

Human genotyping

Axiom™ PangenomeCore Array: pathway-informed genome-wide coverage for multigenic disease and translational research

Highlights

- Over 800,000 probes enable queries of large numbers of genes
- Content has been optimized for research of multifactorial genetic pathologies, population ancestry, pharmacogenomics and infectious respiratory diseases
- Analysis on Applied Biosystems™ SwiftArrayStudio™ Microarray Analyzer enables sample-to-result data generation from 96 samples in approximately 30 hours

Introduction

Genomic analysis has become a cornerstone of modern biomedical research, enabling investigators to identify genetic variants associated with disease susceptibility, therapeutic response, and population diversity. By examining variation across the genome, researchers can uncover the molecular mechanisms underlying complex, multigenic disorders, discover biomarkers, and accelerate translational research. Genome-wide analyses also support pharmacogenomics, ancestry studies, and investigations of immune function, providing insights that improve our understanding of disease biology and inform the development of precision medicine approaches.

A variety of technologies are available for genomic analysis, including whole-genome sequencing (WGS), whole-exome sequencing (WES), targeted sequencing panels, and SNP microarrays. While sequencing helps provide comprehensive variant discovery, it often generates large, complex datasets that require substantial computational resources, longer analysis times, and higher costs, making it less practical for large population studies. As a result, researchers often face trade-offs between breadth of coverage, biological relevance, throughput, and cost.

The Applied Biosystems™ Axiom™ PangenomeCore Array was developed to address these challenges by combining comprehensive genome-wide coverage with specialist-curated, pathway-informed content focused on multigenic disease and translational research. Featuring more than 800,000 variants, the array includes extensive coverage of signaling pathways, immune response genes, pharmacogenomic markers, ancestry-informative loci, and relevant disease-associated variants, while supporting accurate imputation across diverse populations. Integrated with the Applied Biosystems™ Axiom™ SwiftArray™ workflow, the platform facilitates high-quality genotyping results in approximately two days with automated data analysis, enabling researchers to generate biologically meaningful insights rapidly and efficiently for large-scale genomic studies.

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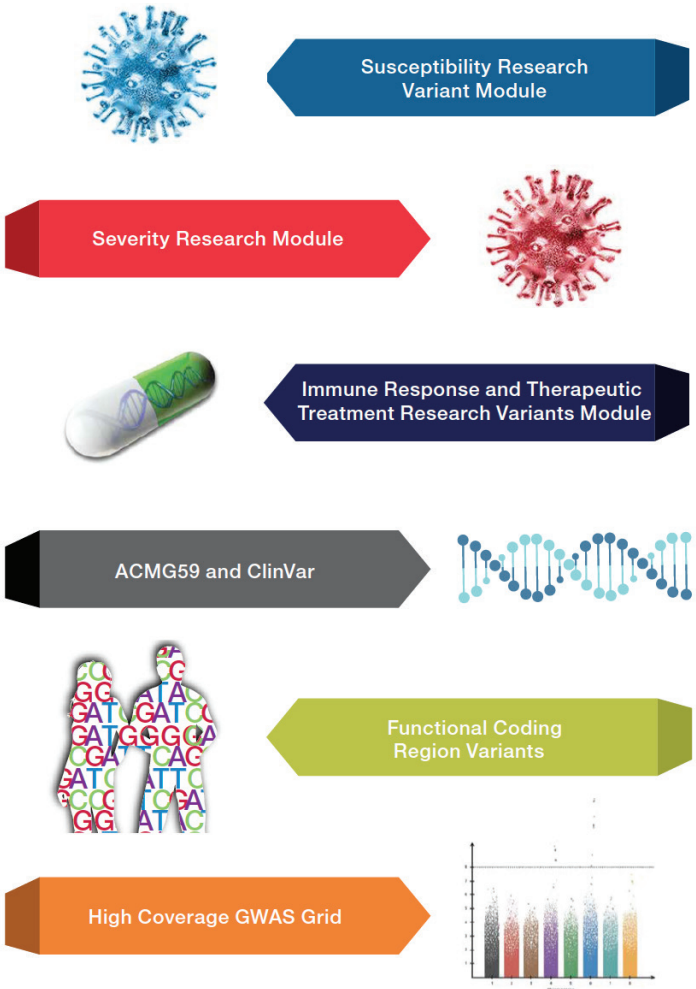


Figure 1. Content modules on the PangenomeCore Array.

Ancestry content

The PangenomeCore Array includes ancestry-informative markers designed to support population genetics research, lineage tracing, and the study of susceptibility to respiratory infectious diseases in specific populations. These markers help provide insights into both recent and ancient population structure, enabling analyses of ancestry, migration patterns, and population diversity. Applied Biosystems™ Axiom™ microarrays are specifically designed with proprietary imputation-based SNP selection algorithms that optimize marker content for both common and low-frequency variation across targeted populations (see our **whitepaper)**. These imputation-aware arrays combine highly accurate direct genotyping with robust downstream imputation (Table 4).

