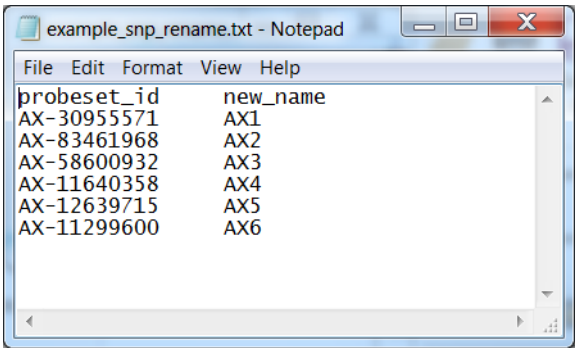
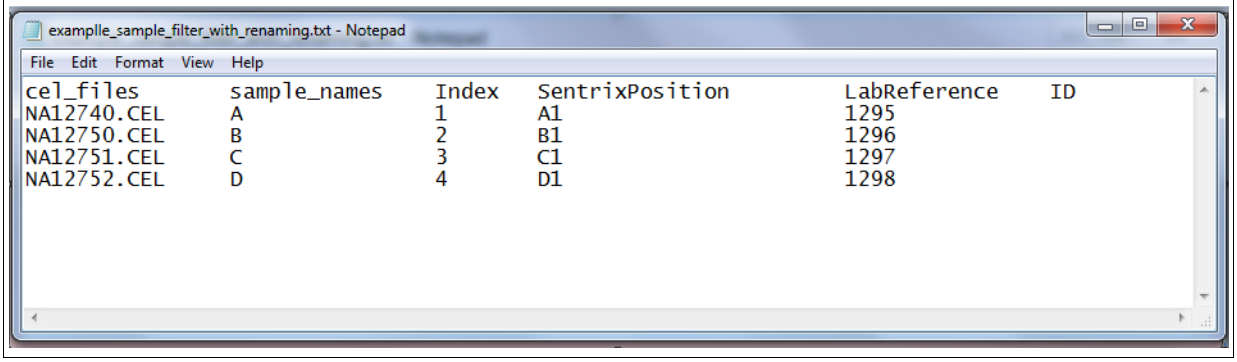


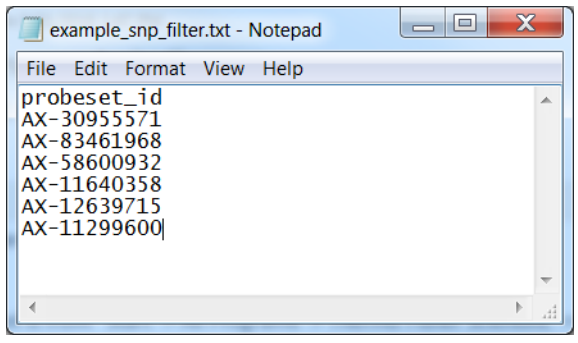
Figure 10 Example sample filter renaming template



Optional: If needed, the columns **Index**, **SentrixPosition**, and **LabReference** can be added to the sample filter renaming template, as shown in Figure 10.

Note: The LabReference column maps to the **Name** value in the **sample_map** file.

Figure 11 Example SNP swap and SNP filter template



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.

Affymetrix, Inc.
3450 Central Expressway
Santa Clara, CA 95051

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Revision	Date	Description
4	August 2018	v1.3 release
3	May 2018	v1.2 release
2	August 2017	v1.1 release
1	May 2017	Initial release

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