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Redefine what's possible with the Axiom® Genotyping Solution

From discovery to translation on a single platform

The Axiom® Genotyping Solution enables enhanced genotyping studies to accelerate your research from discovery to translation on a single platform—quickly, easily, and cost-effectively.

The Axiom Solution has content covering more ethnic populations than any other technology to maximize study power in diverse and admixed cohorts. It is the only genotyping solution that offers a choice of 11 million wet lab-validated and fully-annotated markers so that any investigator can easily and cost-effectively create optimized array designs for their application.

Applications supported by the Axiom Genotyping Solution

Human population genetics

- Variant detection and validation
- Evolutionary biology

Discovery research in human health and disease

- Genome-wide association
- Replication and fine mapping studies
- Exome analysis
- Candidate gene and pathway studies
- Pharmacogenomics

Translational research

- Biomarker identification and validation

Accelerating your research

Ready to use and optimized whole-genome and targeted arrays

- Efficient pre-designed arrays optimized for:
  - Diverse populations
  - Whole-genome and targeted applications

Flexible, innovative, and simplified custom design pipeline

- Axiom® myDesign™ Arrays from 1,500 to 2.6M markers per sample
- Millions of wet lab-validated markers available for diverse populations
- Full annotation by population, function, and frequency
- Online Axiom® Design Center for easy marker selection
- Bioinformatics support for complex designs
- Six to eight week delivery after design submission

Results-focused: automated, hands-free microarray processing

- Enabled by the GeneTitan® Multi Channel (MC) Instrument
- Minimal manual intervention for run-to-run consistency

The Axiom® Genotyping Solution is a scalable and robust platform

The Axiom® Solution was designed specifically to overcome challenges associated with genotyping projects, including:

Study design

- Coverage in non-European populations
- Selection and performance of user-defined marker panels
- Sample diversity and quality
- Cost per sample

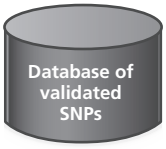
Sample processing

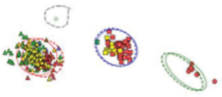
- Reproducibility and reliability across large sample sets
- Sample throughput and time to results
- Hands-on time and workflow automation

Data analysis

- Data quality
- Success rate of user-defined markers
- Meta-analysis and imputation with legacy datasets

The complete workflow solution





Axiom® Genomic Database	**Axiom pre-designed and custom arrays**	**Beckman Biomek FXᴾ Target Prep Express**	**Affymetrix® 2.0 Reagent Kit**	**GeneTitan® MC Instrument**	**Genotyping Console™ Software**
>11M validated common and rare variants	1,500 to 2.6M variants per sample	Automated target preparation*	New robust and reliable assay	Automated, hands-free array processing	Complete and user-friendly software suite

*Manual target preparation protocols are also fully supported.

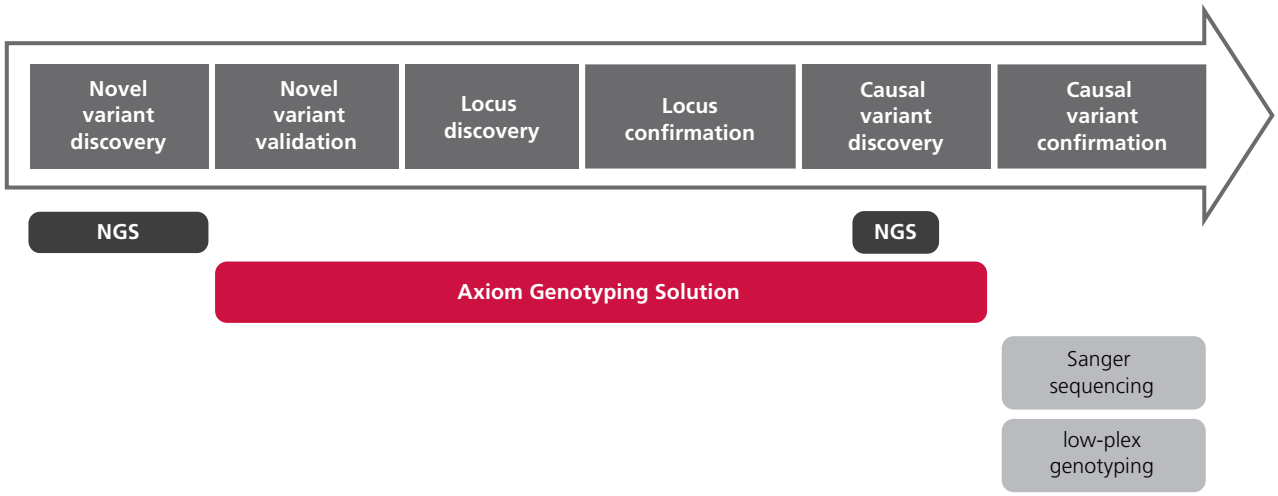
The Axiom® Genotyping Solution enables risk-free, validated, next-generation genotyping

