Axiom PangenomiX and Axiom PangenomiX Plus arrays

Ethnic diversity at your fingertips—the most comprehensive population coverage on a high-throughput array

The Applied Biosystems™ Axiom™ PangenomiX and Axiom™ PangenomiX Plus arrays are human genotyping research arrays designed for whole-genome imputation with globally diverse population coverage. They are an essential research tool in human genomics for applications such as genome-wide association studies (GWASs), population health initiatives, polygenic risk score development and implementation, and clinical research studies in drug discovery. The Axiom PangenomiX and Axiom PangenomiX Plus arrays can scan the whole genome from as little as 100 ng of genomic DNA. This enables identification of single-nucleotide polymorphisms (SNPs), analysis of copy number variants (CNVs), human leukocyte antigen (HLA) typing, and more in a single, cost-effective assay with ready-to-use data analysis.

More than 800,000 markers were selected for high genomic coverage from phase 3 of the 1000 Genomes Project, yielding coverage for European, African, admixed American, East Asian, and South Asian populations. This means variants prevalent in different populations can be accurately represented and accounted for, leading to more inclusive research outcomes.

Features of the Axiom PangenomiX Array:

- **Global population coverage**—more than 800,000 markers across five ancestries: African, admixed American, East Asian, South Asian, and European, including coverage for Hispanic, non-Hispanic White, and non-Hispanic Black
- **Imputation-aware design**—enhanced imputation coverage and accuracy
- **Key disease-related variants**—Alzheimer's disease, cancer, cardiovascular and cardiometabolic diseases, diabetes, neurological disorders, SARS-CoV-2, host response, and immune-related markers
- **Blood group genotyping variants**—comprehensive blood module for the detection of a wide range of extended and rare blood groups, tissue (HLA) types, and platelet (HPA) antigen types, as well as hemoglobinopathy carrier variants that can enhance population research or epidemiology studies
- **HLA typing**—11 major histocompatibility complex (MHC) Class I and Class II loci
- **CNV analysis**—fixed regions and copy number discovery
- **Disease, pharmacogenomic, and pathogenic design**—from broadly referenced public databases, such as ACMG 73, ClinVar, the Clinical Pharmacogenetics Implementation Consortium (CPIC™) guidelines, the Pharmacogenomics Knowledge Base (PharmGKB™ resource), the NHGRI-EBI GWAS catalog, and the absorption, distribution, metabolism, and excretion (ADME) database

Additionally, the Axiom PangenomiX Plus Array provides:

- Over 100 additional markers associated with important haplotypes in genes, including *CYP2D6*, *CYP2A6*, *CYP2B6*, *CYP2C19*, *CYP1A2*, *CYP2C8*, and *SULT1A1*
- Markers in pseudogenes and with high sequence homology
- Reports on 113 genes, a subset with star alleles and phenotype/metabolizer status

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Superior coverage and accuracy

How do we stack up against population-scale disease testing products on the market today? The Axiom PangenomiX and Axiom PangenomiX Plus arrays were designed with your disease and pharmacogenomics (PGx) research studies in mind, based on years of expertise developing and testing arrays used in landmark population disease testing like the PMDA and UK Biobank arrays. Allow us to walk you through the Applied Biosystems™ Axiom™ array difference by **contacting one of our knowledgeable predictive genomics professionals**.

Pharmacogenomics coverage: Axiom PangenomiX and Axiom PangenomiX Plus arrays

The Axiom PangenomiX and Axiom PangenomiX Plus arrays include over 5,000 variants in 1,100 genes of known PGx relevance. This evidence-based content allows researchers to gain valuable insight into an individual's ability to process drugs based upon high, moderate, and preliminary scientific evidence.

The PGx content module includes:

- 2,000 markers in Very Important Pharmacogenes as identified by PharmGKB
- 300 markers associated with PharmGKB level 1A–2B annotations
- >550 reportable alleles mentioned in CPIC guidelines

Table 1. Axiom PangenomiX and Axiom PangenomiX Plus arrays disease-specific risk variants.*

Category	Number of markers
Cancer	>13,000
Mental, behavioral, neurological, and neurodevelopmental	>4,300
Cardiovascular disease	>8,500
Diabetes	>1,500
Musculoskeletal disease	>5,900
Inherited eye disease	>3,700
Autoimmune and inflammatory disease	>1,150
Loss of function, autosomal inheritance	>3,600
Respiratory disorder	>500

* Disease categories as classified by NHGRI, OMIM®, and ClinVar databases.

