Data Sheet

Axiom® Genome-Wide Pan-African Array Set

The first array optimized for coverage of populations of African ancestry

The Axiom® Genome-Wide Pan-African (PanAFR) Array Set maximizes coverage of common and rare variants in populations of Yoruba (YRI), Luhya (LWK), and Maasi (MKK) ancestry and admixed populations with West African ancestry. The markers on this array were selected to provide whole-genome coverage as well as coverage of markers in important disease and biological categories such as coding SNPs (cSNPs), pharmacogenomic genes (ADME), cardiovascular genes (CVS), MHC genes, and immune and inflammation pathway genes (I&I). The array also includes markers from the Sanger Center Cancer Gene Census and the National Human Genome Research Institute (NHGRI) Catalog of Published Genome-Wide Association Studies.

The Axiom Genome-Wide PanAFR Array Set provides:

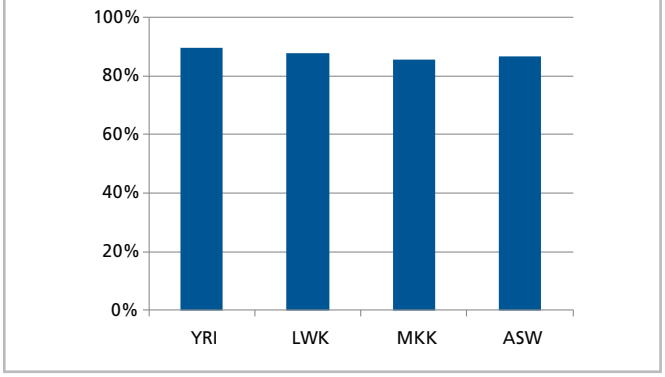
- **Maximum power to detect disease associations in multiple African ancestry populations, with common and rare allele genetic coverage ≥90 percent in YRI and ≥85 percent in LWK and MKK**
- **High coverage of admixed populations, including Americans of African ancestry in Southwestern USA (ASW)**
- **High coverage of common and rare variants in important biological categories**
- **Diverse sources of genomic content, including HapMap, 1000 Genomes, and Southern African Genomes Projects**
- **Fully automated and fast array processing for significantly reduced hands-on time and cost savings**
- **Highly reproducible and reliable data for faster publication**

The Axiom Genome-Wide PanAFR Array Set is part of the Axiom Genotyping Solution, Affymetrix' innovative technology for genotyping studies. Axiom Genome-Wide Arrays are a family of predesigned, population-specific panels that offer optimal coverage for genome-wide association, disease association, and replication studies.

Industry-leading genomic coverage

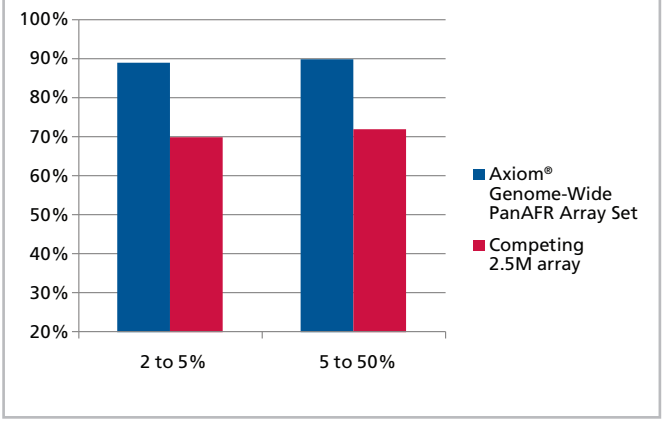
The Axiom Genome-Wide PanAFR Array Set offers the highest genomic coverage of both common and rare alleles from several genomes (including the YRI, LWK, MKK, and ASW genomes), as seen in Figure 1. The high coverage of the ASW genome demonstrates the utility of the Axiom Genome-Wide PanAFR Array Set for association studies in admixed populations.