Translational research content

The array includes dedicated content to support investigations into host genetics and immune response across a range of infectious respiratory diseases, including influenza and SARS-CoV-2 research.

Host susceptibility research module

- Includes structural variants in host pathways associated with pathogen entry, immune function, and inflammatory responses.
- Covers key signaling pathways that help investigate genetic factors influencing individual susceptibility and host responses to infectious diseases, including SARS-CoV-2 and other pathogens.
- Provides dense coverage of the X chromosome to support studies of sex-linked innate immunity.

Disease severity research module

- Includes variants associated with chronic underlying conditions that may influence disease severity.
- Covers blood group markers, including the ABO gene and related blood phenotypes that have been associated with disease outcomes in certain infectious respiratory diseases.

Immune response and therapeutic research module

- Includes variants in genomic regions involved in immune regulation and host-pathogen interactions.
- Supports research into immune responses, vaccine development, therapeutic target identification, and pharmacogenomics for various diseases.

Table 1. Categories of markers on the PangenomeCore Array

Category	Number of Markers
Ancestry	3368
Blood phenotypes	770
Cancer	2565
Cardiac	1487
COVID-19 and Host Response	107799
eQTL	27842
Exome	21797
GWAS Grid	727548
HLA imputation	8515
Human Disease	9647
IGH Imputation	2164
Immunity, Inflammation, and HLA	689
KIR imputation	1459
Lung Function Phenotypes	8243
miRNA gene and miRNA processing variants	1078
miRNA target-site variants	5700
Pathogenic variants (ClinVar)	4313
Pharmacogenetics/ADME	4755
Published GWAS Hits	34190
Sample Tracking	348
SARS-CoV-2 putative receptor variants	1227

Pathologies queried by Axiom PangenomeCore Array

The array also includes dedicated content supporting research into complex, multifactorial diseases (Table 3). Some of these include:

Inflammation & immune activation genes

- Cytokine cascade, macrophage activity, mucosal immunity
- 1,946 inflammation genes, 338 cytokine-cascade genes and 968 macrophage-activity genes are represented.

Autoimmune and rheumatology genes

- The array includes rheumatoid arthritis plus immune pathway biology genes relevant to patient stratification research.
- 1,202 rheumatoid-arthritis associated genes, including *TNF*, *NF-κB*, B-cell, *JAK/MAPK* and *TGF-β* family members

Cardiometabolic genes

- Genes involved in hypertension, diabetes/insulin resistance and renin–angiotensin biology are present.
- Includes 1,050 hypertension genes and 2,056 diabetes/insulin-resistance genes.

Coagulation and thrombosis genes

- Genes associated with general coagulation, hypercoagulability, PE, DIC and protein C/S deficits are on the array.
- 438 coagulation-associated genes plus thrombosis sub-signatures are represented.

Neurology & sensory biology

- Neuro-immune signaling genes, anosmia, ageusia and neuro-immunopathology-related genes are on the array.

Respiratory stress biology

- The array includes content for analyzing ARDS and severe respiratory infections.

Table 2. Full list of signaling pathways covered on the array.

Signaling pathways, regulatory pathways, and cellular mechanisms*	Number of genes	Variants
NF-kappa signaling	103	>2,500
AMPK signaling	128	>4,600
B cell receptor signaling	80	>2,500
TNF signaling	112	>2,300
MAPK signaling	289	>10,500
IFN signaling	473	>14,400
TGF-beta signaling	92	>2,600
FoxO signaling	128	>3,600
Endoplasmic reticulum stress	930	>28,000
Renin angiotensinogen	23	>1,763
Notch signaling and ligands	53	>1,800
NOD-like receptor pathway	175	>3,400
Toll-like receptor signaling	103	>1,600
Apoptosis	4,038	>100,000

* An explanation of various pathways can be found in genome.jp/kegg/pathway.html.

Table 3. Markers classified into selected categories for pathology research. Note that this is not a complete list.

Details on sub-categories of markers	Variants
Mental, behavioral, neurological, and neurodevelopmental risk variants	
• Alzheimer's disease including ApOE markers	>170
• Parkinson's disease	>70
• Schizophrenia	>400
Autoimmune and inflammatory disease risk variants	
• Inflammatory bowel disease	>170
• Celiac disease	>60
• Crohn's disease	>300
• Graves' disease	>30
Inherited eye disease	
• Macular degeneration	>100
• Glaucoma	>70
Blood phenotypes and blood groups	>1,200
Asthma	>500
Body mass index	>1,100
Lung function phenotypes	>8,000
Copy number variation	>3,000
Markers from Applied Biosystems™ UK Biobank Axiom™ Array	>21,700
Diabetes	>800
Cardiovascular disease	>1,300
Stroke	>50
Lung function and COPD	>9,000
Chronic kidney disease/RAAS	>450

Pharmacogenomic content

The array contains a focused pharmacogenomic (PGx) module with over 5,500 ADME alleles that complements genome-wide queries without replacing discovery genetics. This evidence-based content includes data curated from PharmGKB™ and the Clinical Pharmacogenetics Implementation Consortium (CPIC™). These data allow researchers to investigate an individual's ability to process drugs based upon high, moderate, and preliminary levels of scientific evidence. The array includes:

- Metabolic genes, including markers for Phase I/II enzymes that affect exposure times and clearance.
- Transport genes, including those that affect uptake, efflux and distribution of compounds.
- Safety genes, including extensive coverage of *HLA*, *G6PD*, *DPYD*, *TPMT*, *NUDT15* and related modifiers.
- Target genes for research into factors labeled as modifiers of therapeutic response.

