Data Sheet

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Axiom™ Genome-Wide CEU 1 Array Plate

The first array to maximize power for European populations with high coverage of rare variants

The Axiom Genome-Wide CEU 1 Array Plate maximizes genomic coverage of common and rare alleles of the CEU genome, including variants from important biological categories such as coding SNPs, ADME genes, cardiovascular 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 CEU 1 Array:

- **Maximized power for CEU populations**
- **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 CEU 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 design

SNP content and insertions/deletions (in/dels) for the Axiom™ Genome-Wide CEU 1 Array were selected from the Axiom Genomic Database. These validated markers were derived from various public sources, including the International HapMap Project, 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. SNPS were selected to provide high global 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.

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

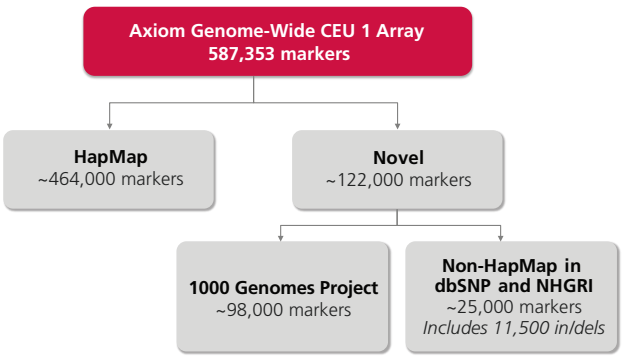


Table 1: Breakdown of SNPs by biological categories.

cSNP – synonymous	5,020
cSNP – nonsynonymous	10,648
Splicing and untranslated regions (UTR)	13,269
MHC	7,914
ADME	4,603
Genic	263,062
Conserved	28,910
Inflammation and immunity pathway	4,532
NHGRI disease associated	1,610
miRNA associated and mitochondrial	266
Chromosome X	16,413
Chromosome Y	1,718
In/dels	11,501
Total biologically relevant SNPs	**369,466**
Genic	263,062
Non-genic	324,290
Total	**587,352**

SNPs on the Axiom Genome-Wide CEU 1 Array were validated using 270 phase I HapMap samples. 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 CEU 1 Array.

Metric	Specification	270 HapMap
Average SNP call rate	>99%	99.6%
Average HapMap concordance	>99.5%	99.8%
Average sample repeatability	>99.8%	99.9%

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

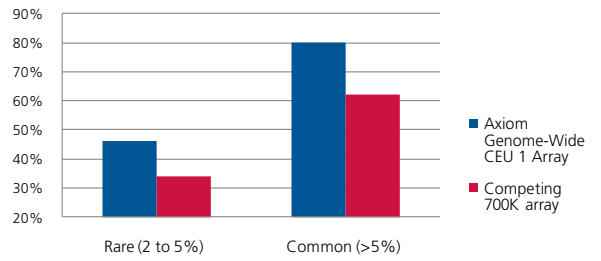
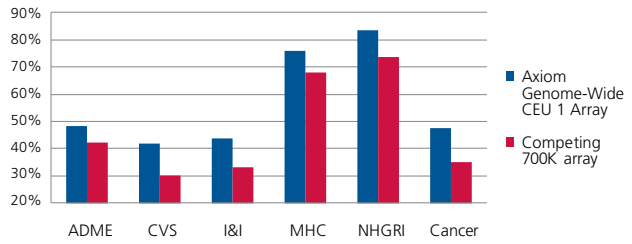


Figure 3: Comparison of genetic coverage of rare CEU alleles across different biological categories by the Axiom CEU 1 Array and a competing 700K SNP array.



Genomic coverage

Figure 2 shows the genomic coverage of the Axiom Genome-Wide CEU 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 CEU genome.

The genomic coverage of the Axiom Genome-Wide CEU 1 Array is shown relative to the common and rare CEU alleles 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 CEU 1 Array also provides higher genetic coverage than a competing 700K SNP array of rare CEU alleles in important biological categories (Figure 3).

Assay performance

The Axiom Genome-Wide CEU 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 CEU 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
901608	Axiom™ Genome-Wide CEU 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

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