applied biosystems

Axiom Biobank Genotyping Solution

The power of discovery is in the design

ThermoFisher SCIENTIFIC

GWAS has evolved—why and how?

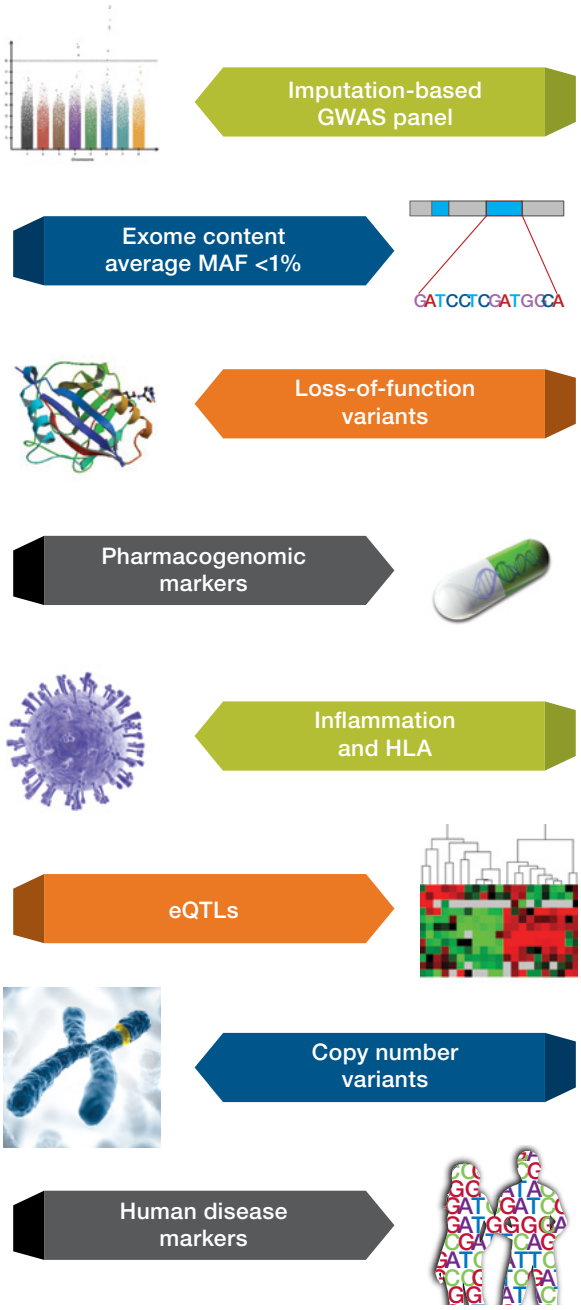
More than 2,000 genetic loci have been published from hundreds of genome-wide association studies (GWAS). In addition, next-generation sequencing has enabled extensive allele discovery in diverse global populations and in functionally important regions of the genome. This new genetic knowledge provides a foundation for the next era of genomic studies.

Investigators at the forefront of complex trait genetic studies have implemented a genotyping paradigm that does more than just cover the genome and exome. The world's leading genetic studies and biobanks are integrating knowledge of population variation, imputation analysis, epidemiology, and biological function into the design of large-scale, state-of-the-art association studies to understand how the complex interactions between genes, environment, and lifestyle relate to health.

Axiom Biobank Genotyping Arrays

Imputation-aware modular designs tailored for your trait and population studies

The Applied Biosystems™ Axiom™ Biobank Genotyping Solution is the platform of choice for large-scale genotyping studies. Scientists from many of the world's largest population genetic epidemiology projects have chosen Thermo Fisher Scientific as their partner to tailor the array design for their studies.



Choose a predesigned array or collaborate with our bioinformatics team to customize an existing design or create an entirely new array for GWAS, replication, or fine-mapping studies.

Axiom array content modules

Our novel modular content approach makes designing custom arrays fast and simple. Axiom™ array content modules exist within the Applied Biosystems™ Axiom™ Genomic Database, which covers 26 million SNPs and indels. Combine the modules into an array design, and easily modify them by adding, removing, and replacing markers to meet the needs of your study.

