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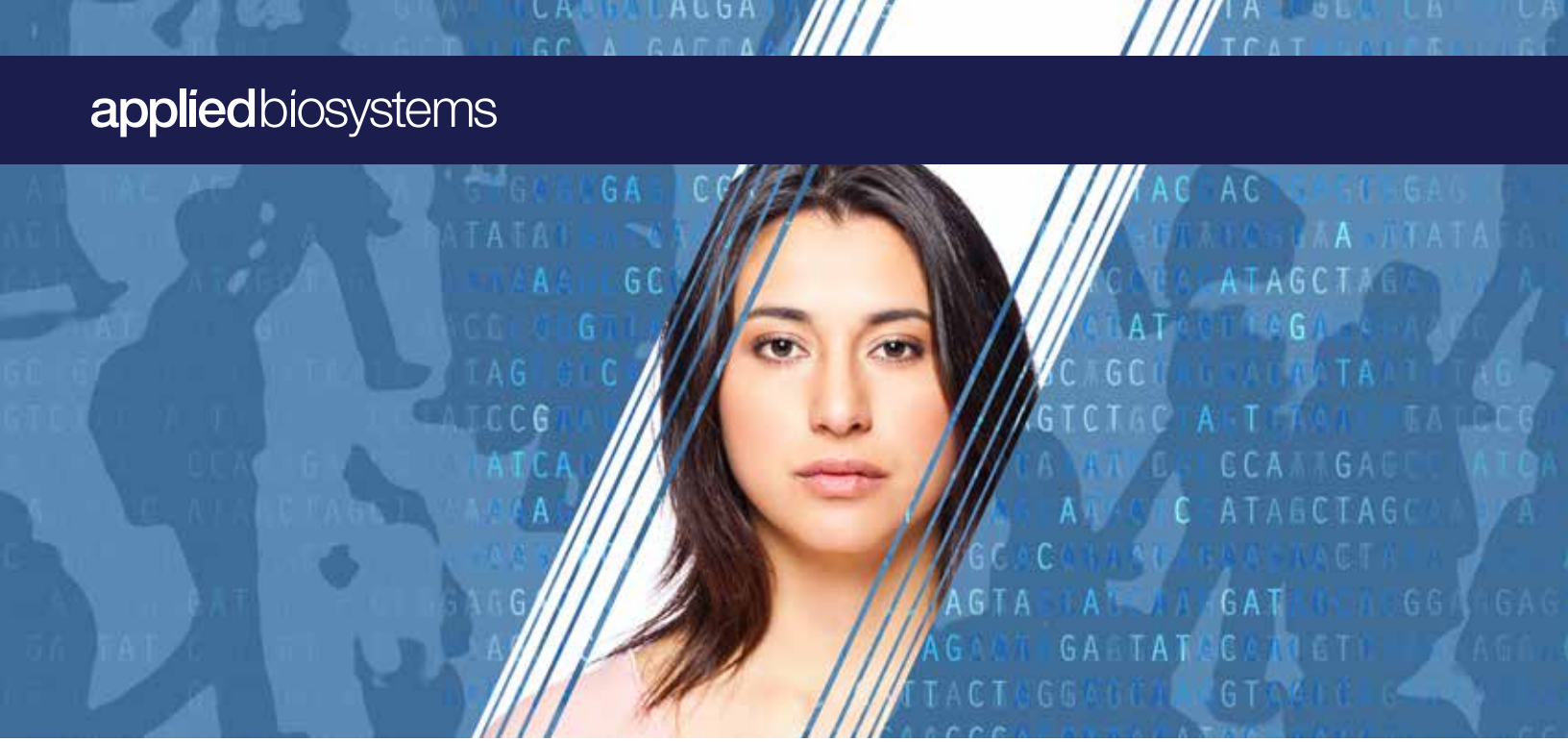

Axiom Precision Medicine Research Array

Content summary

The Applied Biosystems™ Axiom™ Precision Medicine Research Array (PMRA) is a highly affordable genotyping array with comprehensive, high-value content for precision medicine research initiatives, biobanking, and translational research including direct-to-consumer applications. The array includes genomic content to aid in the translation of research results to clinical insight and to help drive the development of new, more effective treatments and wellness plans based on genetic, environmental, and lifestyle factors.

