Data Sheet

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Axiom® Genome-Wide AFR 1 Array
World Array 3

The highest available coverage of disease-associated common and rare variants in African American populations for GWAS, replication, and fine mapping in one study

Highlights

- Whole-genome design with highly saturated marker density in strong disease associations and functional relevance
- Coverage-optimized for African American and Yoruban populations to enable the highest discovery with minimum marker redundancy
- GWAS, replication, and fine mapping in one experiment to save time and cost
- Includes thousands of GWAS hit SNPs so you can benefit from the knowledge of prior GWAS
- Intelligent, innovative SNP selection maximizes efficient imputation of millions of additional SNPs
- Fully automated array processing significantly reduces hands-on time and cost

Axiom® Genome-Wide AFR 1 Array is part of Axiom® Genotyping Solution, an innovative technology that supports the entire genotyping workflow from whole-genome to highly targeted gene and causal variant studies. Axiom® World Arrays are a family of predesigned, population specific panels that offer optimal coverage for genome-wide association, replication, and candidate gene association studies.

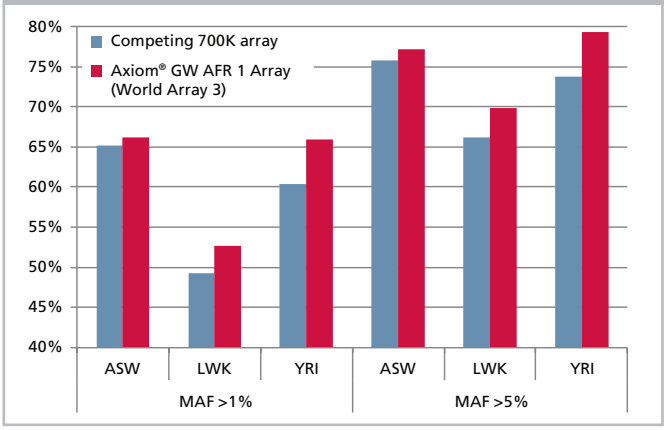
Axiom Genome-Wide AFR 1 Array design

Axiom Genome-Wide AFR 1 Array was designed with the following goals:

- Maximize the number of high-quality SNPs for genome-wide coverage of specific variants and gene regions in African American populations
- Provide coverage down to a minor allele frequency (MAF) of 1% in specific gene-based regions
- Select SNPs to facilitate imputation to the complete HapMap and 1000 Genomes map and provide the highest coverage of rare African American alleles
- Saturate over 5,000 gene regions previously identified as disease-associated from prior GWAS for both replication and fine mapping applications
- Improve coverage of both common and rare variants by utilizing data from the low-pass and high-pass projects of the 1000 Genomes Project
- Incorporate redundant coverage of SNPs with known strong associations with disease or trait outcomes

Genotype imputation was applied to SNP selection for maximum genome-wide coverage. Imputed genomic coverage across three population of African descent can be seen in Figure 1.

Figure 1: Imputed genomic coverage at r^2 >0.8 as measured against rare (MAF >1%) and common (MAF >5%) alleles in three ethnic populations: African Ancestry in SW, US (ASW), Luhya in Webuye, Kenya (LWK), Yoruban in Ibadan, Nigeria (YRI) across a competing 700K array and Axiom Genome-Wide AFR 1 Array. Data generated using 1000 Genomes March 2012 integrated phase 1 release version 3.



SNPs were selected from disease and drug response GWAS databases including the National Human Genome Research Institute (NHGRI) Catalog of Published Associations, the Human Genome Epidemiology Navigator (HuGE) database, the Pharmacogenetics Knowledge Base (PharmaGKB), and the Pharmacogenetics Membrane Transporter (PMT) database. Additionally, common and rare (as low as MAF 1%) SNPs from over 4,000 disease-associated genes were selected from peer-reviewed scientific publications. Table 1 classifies the genomic content of Axiom Genome-Wide AFR 1 Array by SNP type.

Table 1: Count of the Axiom Genome-Wide AFR 1 Array markers by various categories.

Category	Count
SNPs	889,338
Indels	4,293
Mitochondrial SNPs	98
Chromosome Y	234
Chromosome X	26,264
Coding	22,849
ADME	6,246
Cardiovascular	9,401
Cancer	10,371
MHC	9,425
Immune and inflammation	8,495
Total markers	893,631

During array design, SNPs were prioritized based on significance for pharmacogenetic and disease-related traits and were grouped into one of four categories: primary, secondary, tertiary and genome-wide coverage. Table 2 presents a summary of the different tiers, a description of the tier content, sources of the content, and the number of markers in each tier.

The result of this design strategy is Axiom® Genome-Wide AFR 1 Array, which maximizes coverage of known rare and common disease-associated alleles from African and African American genomes, derived from the following HapMap samples: Yoruba (YRI) and Africans in the Southwestern USA (ASW).

