Axiom Biobank Genotyping Arrays

High-value genotyping for large sample cohorts to explore genetics of complex diseases

Highlights

We have collaborated with thought leaders in the industry to design Applied Biosystems™ Axiom™ Biobank Genotyping Arrays, a high-throughput solution for affordable genotyping of large sample collections such as those screened at biobanks, genome centers, and core labs.

The arrays, available in both catalog and custom versions, incorporate multiple categories of content, including a genome-wide association study (GWAS) panel of markers for genome-wide coverage in major ethnic groups, rare-coding SNPs and indels for exome analysis, pharmacogenomic markers, expression quantitative trait loci (eQTLs), and newly discovered loss-of-function variants, including sequence insertions and deletions from recent exome sequencing initiatives. See Table 1 for the categories of content on the arrays.

Table 1. Categories of SNPs and indels on Axiom Biobank Genotyping Arrays.

Category	No. of markers*
Genome-wide association markers	246,000
Exome SNPs and indels	265,000
Novel loss-of-function SNPs and indels	70,000
eQTL	23,000**
Pharmacogenomic markers	2,000
Custom markers (optional)	~115,000
Total	**675,000**

* Content in the categories may overlap.
** Value includes taggable SNPs and indels.

Flexible format

The array plates are configured in two formats:

- Axiom Biobank Genotyping Array
  - Preselected content
  - Designed to fit any budget
  - ~675,000 SNPs and indels
- Axiom Biobank Plus Genotyping Array
  - Includes Axiom Biobank Genotyping Array content plus up to 115,000 markers of your choice

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Array design

GWAS markers are common variants intelligently selected via an imputation-based selection strategy for genome-wide coverage in European, Asian, African, and Latino populations, enabling efficient imputation of millions of additional markers [1–3]. See Table 2 for imputed genomic coverage in several HapMap populations.

Table 2. Imputed genomic coverage of the GWAS markers with the indicated minor allele frequencies (MAF).

	$r^2 > 0.8$		Average r^2	
Population*	MAF ≥1%	MAF ≥5%	MAF ≥1%	MAF ≥5%
CEU	72.8%	87.3%	81.3%	90.3%
FIN	73.3%	86.4%	82.0%	90.2%
TSI	66.0%	82.4%	76.9%	87.7%
YRI	38.8%	45.8%	65.6%	73.1%
MXL	66.5%	78.1%	78.3%	85.8%
CHB	59.7%	72.5%	72.0%	82.8%

* Ethnic populations: Western European (CEU), Finnish in Finland (FIN), Toscani in Italy (TSI), Yoruban in Ibadan, Nigeria (YRI), Mexican ancestry in Los Angeles, USA (MXL), Han Chinese in Beijing, China (CHB).

The GWAS markers were selected from the Applied Biosystems™ Axiom™ Genomic Database of SNPs and indels. All markers have been genotype-tested in reference samples and are known to be polymorphic in HapMap samples.

Exome SNPs and indels are nonsynonymous coding markers in exonic regions of the genome, making this a powerful tool for identification of causal variants in complex diseases. A majority of the variants are very rare with MAF ≤1%. Variant types included are nonsynonymous cSNPs, human leukocyte antigen (HLA) markers, fingerprinting markers, mtDNA variants, Y chromosome markers, miRNA target sites, and ancestry-informative markers.

This content was derived from the Exome Chip Design Consortium, the NHLBI Exome Project, the Genetics of Type 2 Diabetes (GoT2D) program, the 1000 Genomes Project, the Cancer Genome Atlas Project, the SardiNIA Medical Sequencing Study, the Autism Exome Sequencing Study, the UK10K project, and others.

Loss-of-function SNPs and indels are newly discovered markers from a sequencing initiative of 26,000 individuals. This category of markers includes known disease-causing mutations and potential splice variants.

eQTLs were selected from the Axiom Genomic Database to cover markers with known associations to RNA expression traits. Over 70% of unique eQTLs in the NCBI Genotype-Tissue Expression (GTEx) eQTL database are represented on the array.

Pharmacogenomic markers were selected to represent phases of absorption, distribution, metabolism, and excretion (ADME). This content was derived from the PharmaADME and PharmGKB databases. Custom markers are variants of your choosing, allowing you to optimize disease or population coverage. Markers may be *de novo* or selected from the Axiom Genomic Database of genotype-tested SNPs, such as predefined population booster sets.

Low running costs translate to high-powered studies

The Axiom Genotyping Solution utilizes a unique 96-sample format. The Applied Biosystems™ GeneTitan™ Multi-Channel (MC) Instrument automates array processing from target hybridization to data generation and can process up to eight array plates per week. Applied Biosystems™ Genotyping Console™ Software automates data analysis and includes allele-calling algorithms and user-friendly visualization tools.

The assay and workflow are fully automated, significantly reducing hands-on time and allowing you to run more samples for a more powerful study.

Analysis workflow for Axiom Biobank Genotyping Arrays

The following guides detail the use of Genotyping Console Software or Power Tools (APT) to perform quality control analysis and sample or SNP filtering prior to downstream analysis: Axiom Genotyping Solution Data Analysis Guide (P/N 702961) and the Best Practice Supplement to Axiom Genotyping Solution Data Analysis User Guide (P/N 703083). The benefit of the advanced analysis workflow is that it offers the greatest flexibility in finding informative content in each dataset.

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References

1. Hoffmann TJ, et al. (2011) Design and coverage of high throughput genotyping arrays optimized for individuals of East Asian, African American, and Latino race/ethnicity using imputation and a novel hybrid SNP selection algorithm. *Genomics* 98(6):422–430.
2. Hoffmann TJ, et al. (2011) Next generation genome-wide association tool: design and coverage of a high-throughput European-optimized SNP array. *Genomics* 98(2):79–89.
3. Howie BN, et al. (2009) A flexible and accurate genotype imputation method for the next generation of genome-wide association studies. *PLoS Genetics* 5(6):e1000529.

Ordering information

Product	Description	Cat. No.
Axiom Biobank Genotyping Array	Contains one 96-array plate; reagents and GeneTitan Multi-Channel Instrument consumables sold separately	902186
Axiom Biobank Plus Genotyping Array	Contains one Axiom Biobank Genotyping Array plus up to 115,000 additional custom markers; reagents and GeneTitan Multi-Channel Instrument consumables sold separately	000854
Axiom GeneTitan Consumables Kit	Contains all GeneTitan Multi-Channel Instrument consumables required to process one Axiom array plate	901606
Axiom 2.0 Reagent Kit	Includes all reagents (except isopropanol) for processing 96 DNA samples	901758

Find out more at **thermofisher.com/microarrays**

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