## Human genetics with the Axiom® Genotyping Solution



The Axiom® Genotyping Solution is an innovative genotyping technology platform that enables optimized human genetics workflows through flexible array choices, a robust assay, and hands-free, automated array processing on the GeneTitan® Multi-Channel Instrument.

Causal genes **Novel variants Population genetics** Susceptibility loci and variants **Association and** Validation and Ancestry and evolution GWAS, replication, and fine-mapping haplotype mapping confirmation Asian Admixed/ Latin European African ancestry ancestry ancestry ancestry Rapid and cost-effective alternative to The only array designed by optimized SNP The most complete exome genotyping Coverage-Optimized World Arrays: next-generation designs combining GWAS, replication, and fine-mapping into one array. Dense array to screen functional variants for sequencing to create a haplotype map selection for unbiased population and marker coverage of disease-associated SNPs, genes, and regions, plus high whole-genome coverage of common and rare variants. in your population sample evolutionary genetics disease relevance Axiom® Genomic Axiom® EUR 1 Array Axiom® AFR 1 Array Axiom® EAS 1 Array Axiom® LAT 1 Array Axiom® Exome 319 Array Axiom® Human Origins 1 Array MAF ≥0.01 African-American genomes African with European admi MAF >0.005 Screening Service 629,000 SNPs from diverse modern human MAF ≥0.01 >300,000 rare exonic variants 5.4 million SNPs and indels >35,000 simple and complex indels The most complete exome genotyping Population-optimized arrays with high genome-wide coverage of common and rare variants. Maximum discovery power with array plus the most flexible choice to optimal cost-efficiency using highly informative markers selected by screening population samples. add your own SNPs and indels Axiom<sup>®</sup> CEU 1 Array

MAF ≥0.02 Axiom® ASI 1 Array Axiom® Exome Plus Array MAF >0.005 **Array Set** MAF ≥0.02 African genomes including Add your choice of up to 100,000 SNPs and indels YRI, LWK, MKK, ASW Axiom<sup>®</sup> CHB 1 Array Axiom<sup>®</sup> CHB 2 Array MAF 0.05-0.02 Combine validation and genotyping Cost-effective and robust variant The most flexible access to your choice of cost-effective genotyping array content with high SNP conversion and no batch-to-batch screening to eliminate false positives SNP drop-out for the best data consistency available. Our bioinformatics specialists work with you to design the array you need of candidate causal variants from using SNPs and indels from our Axiom® Genomic Database or from your own NGS discovery studies. and create robust haplotype maps your targeted NGS studies Axiom<sup>®</sup> myDesign<sup>™</sup> Axiom® myDesign™ Genome-Wide Arrays r as few as 480 samples, choose from 500,000 to 2.6 million SNPs and ind to create your own genome-wide array for GWAS or replication studies Axiom® myDesign™ Choose to add from 1.500 **Targeted Genotyping Arrays** up to 2.6 million SNPs and For as few as 480 samples, choose indels to your screening array from 1,500 to 500,000 SNPs and indels for your own targeted array Axiom® myDesign® Targeted Genotyping Arrays For as few as 480 samples, choose from 1,500 to 00,000 SNPs and indels to create your own targeted arra

## What is the Axiom Genomic Database?

The Axiom Genomic Database (AGD) is the world's largest resource of >11M genotype-tested and fully-annotated SNPs and indels. It is freely available to investigators developing Axiom myDesign Arrays and overcomes three significant challenges:

- 1. AGD has eliminated false positive SNPs from low-coverage NGS data
- AGD contains only confirmed SNPs and indels. We genotype-tested millions of SNPs from initiatives such as the 1000 Genomes Project, to remove the sequencing errors common in low coverage NGS data.
- 2. No loss of SNPs due to assay incompatibility
- AGD markers have ~100% success rate on Axiom myDesign Arrays because our genotype-testing program, in at least 270 reference samples, proves each marker works robustly with the Axiom® Assay. 3. Easy to define and build an Axiom myDesign Array SNP list:
- AGD markers are fully-annotated from more than 11 online sources for easy SNP selection, saving hours of web searching to collate your SNP list. Our bioinformatic specialists work with you to develop your Axiom myDesign Array using our online Axiom Design Center for simple designs or additional in-house expertise for complex designs.