Remove the risk from variant detection and validation

Reduce error rates and costs by validating all your novel variants from next-generation sequencing (NGS). The Axiom® Genotyping Solution enables scientists to confirm putative variants from any depth of sequencing coverage to identify high-confidence markers that can be carried forward into genotyping studies.

- Eliminate sequencing errors from rare variants
- Obtain robust allele frequency and linkage disequilibrium (LD) estimates
- Design optimized genotyping arrays for follow-up studies

Figure 1: The Axiom Genotyping Solution integrates with next-generation sequencing to enable a complete genetics workflow.



Take the guesswork out of custom array design

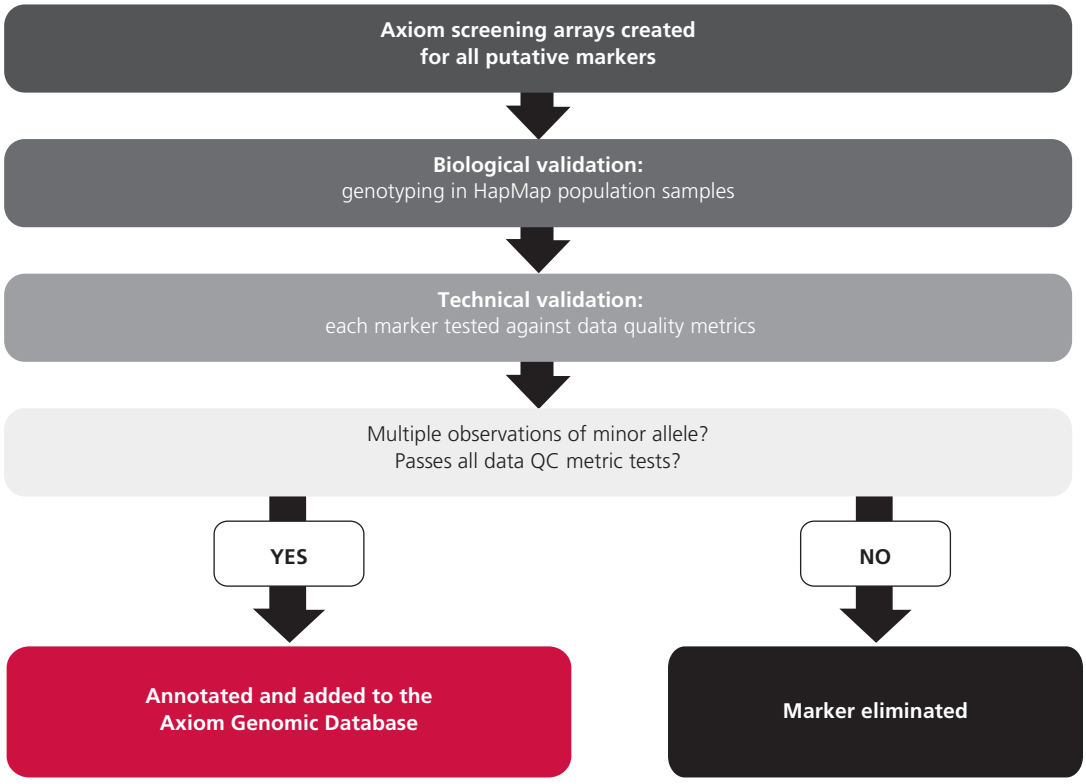
Whether you need whole-genome or targeted genotyping, the Axiom® myDesign™ pipeline enables you to create your own array design quickly, easily, and cost-effectively. Once submitted, your design is manufactured and delivered within six to eight weeks.

