Data Sheet

Affymetrix®
Revolutionize life

Axiom™ Genome-Wide ASI 1 Array Plate

The first array that maximizes coverage of rare variants in East Asian populations

The Axiom Genome-Wide ASI 1 Array Plate maximizes genomic coverage of common and rare alleles of a consensus East Asian (ASI) genome, including variants from important biological categories such as coding SNPs, cardiovascular genes, ADME genes, MHC region genes, Sanger Center Gene Census genes, and the National Human Genome Research Institute (NHGRI) Catalog of Published Genome-Wide Association Studies.

Benefits of the Axiom Genome-Wide ASI 1 Array:

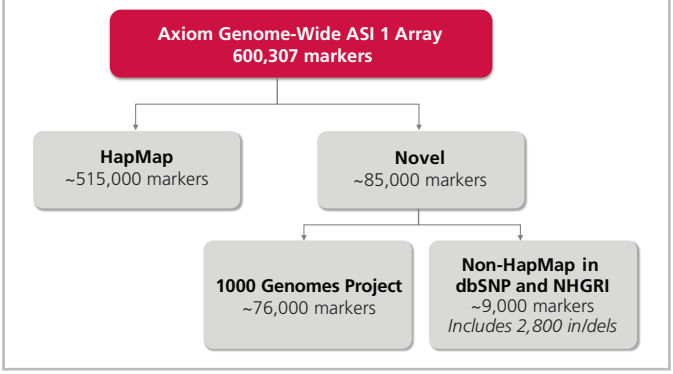
- **Maximized power for ASI populations – the first array with high genomic coverage of rare ASI alleles**
- **Novel common and rare variants**
- **Fully automated and fast array processing significantly reduces hands-on time and saves money**
- **Highly reproducible and reliable data for faster publication**

The Axiom Genome-Wide ASI 1 Array 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, replication, and candidate gene association studies.

Array and kit design

The Axiom Genome-Wide ASI 1 Array maximizes coverage of a consensus East Asian genome derived from the union of the Han Chinese and Tokyo Japanese HapMap genomes. The array also offers high genomic coverage of ASI and CEU admixed populations. SNP and insertion/deletion (in/del) content was derived from various public sources, including HapMap, the Single Nucleotide Polymorphism Database (dbSNP), and 1000 Genomes Project content already in dbSNP (Figure 1). Each marker was tested extensively to ensure reliable detection of the minor allele and performance to stringent performance criteria in the Axiom™ Assay.

Figure 1: Source of genomic content for the Axiom Genome-Wide ASI 1 Array.



SNPS 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 SNPs by biological categories.

cSNP – synonymous	4,427
cSNP – nonsynonymous	10,346
Splicing and untranslated regions (UTR)	12,719
MHC	7,914
ADME	6,736
Genic	266,690
Conserved	28,887
Inflammation and immunity pathway	4,486
NHGRI disease associated	1,481
miRNA associated and mitochondrial	232
Chromosome X	16,410
Chromosome Y	1,584
In/dels	2,834
Total biologically relevant SNPs	**364,746**
Genic	266,690
Non-genic	333,617
Total	**600,307**

SNPs on the Axiom Genome-Wide ASI 1 Array were validated using 190 phase I HapMap samples from the ASI and YRI populations. Arrays that passed the quality control threshold were analyzed using the Axiom GT1 algorithm. Table 2 summarizes the performance specifications for the array as well as the metrics achieved.

Table 2: Performance metrics achieved by the Axiom Genome-Wide ASI 1 Array.

Metric	Specification	190 HapMap
Average SNP call rate	>99%	99.6%
Average HapMap concordance	>99.5%	99.8%
Average repeatability	>99.7%	99.9%

Genomic coverage

Figure 2 shows the genomic coverage of the Axiom Genome-Wide ASI 1 Array as measured against common alleles (minor allele frequency [MAF] greater than 5 percent) and rare alleles (MAF between 2 and 5 percent) of the ASI genome.

Figure 2: Comparison of genomic coverage of common and rare ASI alleles between the Axiom ASI 1 Array and a competing array of 700K SNPs.