Exome module

- A panel of coding SNPs, indels, and other markers selected from global exome sequencing projects

Loss-of-function module

- Variants predicted to be seriously disruptive to protein function and therefore logical targets for disease and drug development

Pharmacogenomic marker module

- Variants associated with drug response include markers from the PharmGKB (**pharmgkb.org**) and the Applied Biosystems™ DMET™ Plus Array, which have been validated on the Axiom platform

Inflammation and HLA module

- Markers for genes in the HLA (chr6) region known to be important in immune response but are not easily assayed directly on other commercial arrays

eQTL module

- Expression quantitative trait locus (eQTL) variants that allow regulatory linkage with transcripts whose expression they may influence and are extremely valuable when a strong eQTL coincides with a GWAS signal

CNV module

- CNV regions thought to be related to developmental delay, neuropsychiatric disorders, and lung function
- Custom arrays can be designed to include markers targeting any CNV region of interest

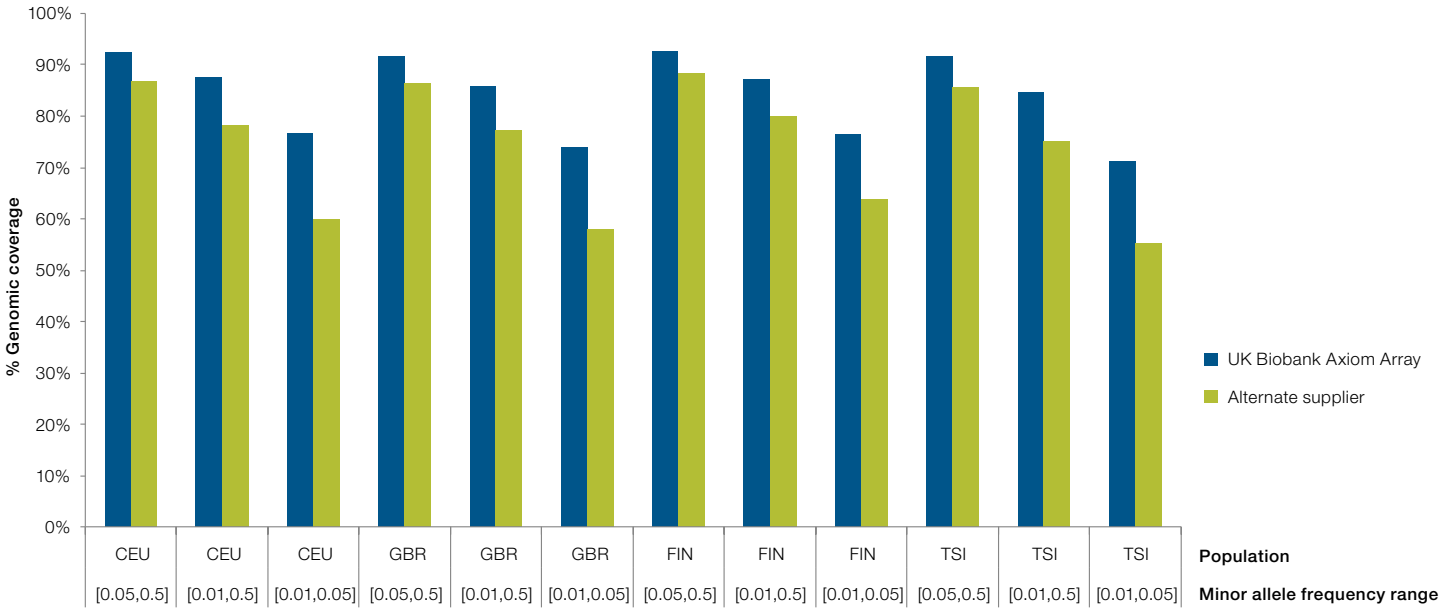
Human disease module

- Markers likely associated with Alzheimer's disease including APOE, autoimmune/inflammatory diseases, blood phenotypes, cancer common variants, cardiometabolic disorders, high blood pressure, diabetes, and asthma

Superior genomic coverage

High-coverage GWAS module

- A series of GWAS panels with superior genomic coverage for every population in the 1000 Genomes Project
- Greater imputed coverage in target populations compared with a tagSNP design approach due to utilization of advanced algorithm strategies for imputation-aware marker selection
- Marker selection methods using technology licensed from the University of Oxford
- Ability to design novel GWAS panels using your own population reference data



Genomic coverage of target populations by minor allele frequency. The Applied Biosystems™ UK Biobank Axiom™ Array offers superior genomic coverage compared to a human whole-genome plus exome array from another supplier.

The UK Biobank Axiom Array was designed by leading researchers in the fields of epidemiology and population genetics for use by the UK Biobank in their prospective genotyping study of 500,000 individuals. This array is now commercially available; for more information, visit **thermofisher.com/axiombiobank**

"There's no doubt that designing a scaffold using an imputation-aware, multimarker tagging approach is a much more efficient way to achieve coverage."