The Axiom PMRA is powered by the Applied Biosystems™ Axiom™ Genotyping Solution used by biobanks worldwide to accelerate their scientific discoveries.

Highlights

- No minimum order commitment; multiyear availability and split shipment options available
- Genome-wide association studies (GWAS) imputation module of 800,000 markers with broad panethnic genomic coverage in more than 25 ethnic and admixed populations around the world
- Coverage of common and rare variants including markers with known associations to human health, pharmacogenomics, cancer variants, immune function, functional variants, and blood phenotype, as well as fingerprint markers for sample tracking and quality control
- Comprehensive analysis support with customization capabilities to empower every study

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The Axiom PMRA reliably addresses the needs of precision medicine and translational researchers with over 900,000 single-nucleotide polymorphisms (SNPs), copy number variants, and insertion/deletion markers, along with a GWAS imputation module covering all five major ancestral population groups for dense genotyping. Comprehensive analysis support and marker customization capabilities are accessible to all researchers and direct-to-consumer providers with no minimum order commitment. Details on array content and marker categories are included in Table 1.

Table 1. Axiom PMRA content summary. Markers were curated specifically to advance precision medicine research.

Category	Number of markers*	Description of content
GWAS markers		
Genome-wide imputation grid	800,000	Markers to maximize panethnic coverage, especially in the 1–5% minor allele frequency (MAF) range, enabling cross-platform and cross-cohort metadata analysis.
NHGRI-EBI** GWAS catalog	>15,000	Content covering the complete NHGRI catalog of published GWAS as of May 2016.
Markers of clinical relevance		
ClinVar	>23,000	Markers with pathogenic or likely pathogenic associations from ClinVar archives.
ACMG	>9,000	A set of markers from the ACMG-published list of genes with intersection in the ClinVar archives.
Exclusive markers	>2,000	Established markers known to be of high clinical importance, such as those in *BRCA1*, *BRCA2*, *CFTR*, *DMD*, and *APOE* genes (with >78% GC content in flanking sequences).
Immune related		
Human leukocyte antigen (HLA)	>9,000	SNPs from the extended major histocompatibility complex region compatible with Applied Biosystems™ Axiom™ HLA Analysis software for improved imputation of HLA alleles in multiethnic populations.
Killer immunoglobulin-like receptor (KIR)	>1,400	Markers to facilitate imputation of the KIR genes.
Autoimmune and inflammatory	>250	Variants with association to specific autoimmune and inflammatory disorders, including ulcerative colitis, Crohn's disease, type 1 diabetes, Graves' disease, Hashimoto's thyroiditis, and celiac disease.
Pharmacogenomic	>1,200	Absorption, distribution, metabolism, and excretion (ADME) markers from the list of variants in the Clinical Pharmacogenetics Implementation Consortium guidelines.
Blood phenotype	>2,000	Markers from GWAS and candidate gene studies associated with red blood cell groups, the regulation of formation of red blood cells and platelets, and the regulation of blood homeostasis.
Cancer common variants	>300	Variants from the list of published common variants associated with cancer phenotypes identified via GWAS, as per NHGRI-EBI GWAS Catalog as well as some recently published and unpublished cancer-associated SNPs as of June 2013.
Functional variants		
Loss of function	>33,000	Markers from Applied Biosystems™ Axiom™ Biobank Genotyping Array as well as human disease mutation and exome databases.
Expression quantitative trait loci	>16,000	Markers to support mapping functional noncoding variations to identify associations with gene transcription variability and differential gene expression.
Fingerprint or sample tracking	>300	Markers shared among several major genotyping platforms, including Rutgers University Identification markers and a set of SNPs used by the University of Washington and the Broad Institute to facilitate sample tracking.
Total number of markers	**>903,000**	

* Markers may be selected for more than one category, but only appear on the array once.

** National Human Genome Research Institute-European Bioinformatics Institute (NHGRI-EBI).

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

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