



Axiom™ Genotyping Solution

The Axiom Genotyping Solution is a fully automated workflow that enables you to process more than 750 samples per week on a single Affymetrix system. With the Axiom Solution, you can discover new disease associations in less time and with little effort.

Benefits of the Axiom Genotyping Solution:

- **Novel variants**
- **Highest throughput**
- **Fully automated**
- **Reliable results**

Introduction

The Axiom Genotyping Solution is Affymetrix' newest product for genome-wide association studies (GWAS) and candidate gene association studies. It includes array plates with validated genomic content, complete reagent kits, data analysis tools, and a fully automated workflow utilizing the GeneTitan™ Multi-Channel (MC) Instrument.

The Axiom Genotyping Solution includes:

- Axiom™ Genome-Wide Human Array Plates, designed to maximize genomic coverage of common and novel SNP and indels in Caucasian, Asian, and African populations
- A proprietary database of validated genomic markers, including HapMap and content from the 1000 Genomes Project, that can be leveraged as part of a custom Axiom design process
- Axiom™ Reagent Kits for automated, parallel processing of 96 samples per plate—process between 192 and 768 samples per run
- A fully automated workflow with validated robotic methods for target preparation and array processing, based on the Beckman Biomek® FX⁹ Target Prep Express System and the GeneTitan™ MC Instrument
- Genotyping Console™ Software (GTC) with a new genotyping algorithm (Axiom GT1 algorithm) for automated allele calling and quality assessment of called genotypes

Highly informative genomic content

The initial Axiom Genome-Wide Human Array contains more than 560,000 single nucleotide polymorphisms (SNPs) and more than 6,000 biallelic insertion & deletions (indels). SNPs and indels on this array were from the dbSNP database and were screened against the original 270 HapMap samples.

SNPs were selected to represent chromosomes X and Y, as well as mitochondrial SNPs, cSNPs, SNPs in recombination hotspots, ADME SNPs, miRNA SNPs, and disease-associated SNPs (see Table 1). The indels were selected to supplement the genomic coverage provided by the SNPs.

Table 1: Total number of SNPs of each class on the Axiom Genome-Wide Human Array

Chromosome X SNPs	15,790
Chromosome Y SNPs	2,025
Mitochondrial genome and miRNA SNPs	287
Disease associated	809
Total cSNPs	13,049
UTRs and splicing SNPs	9,311
MHC	7,391
ADME	4,193
Conserved regions	28,254
Genic regions	227,312
Total markers	567,096

High-quality data

To validate the performance of the Axiom Genotyping Solution, Affymetrix processed 270 samples from phase I of the International HapMap Project and 43 matched pairs of blood and saliva samples collected with the Oragene® Sample Collection Kit (DNA Genotek, Inc.). An external customer also ran our end-to-end 8-plate workflow at their laboratory using a combination of HapMap samples and their own samples, for a total of 192 samples. In addition, we have data from another set of 48 customer samples run by Affymetrix. Arrays that passed the quality control (QC) threshold were analyzed using the Axiom GT1 algorithm. The results are shown in Table 2 (page 2).



Table 2: Preliminary data analyzed with the Axiom GT1 algorithm (0.15)

Performance metrics	Specifications*	270 HapMap	8-plate workflow run by external customer	48 samples from external customer run at Affymetrix	Paired blood and saliva sample set
Avg. call rate	99%	99.7%	99.5%	99.8%	99.6%
HapMap concordance	99.5%	99.8%	Pending**	N/A [†]	N/A [†]
Reproducibility	99.8%	Pending**	99.9%	99.9%	99.9%

*Observed data may vary depending on sample preparation method and sample quality.

**Data analysis in progress [†]Not applicable

Ordering information

Reagent and array kits		
Axiom™ Genome-Wide Human Reagent Kit*	901281	Includes all reagents (except isopropanol) to process 96 gDNA samples
Axiom™ Genome-Wide Human Array Plates	901591	Includes 1 96-array plate and required GeneTitan™ plastic consumables
Axiom™ Genome-Wide Human Array Plates	901592	Includes 2 96-array plates and required GeneTitan™ plastic consumables
Axiom™ Genome-Wide Human Array Plates	901596	Includes 8 96-array plates and required GeneTitan™ plastic consumables
Services		
Axiom™ Genotyping Services	000740	Sample processing and data generation services for gDNA samples (sample minimum 1,000 Samples) using the Axiom Genotyping Solution

*Reagent kits do not include Beckman plastic consumables required to run the assay on the Beckman Biomek® FX[†] Target Prep Express System.

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P/N DS104 Rev. 1

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