Table 2. Examples of key genes covered by the Axiom PangenomiX and Axiom PangenomiX Plus arrays.**

Cardiology	Infectious diseases
CYP2D6	*HLA-A/B/C*
CYP2C19	*UGT1A1*
CYP2C9	*CYP2B6*
VKORC1	*CYP2C19*
CYP4F2	*APOE*

Oncology	Pain management	Mental health
DPYD	*CYP3A5*	*CYP2B6*
CYP2D6	*CYP3A4*	*CYP2D6*
CYP2C19	*OPRM1*	*CYP2C19*
CYP2A6	*CYP2D6*	*CYP2C9*
TPMT	*CYP2B6*	*CYP3A4*
UGT1A1	*COMT*	*CYP3A5*
SLCO1B1	*CHRNA5*	*MTHFR*
NUDT15	*OPRM1*	*HLA-B (HLA-B*15 :02:01)*

** PharmGKB clinical annotation levels 1A–2B, accessed May 2021.

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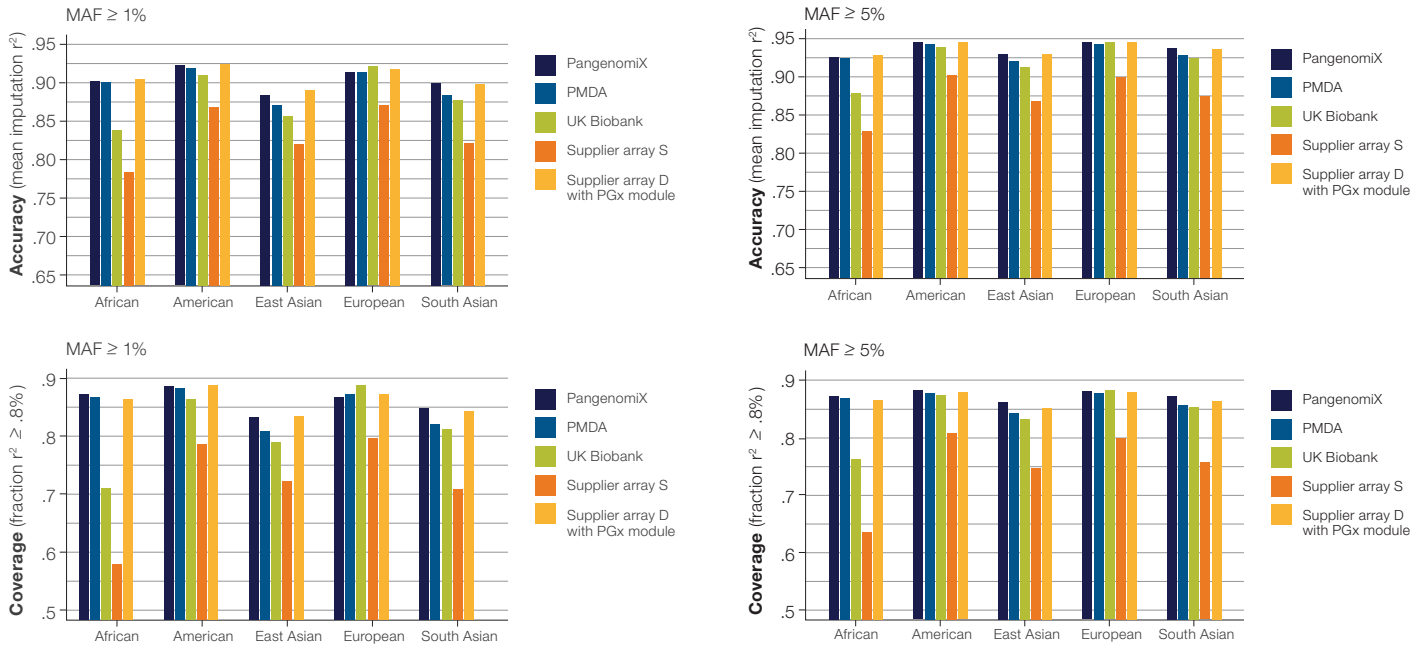

Figure 1. Comparison of coverage and accuracy by minor allele frequency (MAF) and ancestral population. The Axiom PangenomiX and Axiom PangenomiX Plus arrays use imputation to deliver our highest level of accuracy and coverage in human genotyping arrays for all main ancestral populations. PMDA: Applied Biosystems™ Axiom™ Precision Medicine Diversity Array; UK Biobank: Applied Biosystems™ UK Biobank Axiom™ Array.