Superior performance

Table 3 summarizes the performance metric achieved for Axiom Genome-Wide AFR 1 Array.

Table 3: Performance metrics achieved by Axiom Genome-Wide AFR 1 Array.

Metric	Sample set 1	Sample set 2
Sample type	Cell lines	Saliva
Number of samples	95	768
Percent of samples that passed DQC and call rate cutoffs	100%	96.61%
Average sample call rate	99.67%	99.48%
Average HapMap concordance	99.76%	N/A
Average reproducibility	99.92%	N/A

Sample types

In addition to supporting cell line gDNA as an assay template, 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)

Analysis workflow for Axiom® Genotyping Array

An analysis workflow is recommended to utilize the content of the array. The following guides detail the use of Genotyping

Table 2: Descriptions of the different tiers of markers included on the Axiom Genome-Wide AFR 1 Array.

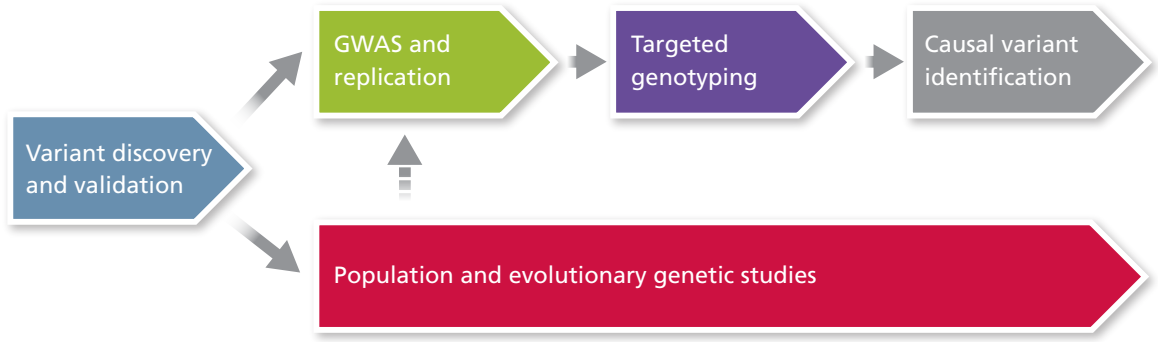
Tier	Description	No. of SNPs
Primary	SNPs from HuGE database and NHGRI Catalog with strong confirmed p-value	270
Secondary	QC SNPs tagging SNP in high LD ($r^2 > 0.6$); imputation SNP ±100 kb of 1' SNP, SNPs suggestive of association but not yet replicated, p-value $<10^{-5}$ association	16,669
Tertiary	SNPs mined from a variety of sources and selected for functional significance (miRNA, splice-site SNPs)	43,398
Genome-wide coverage	Additional SNPs selected to ensure redundancy and genome-wide coverage	833,294
Total number of markers on the array		893,631

Console™ Software or Affymetrix Power Tools to perform quality control analysis and sample or SNP filtering prior to downstream analysis: Axiom Genotyping Solution Data Analysis Guide (P/N 702961) and the *Best Practices Supplement to Axiom Genotyping Solution Data Analysis User Guide for Axiom Genome-Wide EUR 1, EAS 1, LAT 1, and AFR 1 Arrays* (P/N 703106). The benefit of the advanced analysis workflow is that it provides the greatest flexibility in finding the most informative content in each dataset.

Genomics journal publications

Hoffmann T. J., *et al.* Design and coverage of high throughput genotyping arrays optimized for individuals of East Asian, African American, and Latino race/ethnicity using imputation and a novel hybrid SNP selection algorithm. *Genomics* **98**(6):422-430 (2011).

Figure 2: Axiom Genotyping Solutions can help you discover more. Affymetrix offers cost-effective high-coverage arrays enabling a wide range of human genetic research workflows.



Ordering information

Part number	Product name	Description
901848	Axiom® Genome-Wide AFR 1 Array Plate	Includes one 96-array Axiom Genome-Wide AFR 1 Plate (World Array 3)
901606	Axiom® GeneTitan® Consumables Kit	Contains all GeneTitan® Instrument consumables required to process one Axiom® Array Plate
901758	Axiom® 2.0 Reagent Kit	Includes all reagents (except isopropanol) for processing 96 DNA samples

Affymetrix, Inc. Tel: +1-888-362-2447 ■ **Affymetrix UK Ltd.** Tel: +44-(0)-1628-552550 ■ **Affymetrix Japan K.K.** Tel: +81-(0)3-6430-4020
Panomics Solutions Tel: +1-877-726-6642 panomics.affymetrix.com ■ **USB Products** Tel: +1-800-321-9322 usb.affymetrix.com

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