Rapid turnaround time on SwiftArrayStudio Microanalyzer

To enable greater efficiency for researchers in high-throughput microarray studies, the Applied Biosystems™ Axiom™ SwiftArray™ Assay with automated array processing on the

Applied Biosystems™ SwiftArrayStudio™ Microarray Analyzer (Figure 1; for further information, see the **SwiftArrayStudio Microarray Analyzer webpage**). This solution consists of a target preparation that can be completed in a single work day, and the SwiftArrayStudio Microarray Analyzer that automates hybridization, washing and scanning, generating genotypes in 30 hours. This assay reduces the sample-to-answer time from five days to two days, allowing researchers to gather and interpret genetic data to keep pace with their needs. Of course, multiple runs can be initiated in the same week, increasing the throughput to 16 plates (for details, see **this application note**). Finally, the Applied Biosystems™ Automated Axiom™ Analysis Suite Software automates data analysis with allele-calling algorithms, integrated CNV detection, and intuitive visualization tools.

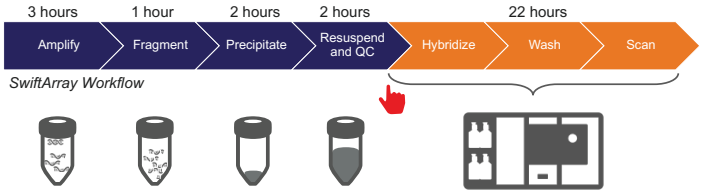


Figure 1. The SwiftArrayStudio Microarray Analysis solution. The Applied Biosystems™ SwiftArray™ Assay target preparation steps can be completed in about eight hours in a single working day. On-instrument, autonomous array processing can generate data within 22 hours of loading. Overall, a full set of genotypes and copy number calls from an Axiom plate can be generated in about 30 hours. Different colors represent workflow steps on different days; finger represents touch point on the instrument, where plates and/or reagents are loaded onto the instrument.

Performance

Thermo Fisher Scientific maintains rigorous performance standards for all microarray products, and the Axiom PangenomeCore microarray meets or exceeds these criteria. These arrays were extensively verified using four different gDNA sources and run with the Axiom SwiftArray PangenomeCore kits. The **Axiom Analysis Suite software** calculated quality-control metrics, including dishQC, QC call rates, and reproducibility measurements. All array examples met the performance specification requirements (Table 5).

Table 4. Number and accuracy of markers with $r^2 > 0.8$ covering the ancestral populations in phase 3 of the 1000 Genomes Project.

	Imputation Accuracy		Number of imputed markers with $r^2 > 0.8$
Ancestral population covered on array	MAF > 1%	MAF > 5%	MAF > 1%
African (AFR)	90%	92%	14.9M
Admixed American (AMR)	92%	94%	10.2M
East Asian (EAS)	87%	92%	7.3M
European (EUR)	91%	94%	8.8M
South Asian (SAS)	88%	93%	8.7M

* Accuracy is the mean r^2 calculated across autosomal SNPs from the highest-ranked markers.

Table 5. Verification quality performance of PangenomeCore Array on SwiftArrayStudio Microanalyzer using a variety of genomic DNA samples.

	Spec	Cell Lines	Saliva	Buccal	Blood
Number of samples		96	72	94	72
Percent passing DishQC and QC Call Rate	95.0%	100%	100%	100%	100%
Average total call rate (best and recommended)	99.5%	100%	99.9%	100%	100%
Reproducibility	99.8%	100%	99.9%	100%	100%
Average call rate for mPCR genes	99.5%	100%	99.7%	99.9%	100%
Concordance to known truth (all calls)	99.8%	99.9%	N/A*	N/A*	N/A*

* N/A - not applicable; known truth is not available for these samples.

Ordering information

Description	Quantity	Cat. No
Axiom™ SwiftArray™ PangenomeCore Assay, 1X	1 kit for 1x96 samples	952696
Axiom™ SwiftArray™ PangenomeCore Assay, 2X	1 kit for 2x96 samples	952697
Axiom™ SwiftArray™ PangenomeCore mPCR Assay, 1X	1 kit with mPCR for 1x96 samples	952700
Axiom™ SwiftArray™ PangenomeCore mPCR Assay, 2X	1 kit with mPCR for 2x96 samples	952701
Axiom™ PangenomeCore 96F Array Plate	1 Array Plate for 1x96 samples	952703

Learn more at **thermofisher.com/swiftarraystudio**

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