Axiom PangenomiX Plus Array

Broad-coverage, population-scale disease and PGx testing

The Axiom PangenomiX Plus Array includes all the same great benefits as the Axiom PangenomiX Array but is designed to enhance PGx testing. When used in conjunction with the Applied Biosystems™ Axiom™ 2.0 Plus Assay or Axiom™ Propel Plus Assay, the PangenomiX Plus Array unlocks over 100 additional markers associated with important haplotypes in genes, including *CYP2D6*, *CYP2A6*, *CYP2B6*, *CYP2C19*, *CYP1A2*, *CYP2C8*, and *SULT1A1*. This unique assay opens up the ability to genotype these important PGx markers that are in highly homologous regions of the genome. Based on gene-specific amplification, the Axiom 2.0 Plus Assay overcomes limitations observed in other microarray technologies, making it the array of choice for PGx research.

Furthermore, the Axiom PangenomiX Plus Array comes with Pharmacogenomic Translation Reports that include star allele calling and phenotype prediction. Star allele calling is informed by copy number measurement for genes like *CYP2D6*.

Pharmacogenomic Translation Reports include:

- >110 genes
- >75 relevant gene haplotypes
- >1,000 important haplotypes
- >30 genes with phenotype predictions (e.g., intermediate metabolizer)

Table 3. The *CYP2D6* gene requires both determination of copy number state and SNP genotype calling for accurate star allele reporting. This table shows the workflow in the Applied Biosystems™ Axiom™ Analysis Suite Software to get to the final diplotype/metabolizer status for a sample.

	Step	Output	Interpretation
1	CNV state detection	*CYP2D6*	There is one copy number state present for the *CYP2D6* gene in this sample
2	SNP genotype calling	*rs16947 'A'* *rs1135840 'G'*	Variant haploid genotypes
3	Star allele calling	Diplotype: *CYP2D6* *2/*5 Phenotype: intermediate metabolizer	A sample with one normal-function allele (*2) and one no-function allele (*5) is considered an "intermediate metabolizer" for *CYP2D6*

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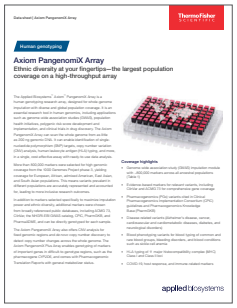
Table 4. Content of the Axiom PangenomiX Array compared with other catalog Axiom arrays.

Category	Description	Axiom PangenomiX Array	Axiom PMD* Research Array	Axiom PMR* Array	UK Biobank Axiom Array
PGx (ADME)	Number of ADME genes covered	>1,100	>1,100	>660	>900
	Markers from PharmGKB with known relevance to drug metabolism	>5,000	>5,000	>1,950	>2,400
	Total number of markers in ADME genes	>92,000	>92,000	>49,000	>67,000

* PMD: Precision Medicine Diversity; PMR: Precision Medicine Research.

Ordering information

Product	Quantity	Cat. No.
Axiom PangenomiX Array—combo kits (array, reagents, and consumables)		
Axiom PangenomiX Array Kit with Axiom 2.0 Assay	96 samples	952519
Axiom PangenomiX Array Kit with Axiom Propel 4X Assay and Fast Wash	4 x 96 samples	952528
Axiom PangenomiX Array Kit with Axiom Propel 8X Assay and Fast Wash	8 x 96 samples	952529
Axiom PangenomiX Plus Array—combo kits (array, reagents, mPCR reagents, and consumables)		
Axiom PangenomiX Plus Array Kit with Axiom 2.0 Plus Assay	96 samples	952521
Axiom PangenomiX Plus Array Kit with Axiom Propel Plus 4X Assay and Fast Wash	4 x 96 samples	952530
Axiom PangenomiX Plus Array Kit with Axiom Propel Plus 8X Assay and Fast Wash	8 x 96 samples	952531
Axiom PangenomiX Array—training kits (array, reagents, consumables, and DNA sample plate)		
Axiom PangenomiX Array Training Kit with Axiom 2.0 Assay	96 samples	952522
Axiom PangenomiX Array Training Kit with Axiom Propel 4X Assay and Fast Wash	4 x 96 samples	952416
Axiom PangenomiX Array Training Kit with Axiom Propel 8X Assay and Fast Wash	8 x 96 samples	952417
Axiom PangenomiX Plus Array—training kits (array, reagents, mPCR reagents, consumables, and DNA sample plate)		
Axiom PangenomiX Plus Array Training Kit with Axiom 2.0 Plus Assay	96 samples	952523
Axiom PangenomiX Plus Array Training Kit with Axiom Propel Plus 4X Assay and Fast Wash	4 x 96 samples	952524
Axiom PangenomiX Plus Array Training Kit with Axiom Propel Plus 8X Assay and Fast Wash	8 x 96 samples	952525



Download the data sheet for the Axiom PangenomiX Array

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