Figure 1: Axiom® Genome-Wide PanAFR Array Set's genomic coverage of common and rare alleles (MAF 2 to 50%) from several diverse African genomes (YRI, LWK, MKK) and the genome of an admixed population with African ancestry (ASW).



The Axiom Genome-Wide PanAFR Array Set offers superior genomic coverage of the YRI genome compared to competing arrays with similar numbers of markers (Figure 2).

Figure 2: Comparison of genomic coverage of common and rare YRI alleles between the Axiom Genome-Wide PanAFR Array Set and a competing array of 2.5 million SNPs.



In addition to providing excellent genome-wide coverage, the Axiom Genome-Wide PanAFR Array Set also provides higher genetic coverage than a competing 2.5 million SNP array of

rare and common YRI alleles in important biological categories (Figures 3 and 4).

Figure 3: Comparison of genetic coverage of rare YRI alleles (MAF 2 to 5%) across biological categories of the Axiom® Genome-Wide PanAFR Array Set and a competing 2.5 million SNP array.

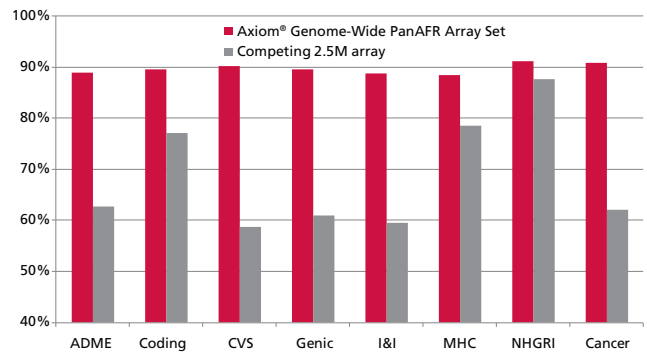
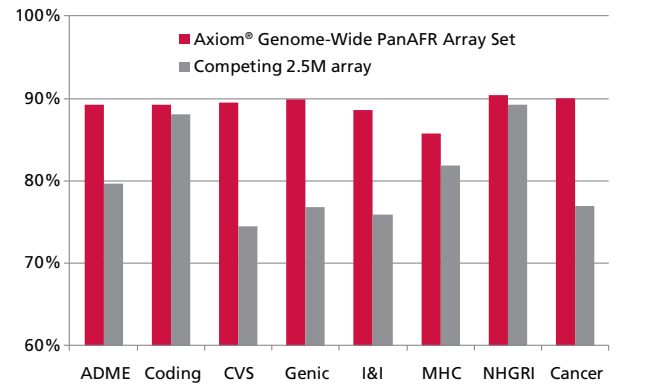


Figure 4: Comparison of genetic coverage of common YRI alleles (MAF 5 to 50%) across different biological categories of the Axiom Genome-Wide PanAFR Array Set and a competing 2.5 million SNP array.

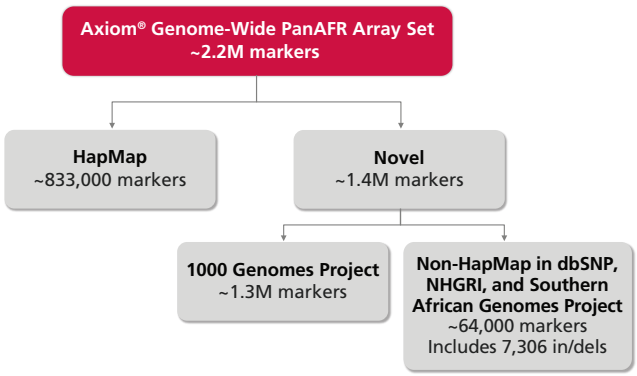


The genomic coverage of the Axiom® Genome-Wide PanAFR Array Set is shown relative to the common and rare YRI alleles found in the Axiom Genomic Database (which includes content from HapMap, dbSNP, all three 1000 Genomes pilot projects, and the Southern African Genomes Project), from which the Axiom Genome-Wide PanAFR Array Set content was selected. For 1000 Genomes Project content, variants were included that were validated by the Axiom 2.0 Assay and/or variants that were discovered by both shallow and deep sequencing projects. Variants that were only discovered using shallow sequencing were not included in the database because these variants tend to have higher false-positive rates and lower genotyping performance compared to the other variants in the database. The genomic coverage of the competing 2.5 million array is taken directly from the manufacturer's published technical specifications.