To simplify marker selection, we wet lab-validated and annotate millions of variants from international discovery projects to create the Axiom® Genomic Database (AGD). This is the world's largest resource of high-performance genetic markers which are informative in major populations.

Markers in the AGD are fully annotated to enable designs from 1,500 to 2.6 million markers for targeted genotyping of genes, pathways, regions, diseases, or for whole-genome applications.

- Greater than11M common and rare SNPs and insertion/deletions (in/dels)
- Confirmed polymorphisms with no false positives
- Easy marker selection via the Axiom® Design Center
- Proven genotype performance with the Axiom Assay
- Greater than 95 percent "marker success rate" on myDesign Arrays

Figure 2: Markers are stringently wet lab-validated for inclusion in the Axiom Genomic Database, enabling confident selection for custom myDesign™ Arrays.



The Axiom® Genotyping Solution enables genetic studies in diverse populations

One size does not fit all – choose population-optimized genotyping

Choose from a portfolio of population-optimized, pre-designed Axiom® Arrays for African, Asian, and other populations. It is no longer necessary to settle for a European or generic array design to study genetically complex or admixed populations. Population-optimized designs are available for genome-wide association, replication, and population genetics studies.

Table 1: Optimized pre-designed arrays offer increased study power in diverse populations and applications

Target populations	Common variant studies	Common and rare variant studies	Replication and epidemiology
Northern Europe		Axiom® CEU	Axiom® EUR
Asian	Axiom® CHB	Axiom® ASI	Axiom® EAS
African		Axiom® PanAFR	Axiom® AFR
Southern European and Latin American			Axiom® LAT
	MAF>5%	**MAF>2.5%**	**MAF>1%**

Create a haplotype map in novel, uncharacterized populations

The Axiom® Genomic Screening Service offers scientists an alternative to intensive and costly sequencing for characterization of novel populations. Population samples can be quickly screened against 5.4M common and rare variants to estimate allele frequency and linkage disequilibrium. The Genomic Screening Service report enables selection of the most informative tagging SNPs for design of downstream genetic studies.

Sample submission

Investigator submits sample set from target population

↓

Axiom Genomic Screening Service

Samples screened against 5.4 million SNPs and in/dels on Axiom® Arrays

↓

Axiom Arrays Screening Service Report

- Population-specific marker list
- Allele frequencies
- Pairwise linkage disequilibrium

↓

TagSNP selection for optimized genotyping

Investigator can develop population-optimized genotyping strategies and myDesign™ Arrays

Confidence in your data

Automated high-quality data output with the Axiom® Genotyping Solution
The robust Axiom® Assay combines with the hands-free GeneTitan® MC Instrument to produce high-quality data from multiple samples with minimal operator intervention.

Very large studies numbering tens of thousands of samples have successfully implemented the Axiom Genotyping Solution and reported consistently high-quality data even on variable sample types such as saliva.

Table 2: Typical genotyping data quality metrics from an Axiom® Array Plate

Metric	Specification	270 HapMap
Average SNP call rate	>99%	99.60%
Average HapMap concordance	>99.5%	99.80%
Average sample repeatability	>99.8%	99.90%

For more information on the Axiom Genotyping Solution, visit www.affymetrix.com or contact your local Affymetrix Account Manager.

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