Professor Mark McCarthy, University of Oxford, UK

Accelerate your research

From discovery to translational medicine on a single platform—quickly, easily, and cost-effectively

GWAS

- Comprehensive genome-wide scans
- Find candidate associations

Replication

- Confirm putative results with different samples

Fine mapping

- Narrow down regions or gene associations
- Identify causal genes or putative causal variants associated with traits
- Gain insight into complex disease mechanisms

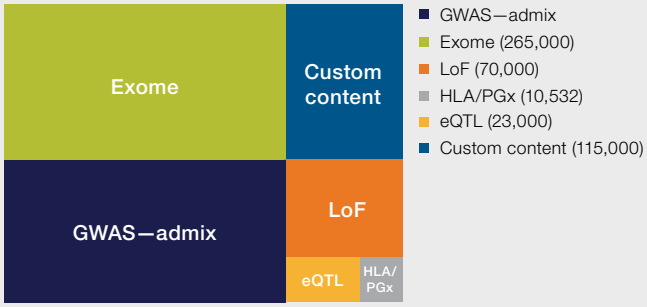
Your design. Your way. Fast.

Array customization flexibility

Customize one of our predesigned arrays by modifying content modules to fit your population or study focus.

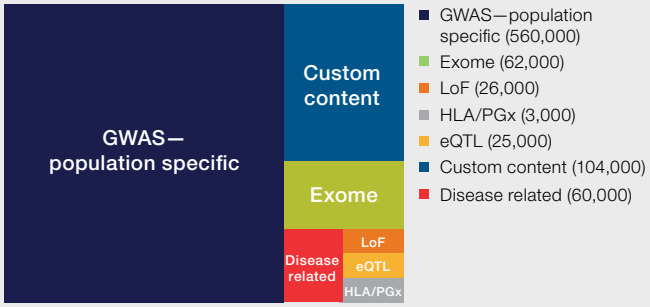
Upon design completion, custom Axiom array designs can be produced in just 4–6 weeks, which is a fraction of the time it takes from other vendors.

Axiom Biobank Genotyping Array



Content with relative number of markers in each major biological category

Example of a modified Axiom Biobank Genotyping Array



Content selected for specific study objectives

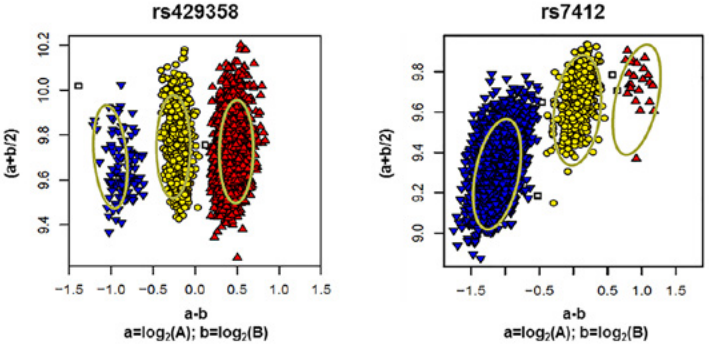
100% reproducible content

For larger longitudinal studies that extend over a significant period of time, it is important that the markers on the array do not change over the course of the project. Axiom arrays may be manufactured on demand at any time with no loss of content. Every marker requested is present on every array, every time it's manufactured.

If part of the content on an array needs to be changed, subsequent designs will always contain the exact retained subset of content from the original design, which is not possible with bead array technology.

Genotype difficult markers

Two isoforms of APOE implicated in Alzheimer's disease can be challenging to genotype and perform poorly on arrays from other manufacturers. Our advanced probe design algorithms enable successful genotyping of these markers with 100% concordance and call rates.



SNP cluster plots of APOE markers.

Axiom Genotyping Solution for human genetic research

A cost-effective, comprehensive platform for all stages of research

The Applied Biosystems™ Axiom™ Genotyping Solution is a complete and affordable solution for applications ranging from genome-wide analysis to fine-mapping studies with high accuracy and reproducibility. Its streamlined workflow and automation enable ultrahigh-throughput studies. The Axiom Genotyping Solution includes arrays with tested and reliable content from the Axiom Genomic Database and *de novo* markers important for your study.

Genotype-tested or newly discovered SNPs

Customized design consultation

Axiom predesigned and custom arrays

96 or 384 samples per array plate

Axiom target preparation solutions

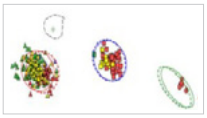
Manual and automated options available

Axiom reagent kit

Robust and reliable assay

Applied Biosystems™ GeneTitan™ Multi-Channel (MC) Instrument

Automated array processing and imaging



Genotyping Console Software and Power Tools (APT)

Primary genotyping analysis software

Experience the industry's highest level of support from design to analysis

Thermo Fisher Scientific is a leader in providing cost-effective solutions and world-class support for genotyping studies. Biobanks worldwide are partnering with us because of the distinct advantages Applied Biosystems™ Axiom™ technology offers over other technologies. Axiom technology offers superior flexibility in design, no marker loss, and a simple workflow.

Talk to your genotyping specialist about the Axiom Genotyping Solution and how our bioinformatics professionals can help you tailor an array design best suited for your study.

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Find out more at **thermofisher.com/axiombiobank**

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