Array design

The Axiom Genome-Wide PanAFR Array Set maximizes coverage of common and rare alleles from several African genomes (derived from the Axiom Genomic Database). SNP and insertion/deletion (in/del) content was chosen from the sources shown in Figure 5.

Figure 5: Source of genomic content for the Axiom Genome-Wide PanAFR Array Set.



Each marker was empirically tested to ensure reliable detection of the minor allele and performance to stringent performance criteria in the Axiom Assay. Markers were selected to provide high genomic coverage and to represent chromosomes X and Y, mitochondrial SNPs, cSNPs, SNPs in recombination hotspots, ADME SNPs, miRNA SNPs, and disease-associated SNPs (Table 1). The in/dels were selected to supplement the genomic coverage provided by the SNPs.

Table 1: Breakdown of the Axiom Genome-Wide PanAFR Array Set content by biological category.

Markers in functional categories	
cSNP – synonymous	12,142
cSNP – nonsynonymous	12,250
Genic	1,010,216
Conserved	93,548
miRNA associated and mitochondrial	288
Chromosome X	63,902
Chromosome Y	2,337
In/dels	7,306
Intragenic markers by gene function	
ADME	13,473
Cardiovascular	19,837
Cancer	27,576
MHC	2,855
Inflammation and immunity pathway	16,210
NHGRI disease associated	1,340
Total biologically relevant SNPs	
Genic	1,010,216
Non-genic	1,207,186
Total	**2,217,402**

Assay and array performance information

The Axiom® Genome-Wide PanAFR Array Set is part of the Axiom Genotyping Solution, which utilizes a ligation-based assay with a two-color readout. After hybridization of amplified DNA target to an oligonucleotide array, the assay leverages the selectivity of ligation to resolve genotypes.

The Axiom assay supports both manual and fully automated protocols with the same reagent kit and the least amount of hands-on time per sample in the industry.

SNPs on the Axiom Genome-Wide PanAFR Array Set were validated using 180 samples from the YRI extended HapMap population, additional samples from the LWK, MKK, ASW, CEU, CHB, and JPT HapMap populations, as well as 24 paired blood and saliva samples from non-HapMap individuals. Arrays that passed the quality control threshold were analyzed using the Axiom GT1 algorithm. The performance specifications for the array, as well as the metrics achieved, are summarized in Table 2.

Table 2: Performance metrics achieved by the Axiom Genome-Wide PanAFR Array Set.

Performance metric	Specification	HapMap	Blood	Saliva
Average SNP call rate	≥99%	99.63%	99.5%	99.3%
Average HapMap concordance	≥99.5%	99.79%	N/A	N/A
Average reproducibility	≥99.8%	99.87%	99.9%*	

*There were no repeat samples within the set of blood samples; therefore reproducibility was measured by comparing the blood sample genotypes to the genotypes of the matching paired saliva samples.

Sample types

In addition to cell line gDNA, the Axiom Genotyping Assay also supports the following sample types as starting material in the target preparation assay:

- gDNA derived from fresh blood
- gDNA derived from saliva (collected using Oragene® DNA collection kits from DNA Genotek)
- Whole-genome amplified DNA (amplified from gDNA using Qiagen REPLI-g® kits)

Ordering information

Part number	Product	Description
901788	Axiom® Genome-Wide PanAFR Genotyping Bundle	Sufficient for processing 96 gDNA samples Contains: One Axiom® Genome-Wide PanAFR Array Plate Set Each set consists of: ▪ Axiom® Genome-Wide PanAFR Plate A ▪ Axiom® Genome-Wide PanAFR Plate B ▪ Axiom® Genome-Wide PanAFR Plate C Three Axiom® 2.0 Reagent Kits Three Axiom® GeneTitan® Consumable Kits

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