Data Sheet

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Axiom® Genome-Wide EUR 1 Array
World Array 1

The highest available coverage of disease-associated common and rare alleles in Western and Northern European 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 populations of European descent to enable the highest discovery with minimum marker redundancy
- GWAS, replication, and fine mapping in one experiment to save time and cost
- 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 EUR 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.

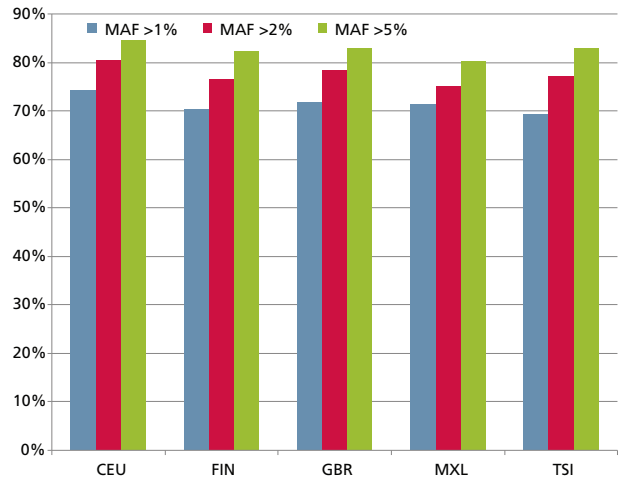
Optimized genome-wide coverage to drive discovery in complex traits and diseases

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

- Select SNPs to facilitate imputation to the complete HapMap and 1000 Genomes map
- Provide coverage with a minor allele frequency (MAF) of 1% in specific gene-based regions
- Saturate regions previously identified as disease-associated from prior GWAS studies for both replication and fine mapping applications
- Improve coverage of both common and rare variants by making use of data from 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 EUR 1 Array as measured against common and rare alleles of the EUR genome.

Figure 1: Imputed genomic coverage at r^2 >0.8 as measured against common and rare (MAF >1%, 2% and 5%) alleles in five Caucasian populations: Western European (CEU), Finnish (FIN), British from England and Scotland (GBR), Mexican in LA, USA (MXL) and Toscani in Italy (TSI) across the Axiom Genome-Wide EUR 1 Array. Data generated using 1000 Genomes March 2012 integrated phase 1 release version 3.



Twenty thousand SNPs were selected from disease and drug response relevant GWAS databases including the National Human Genome Research Institute (NHGRI) Catalog of Published Associations, the Human Genome Epidemiology Navigator (HuGE), 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%. Refer to Table 1 for the number of SNPs in each of these categories.

Table 1: Breakdown of SNPs by biological categories and disease types.

Category	No. of SNPs
SNPs	668,265
Indels	6,252
Mitochondrial SNPs	116
Chromosome Y	289
Chromosome X	13,123
Coding	16,474
ADME	4,542
Cardiovascular	6,959
Cancer	7,187
MHC	9,526
Immune and inflamation	5,496
Total no. markers	674,518

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 contains a summary of the different tiers, a description of the tier content, sources of the content, number of targeted markers in each tier, and the MAF range for each tier.

The result of this design strategy is Axiom® Genome-Wide EUR 1 Array, which maximizes coverage of extremely rare and known disease-associated alleles from Northern/Western European genomes (derived from the CEU HapMap samples).

Superior performance

Markers on Axiom Genome-Wide EUR 1 Array were validated using 95 phase 1 HapMap samples from the CEU population. Arrays that passed the quality control threshold were analyzed using the Axiom® GT1 algorithm. Table 3 summarizes the performance metrics achieved for the array.

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

Performance metric	Specification	Sample data	
Sample type	_	Cell line	Saliva
Number of samples	_	95	760
Percent of samples that passed DQC and call rate cutoffs	_	100%	97.37%
Average SNP call rate	>99%	99.81%	99.77%
Average HapMap concordance	>99.5%	99.76%	N/A
Average reproducibility	>99.8%	99.96%	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).

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

Tier	Description	Source	No. of markers	MAF range
Primary	Markers with a strong disease association as determined by a previous GWAS	NHGRI, HuGE	245	1-5%
Secondary	Markers suggested as being disease associated but not strongly replicated	Literature and public db, PharmaGKB, PMT, NHGRI, HuGE	8,960	1-5%
Tertiary	Functionally significant (miRNA, splice, MHC, coding, 1000 Genomes low pass in functional regions)	Various, 1000 Genomes Project	42,707	1-5%
Genome-wide coverage	Additional SNPs selected to ensure redundancy and genome-wide coverage	HapMap, dbSNP & 1000 Genomes Project	622,606	2-5%
			674,518 (total)	

Analysis workflow for Axiom® Genotyping Arrays

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

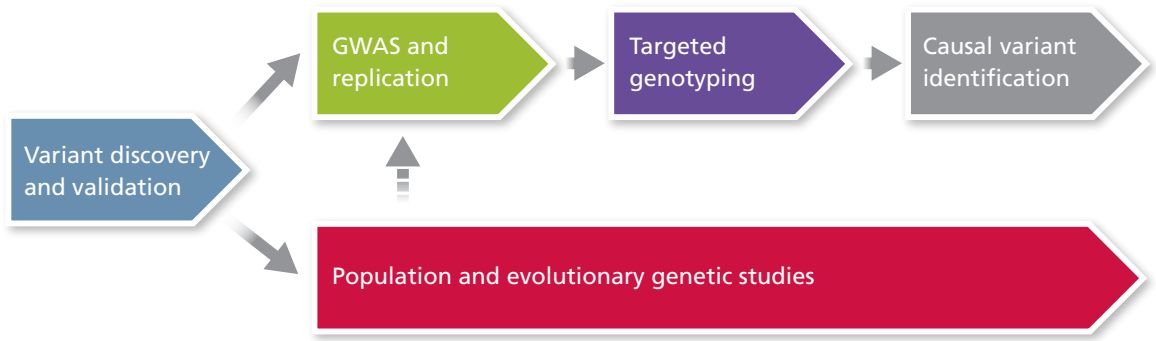
Genomics journal publications

For more information about this array (including design strategy and performance) please refer to the following publications:

Hoffman T. J., *et al.* Next generation genome-wide association tool: Design and coverage of a high-throughput European-optimized SNP array. *Genomics* **98**(2):79-89 (2011).

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

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
901787	Axiom® Genome-Wide EUR 1 Array Plate	Includes one 96-array Axiom Genome-Wide EUR 1 Plate (World Array 1)
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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