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Axiom Asia Precision Medicine Research Array

Content summary

The Applied Biosystems™ Axiom™ Asia Precision Medicine Research Array (Asia 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 Asia PMRA is powered by the Applied Biosystems™ Axiom™ Genotyping Solution used by biobanks worldwide to accelerate their scientific discoveries.

Highlights

- No minimum sample volume commitments; multiyear availability and split shipment options available
- Genome-wide association studies (GWAS) imputation module of >540,000 markers from South and East Asia populations
- Coverage of common and rare variants, including markers with known associations to human health, pharmacogenomics, cancer variants, immune function, and functional variants, 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 Asia PMRA reliably addresses the needs of precision medicine and translational researchers with over 750,000 single-nucleotide polymorphisms (SNPs), copy number variants (CNVs), and insertion/deletion (indel) markers, along with a GWAS imputation module covering South and East Asian populations for dense genotyping.

Comprehensive analysis support and marker customization capabilities are accessible to all scientists with no minimum order commitments. Details on array content and marker categories are included in Table 1.

Table 1. Axiom Asia PMRA content summary. The table provides a summary of the content on the Axiom Asia PMRA. Markers were curated specifically to advance precision medicine research.

Category	Number of markers	Description of category content
GWAS markers		
Genome-wide imputation grid	>540,000	Markers to maximize coverage in all East and South Asian populations, especially in the 1–5% minor allele frequency (MAF) range, enabling cross-platform and cross-cohort metadata analysis
NHGRI-EBI GWAS Catalog	>23,400	Content covering the complete National Human Genome Research Institute (NHGRI) catalog of published GWAS as of May 2017
Markers of clinical relevance		
ClinVar	>43,000	Markers with pathogenic or likely pathogenic associations from ClinVar (accessed February 2017) archives
ACMG	>9,200	A set of markers from the American College of Medical Genetics (ACMG)–published list of genes with intersection in the ClinVar archives
Pharmacogenomic	>2,600	Absorption, distribution, metabolism, and excretion (ADME) markers from the list of variants in the Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines
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 (MHC) 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 or inflammatory	~250	Variants with association to specific autoimmune and inflammatory disorders, including ulcerative colitis, Crohn's disease, Graves' disease, Hashimoto's thyroiditis, and celiac disease

Category	Number of markers	Description of category content
Functional variants		
Loss of function	>43,000	Polymorphic markers in East and South Asian populations from the Applied Biosystems™ Axiom™ Biobank Genotyping Array as well as human disease mutation and exome databases
Expression quantitative trait loci	~16,000	Variants with MAF >0.01% in East and South Asian populations to support mapping functional noncoding variations to identify associations with gene transcription variability and differential gene expression
Nonsynonymous variants	~35,000	Includes rare and nonsynonymous coding variations with MAF >0.01% in East and South Asian populations
Lung phenotypes	>7,600	A set of markers having an established or putative association with lung function, lung disease (asthma, cystic fibrosis, chronic obstructive pulmonary disease, idiopathic pulmonary fibrosis, and lung cancer), and smoking behavior
Disease related		
Alzheimer's disease	>900	Variants associated with Alzheimer's disease were selected from a meta analysis of Alzheimer's disease association studies and a set of mitochondrial markers suspected to be associated with the disease from an Alzheimer's disease research group
Cardiometabolic	>360	Known variants associated with various cardiometabolic traits (i.e., coronary disease, lipids, anthropometry, glycemic markers, and blood pressure)
Neurological disorders	~16,000	Markers include a large number of rare mutations with possible association to a variety of neurological diseases, recently identified association hits, and a number of markers chosen from exome-sequencing studies. Cohorts of diseases considered include Alzheimer's disease, frontotemporal dementia, progressive supranuclear palsy, amyotrophic lateral sclerosis, and Parkinson's disease
Diabetes	>500	Variants associated with diabetes identified via GWAS as per the NHGRI-EBI GWAS Catalog (accessed May 2017) and from recent publications that have shown to be associated with diabetes in East and South Asian populations
Cancer common variants	>300	Variants from the list of published common variants associated with cancer phenotypes identified via GWAS, as per the NHGRI-EBI GWAS Catalog as well as some recently published and unpublished cancer-associated SNPs as of May 2017
Rare cancer variants	>2,600	Markers for rare missense variants in proven cancer predisposition genes, selected from sources including locus-specific and human disease mutation databases
Rare cardiac variants	>830	Markers for cardiac disease predisposition include rare variants in the genes *MYBPC3* and *MYH7*, markers chosen from the ARVD/C locus-specific database to look at variants in the genes *DSC2*, *DSG2*, *DSP*, *JUP*, and *PKP2*, and a set of markers chosen from human disease mutation databases for genes related to cardiac disease and hemochromatosis
Other rare variants	>4,700	A set of markers for investigation of the frequency of a set of rare, possibly disease-causing mutations for disorders relevant to lung function, other phenotypes, or that are polymorphic in the Exome Aggregation Consortium (ExAC) data

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Category	Number of markers	Description of category content
Tracking purposes		
Fingerprint and 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
Mitochondrial	~500	Common mitochondrial DNA (mtDNA) variants including coding and noncoding variants associated with mtDNA haplogroups in East and South Asian populations, which can be used as a framework to generate complex phylogenetic networks and conduct comparison between mtDNA haplogroups; also include variants associated with the most common mitochondrial disorders
Y chromosome	~400	Markers defining lineages on the male-specific region of the Y chromosome including markers specific to East and South Asian–specific Y chromosome haplogroups and from the main branches of the Y phylogeny
Total number of markers	>750,000 with ability to add 50,000 novel markers	

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

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

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