

## Axiom™ Genome-Wide CHB 1 Array Plate

The highest coverage available of common variants to detect disease associations in Han Chinese populations

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

### Benefits of the Axiom Genome-Wide CHB 1 Array:

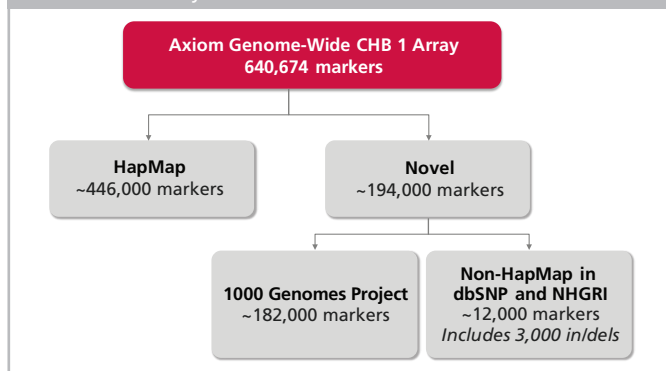
- **Maximized power for CHB populations with the highest genomic coverage of common alleles**
- **High genomic coverage of common variants in biological categories**
- **Most validated 1000 Genomes Project content**
- **Fully automated and fast array processing significantly reduces hands-on time and saves money**
- **Highly reproducible and reliable data for faster publication**

The Axiom Genome-Wide CHB 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

Genomic content for the Axiom Genome-Wide CHB 1 Array was selected from the Axiom™ Genomic Database, which contains validated markers derived from various public sources, including the International HapMap Project, the Single Nucleotide Polymorphism Database (dbSNP), and the 1000 Genomes Project (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 CHB 1 Array.



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 insertions and deletions (in/dels) were selected to supplement the genomic coverage provided by the SNPs.

Table 1: Breakdown of SNPs by biological categories.

cSNP – synonymous	3,243
cSNP – nonsynonymous	4,225
Splicing and untranslated regions (UTR)	7,641
MHC	1,873
ADMET	3,723
Genic	277,429
Conserved	25,507
Inflammation and immunity pathway	4,387
NHGRI disease associated	2,114
miRNA associated and mitochondrial	217
Chromosome X	12,545
Chromosome Y	1,613
In/dels	3,112
<b>Total biologically relevant SNPs</b>	<b>347,629</b>

SNPs on the Axiom Genome-Wide CHB 1 Array were validated using 95 Chinese and Japanese HapMap samples. Arrays that passed the quality control threshold were analyzed using the Axiom GT1 algorithm. The performance specifications for the array as well as the metrics achieved are summarized in Table 2.

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

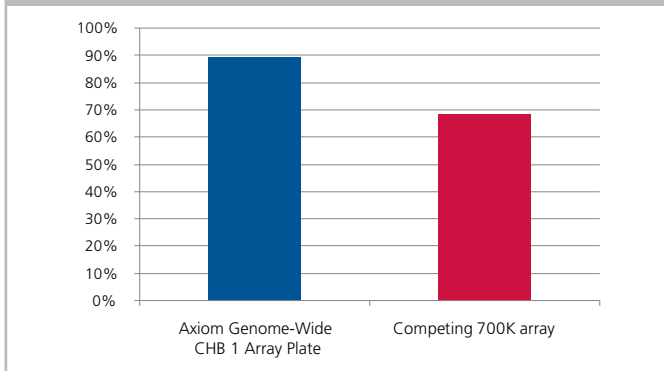
Metric	Specification	95 CHB HapMap
Average SNP call rate	>99%	99.8%
Average HapMap concordance	>99.5%	99.9%
Average reproducibility	>99.7%	100%

### Genomic coverage

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

The genomic coverage of the Axiom Genome-Wide CHB 1 Array is shown relative to the common Han Chinese 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

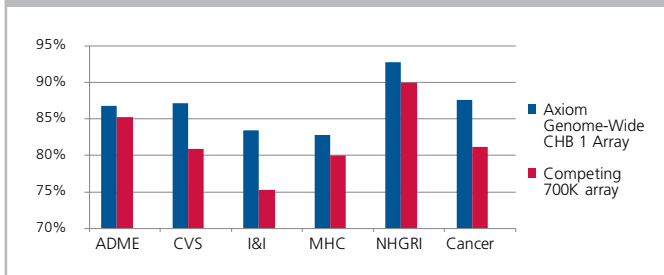
**Figure 2:** Comparison of genomic coverage of common CHB alleles between the Axiom CHB 1 Array and a competing array of 700K SNPs.



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](http://www.illumina.com).

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

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



## Ordering information

Part number	Product	Description
901764	Axiom™ Genome-Wide CHB 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

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## Assay performance

The Axiom Genome-Wide CHB 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 <dye 1> 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 CHB 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)