

# Axiom™ Analysis Suite (AxAS) v5.3

## APT version

- AxAS v5.3 is associated with APT 2.11.8

## New analysis features and changes

- The new Multiplate Normalization feature improves results for some biallelic probesets that have a non-recommended **ConversionType**.
- Dual Workflow (when available) auto-populates and uses the **recommended.ps** file from the original batch as the SNP List File.
- The **PSMetrics Results.txt** file in the SNPolisher folder is no longer removed after a genotyping analysis successfully completes.
- The Sample QC engine has been updated. In some cases, metrics like **DishQC** located in a batch's "**QC\Geno-qcResults.txt**" file may contain slightly different values. Well position and the plate barcode for each sample in this file are now displayed.
- Copy number reference creation workflows now properly handle probesets with no annotated genomic position. Copy number analysis using a new reference has also been improved for arrays with these probesets.
- The log2 transformation function is now standardized.
- The artifact reduction parameter is now tuned to the specific GeneTitan camera type.
- The **Dashboard** now supports opening an Initial Copy Number Reference Creation analysis batch for review even if the batch fails QC.

## Batch viewer updates

- The splitting of a VCF genotype export by sample has been improved when using larger batches. For example, using a 1,132 **Axiom\_PMRA** sample export produced a total file count of 2.25TB.
- The Whole Genome View for Copy Number Discovery batches now has better memory handling for larger batches.
- Whole Genome View for Copy Number Discovery histograms now use all samples in Sample Table to compute percentages. **Note:** The OMIM track is only displayed if annotations exist.
- The word **gender** has been replaced with the word **sex** in the viewer, however the files inside the batch folder may still use the word **gender**.
- Viewing Plate QC Summary details has been improved on Windows 11 systems.
- The issue with Import Sample Attributes reporting an *Object reference not set to instance of object* message has been resolved.
- The CN Region Plot now has better handling of sample selection and drawing of the dashed line.

## Known issues and limitations

- When regenerating SNP Metrics or running OTV Caller, AxAS may freeze while the **Please Wait...** message is displayed. To remedy this, close and re-launch the Viewer. After the Viewer opens, regenerate your SNP Metrics or run the OTV Caller again.
- If the AxAS software window size is not maximized or too small, the **Preferences** tab may not display all available options.
- The new **Rare Het Adjustment** and **Multiplate Normalization** genotyping options are not available for custom analysis settings that were created in previous AxAS versions that did not feature these options.
- **Percent Concordance** is shown at full precision, 5 decimal places and not the default of 3.
- In certain Windows 10 environments, the **Export IGV | Output File Prefix** field may be partially hidden after sample attributes are loaded.
- Known behaviors when using the General Analysis **Inbred File | Value** option:
  - Inbred file is still accepted even if it contains an invalid value.
  - If a Value and a File are supplied for Inbred, the File is used even if the **Value** checkbox was checked.
  - If the **Inbred** checkbox was checked prior to restoring an Analysis configuration, it remains checked.
- Batches that contain special characters (e.g. , or - ) in their names will error out when attempting to run **Regenerate SNP Metrics**.
- The Batch Viewer may crash while analyzing selected samples from batches that were generated using AxAS versions earlier than v5.0.