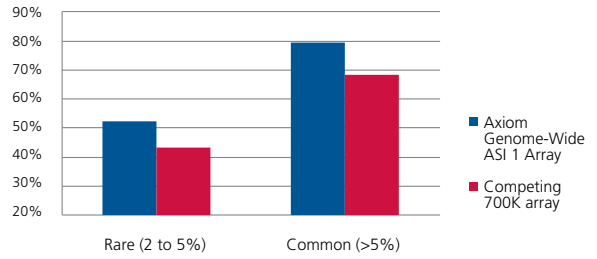
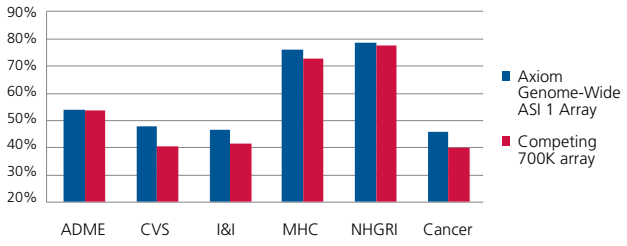


Figure 3: Comparison of genetic coverage of rare ASI alleles across different biological categories by the Axiom ASI 1 Array and a competing 700K SNP array.



The genomic coverage of the Axiom Genome-Wide ASI 1 Array Plate is shown relative to the common and rare ASI alleles found in the Axiom™ Genomic Database, which includes content from HapMap, dbSNP, and all three 1000 Genomes pilot projects. For 1000 Genomes content, variants were included that were validated by the Axiom™ 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 this coverage calculation because of the high false-positive rate associated with shallow sequencing. The genomic coverage of the competing 700K array is taken directly from www.illumina.com.

In addition to providing excellent genome-wide genomic coverage, the Axiom Genome-Wide ASI 1 Array also provides higher genetic coverage than a competing 700K SNP array of rare ASI alleles in important biological categories (Figure 3).

Assay performance

The Axiom Genome-Wide ASI 1 Array is based on the Axiom™ Genotyping Solution, which uses the Axiom™ 2.0 Assay. This ligation-based assay, with a two-color readout, exploits the selectivity of ligation to resolve genotypes subsequent to the amplification of an entire genome via hybridization to an oligonucleotide array, for which hybridization alone may be insufficient.

Oligonucleotide probes are constructed on the surface of the array in 5′ to 3′ order, with a final phosphate group attached to the end of the bound oligonucleotide to enable ligation. The unlabeled target is hybridized to the array, where ligation to 9-mer short oligonucleotides occurs, with the short solution probes beginning with A/T labeled with <dye1> and those beginning with C/T labeled with <dye 2>. In this way, surface probes on the array with the 3′ end immediately before a SNP position can be used to resolve any marker with a weak base against a strong base by analyzing the ratio between the dyes.

Total genomic DNA (200 ng) is amplified and randomly fragmented into 25- to 125-base-pair (bp) fragments, which are purified, re-suspended, and hybridized to Axiom Genome-Wide ASI 1 Array Plates. Following hybridization, the bound target is washed under stringent conditions to minimize background noise caused by random ligation events. Each polymorphic nucleotide is queried via a multicolor ligation event carried out on the array surface. After ligation, the arrays are stained and imaged on the GeneTitan® MC Instrument.

Sample types supported

In addition to cell line gDNA, the Axiom™ 2.0 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
901640	Axiom™ Genome-Wide ASI 1 Array Plate	Contains one 96-array plate
901606	Axiom™ GeneTitan® Consumables Kit	Contains all GeneTitan® consumables required to process one Axiom array plate
901758	Axiom™ 2.0 Reagent Kit	Contains all reagents (except isopropanol) required to process 96 gDNA 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 Products Tel: +1-877-PANOMICS www.panomics.com ■ **USB Products** Tel: +1-800-321-9322 www.usb.affymetrix.com

www.affymetrix.com Please visit our website for international distributor contact information.

For Research Use Only. Not for use in diagnostic procedures.

P/N DNA00450 Rev. 1