Data Sheet

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Axiom® Genome-Wide EAS 1 Array
World Array 2

The highest available coverage of disease-associated common and rare variants in populations with European and East Asian heritage 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 populations with East Asian and European ancestries 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 saves money

Axiom® Genome-Wide EAS 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® Genome-Wide Arrays are a family of predesigned, population-specific panels that offer optimal coverage for genome-wide association, replication, and candidate gene association studies.

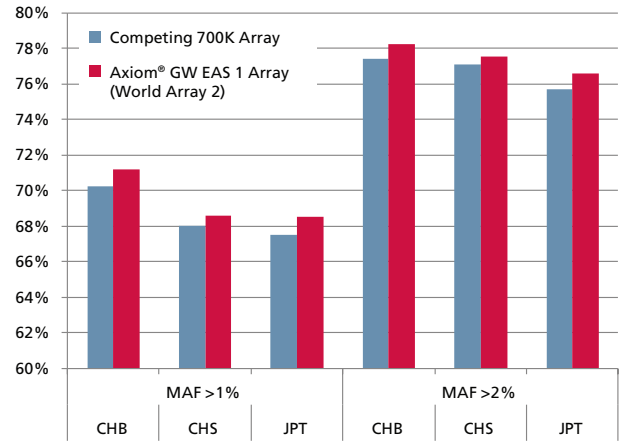
Optimized genome-wide coverage to drive discovery in complex traits and diseases

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

- Select SNPs to facilitate imputation to the complete HapMap and 1000 Genomes map
- Provide coverage down to a minor allele frequency (MAF) of 1% in specific gene-based regions
- 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

Figure 1 shows the genomic coverage of Axiom Genome-Wide EAS 1 Array as measured against rare alleles of the East Asian genome.

Figure 1: Imputed genomic coverage at r^2 >0.8 as measured against rare alleles (MAF >1%, >2%) in three ethnic populations: Han Chinese in Beijing (CHB), Han Chinese South (CHS), and Japanese in Tokyo (JPT) across Axiom® Genome-Wide EAS 1 Array and a competing 700K 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, over 4,000 disease associated genes were selected from peer-reviewed scientific publications to be covered with SNPs with MAF as low as 1%. Table 1 classifies the genomic content of Axiom Genome-Wide EAS 1 Array by biological category.

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

Category	Count
SNP	709,104
Indel	3,846
Mitochondrial SNPs	83
Chromosome Y	158
Chromosome X	13,385
Coding	18,168
ADME	4,375
Cardiovascular	7,070
Cancer	7,546
MHC	10,324
Immune and inflammation	5,747
Total no. of markers	712,950

SNPs were prioritized during the design process 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 contains a summary of the different tiers, a description of the tier content, sources of the content and number of markers in each tier.

Superior performance

Table 3 summarizes the performance achieved in various studies.

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

Metric	Sample set 1	Sample set 2
Sample type	Cell line	Saliva
Number of samples	95	768
Percent of samples that passed DQC and call rate cutoffs	98.95%	96.61%
Average sample call rate	99.82%	99.70%
Average HapMap concordance	99.62%	N/A
Average reproducibility	99.91%	N/A

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 Console™ Software or Affymetrix Power Tools to perform quality control analysis and sample or SNP filtering prior to downstream analysis:

Table 2: Prioritization of SNP selection during design. Descriptions of the different tiers of markers included on the Axiom Genome-Wide EAS 1 Array design.

Tier	Description	No. of SNPs
Primary	SNPs from HuGE database and NHGRI Catalog with strong confirmed p-value	258
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	9,674
Tertiary SNPs	SNPs mined from a variety of sources and selected for functional significance (miRNA, splice-site SNPs)	43,908
Genome-wide coverage	Additional SNPs selected to ensure redundancy and genome-wide coverage	659,110
Total number of SNPs on the array		712,950

Axiom Genotyping Solution Data Analysis Guide (P/N 702961) and the *Best Practice 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.

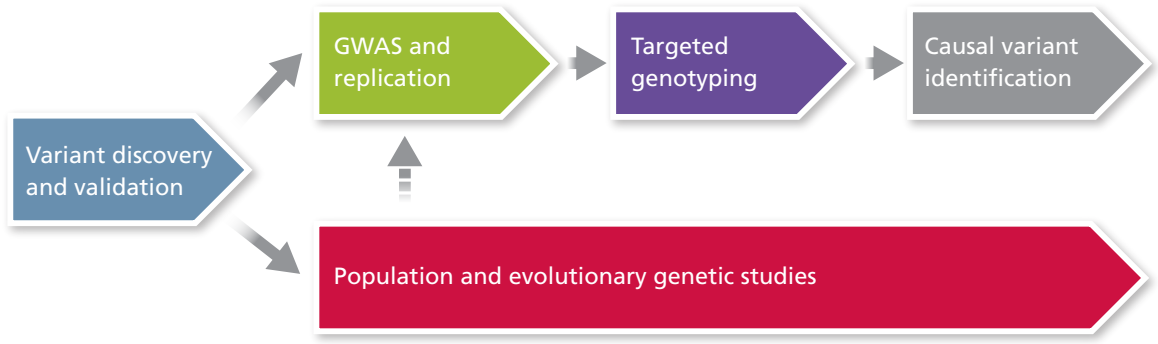
Genomics journal publications

For more information about this array (including design strategy and performance), please refer to the following 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). PMID: 21903159

Altshuler, D., *et al.* A map of human genome variation from population-scale sequencing. *Nature* **467**(7319):1061-1073 (2010). PMID: 20981092

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
901847	Axiom® Genome-Wide EAS 1 Array Plate	Includes one 96-array Axiom Genome-Wide EAS 1 Plate (World Array 2)
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

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