Data Sheet

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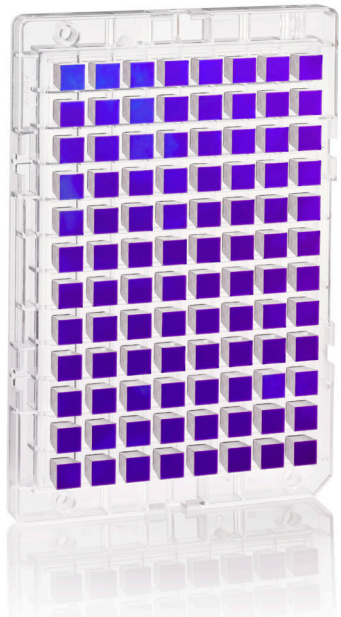
Revolutionize life

Axiom® Exome Genotyping Array Plates

The most complete exome genotyping arrays to screen functional variants for disease relevance



Axiom® Exome Genotyping Array Plates contain the most comprehensive panel to screen for putative functional variants in protein coding regions of the human genome. When combined with genotype data from genome-wide arrays, this panel of rare non-synonymous coding SNPs, insertion/deletion (indel) mutations and other key GWAS markers is a powerful tool to identify casual variants in complex diseases.

Axiom® Exome Array Plates are part of the Axiom® Genotyping Solution, Affymetrix' innovative technology for genome-wide association, replication, and candidate gene association studies.

Introduction

Axiom Exome Array Plates are based on genomic content derived from a pool of novel variants discovered in over 16 major human exome sequencing initiatives, most notably the Exome Chip Design Consortium, the NHLBI Exome Project, the Genetics of Type 2 Diabetes program (GoT2D), the 1000 Genomes Project, the Cancer Genome Atlas Project, the SardiNIA exome study, the Autism Exome Sequencing Study, the UK10K project, and others. The variants were ascertained by sequencing 12,000 ethnically diverse samples of European, African, Latino, and Asian ancestry at 20x coverage or higher and represent multiple disease cohorts, including type 2 diabetes, cancer, infectious disease, cardiovascular disease, and neurological/psychiatric disorders. A final "super set" of putative functional variants was selected from this pool if the SNP or indel was discovered in at least two separate individuals. The minor allele frequency for most of these variants is lower than 0.1% (see Table 3 for a distribution of coding SNPs by minor allele frequency).

Benefits of Axiom Exome Genotyping Array Plates:

Comprehensive content

- The most commercially available novel coding SNPs and indels from 1000 Genomes and other exome resequencing initiatives
- Over 35,000 novel single-base and complex indels (see Table 2 for a distribution of indels by size) from draft Phase 1 1000 Genomes Project exome calls

Informative markers

- Ancestry informative markers (AIMs), GWAS hit SNPs, identity-by-descent (IBD) markers, and more (see Table 1).

High-quality data

- Genotype-tested against 1,256 reference samples in the Axiom® Assay to ensure high call rate and reproducibility

Flexible format

- Customize up to 100,000 additional markers for replication and fine mapping of candidate genes

Less hands-on time

- Fully automated and fast array processing significantly reduces hands-on time, saves money, and reduces errors

Flexible format

The array plates are configured in two formats:

Axiom® Exome 319 Array Plate	
▪ High exon coverage	▪ Fits any budget
▪ Large set of novel indels	▪ Fixed format

Axiom® Exome Plus Array Plate
Includes *Axiom Exome 319 Array Plate content plus:*
▪ Flexible format up to 100,000 custom markers

Proven performance

Axiom® Exome Array Plates are genotype-tested in the Axiom Genotyping Assay by genotyping 1,256 samples from the International HapMap Project and 1000 Genomes sample collection, and performance has been evaluated against stringent quality control metrics including call rate, concordance, and

reproducibility (see Table 4 for performance metrics). The performance and genotype data has been released into the public domain and is currently downloadable from the Affymetrix website.

Analysis workflow for Axiom® Exome Genotyping Array Plates

A two-stage analysis workflow is recommended to fully utilize the content of the arrays. The goal of the first stage of the analysis workflow is to identify the samples with performance metrics that meet and/or exceed the recommended QC thresholds. These QC-verified samples are then included in the second stage of the genotyping workflow, where all markers on the arrays will be genotyped. An advanced analysis technique is then used to identify monomorphic (uninformative) markers and remove these from the analysis. The benefit of this advanced analysis is that it provides the greatest flexibility in finding the most informative content for each dataset.

For more information on how to analyze these arrays, please consult the *Best Practice Supplement to Axiom Genotyping Solution Data Analysis User Guide*, P/N 703083.

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 1: Count of markers included in Axiom Exome Array Plates in various categories.

Categories	Number of markers		
	Axiom® Exome 319 Array Plate	**Axiom® Exome Plus Array Plate**	**Competitive Exome Array**
Total validated markers	319,000	319,000	240,000
Non-synonymous coding	261,000	261,000	220,000
Splice and stop	17,100	17,100	16,400
Synonymous SNPs	4,400	4,400	4,100
Ancestry informative markers[1]	4,500	4,500	3,500
ESP SNPs[2]	860	860	810
SNPs for DNA fingerprinting	260	260	250
Identity-by-descent SNPs	5,500	5,500	3,400
GWAS tag markers	5,100	5,100	4,800
HLA region	2,300	2,300	2,100
Indels[3]	35,100	35,100	180
Chromosome X/Y	6,900/160	6,900/160	470/100
Mitochondrial DNA/miRNA	210	210	180
Customizable markers	N/A	100,000	Varies

[1] Includes SNPs from the Latino Ancestry Cancer Epidemiology Consortium
[2] SNPs derived from the NHLBI GO Exome Sequencing Project (ESP)
[3] Single-base and complex indels ascertained in the draft Phase 1 1000 Genomes Project exome calls
Numbers in tables 1, 2 and 3 are rounded to multiples of ten.

Table 2: Number of markers by indel size.

Size (bp)	Number of markers
1 bp	17,500
2–10 bp	16,400
11–50 bp	1,200
51–140 bp	18

Table 3: Distribution of coding SNPs by minor allele frequency (MAF) TT (estimated over 1,256 samples).

MAF	Number of markers
<1%	255,500
1–5%	21,300
>5%	42,200

Table 4: Performance metrics.

Metric	Specification	Actual performance[4] (n = 1,256 samples from HapMap and 1000 Genomes Projects)
Call rate	>99%	99.76%
Sample pass rate	>95%	99.8%
Reproducibility	>99.8%	99.99%
Concordance	>99.5%	99.5%

[4] Performance metrics based on genotype test against 1,256 samples from the International HapMap and 1000 Genomes reference sample collection.

Ordering information

Part number	Product	Description
902106	Axiom® Exome 319 Array Plate	Contains one 96-array plate, 319K markers per array
000838	Axiom® Exome Plus Array Plate	Contains one 96-array plate, 319K markers plus up to 100K markers per array with custom markers
901758	Axiom® 2.0 Reagent Kit*	Includes all reagents (except isopropanol) for processing 96 DNA samples
901606	Axiom® GeneTitan® Consumables Kit	Contains all GeneTitan® Instrument consumables required to process one Axiom Array Plate

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

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