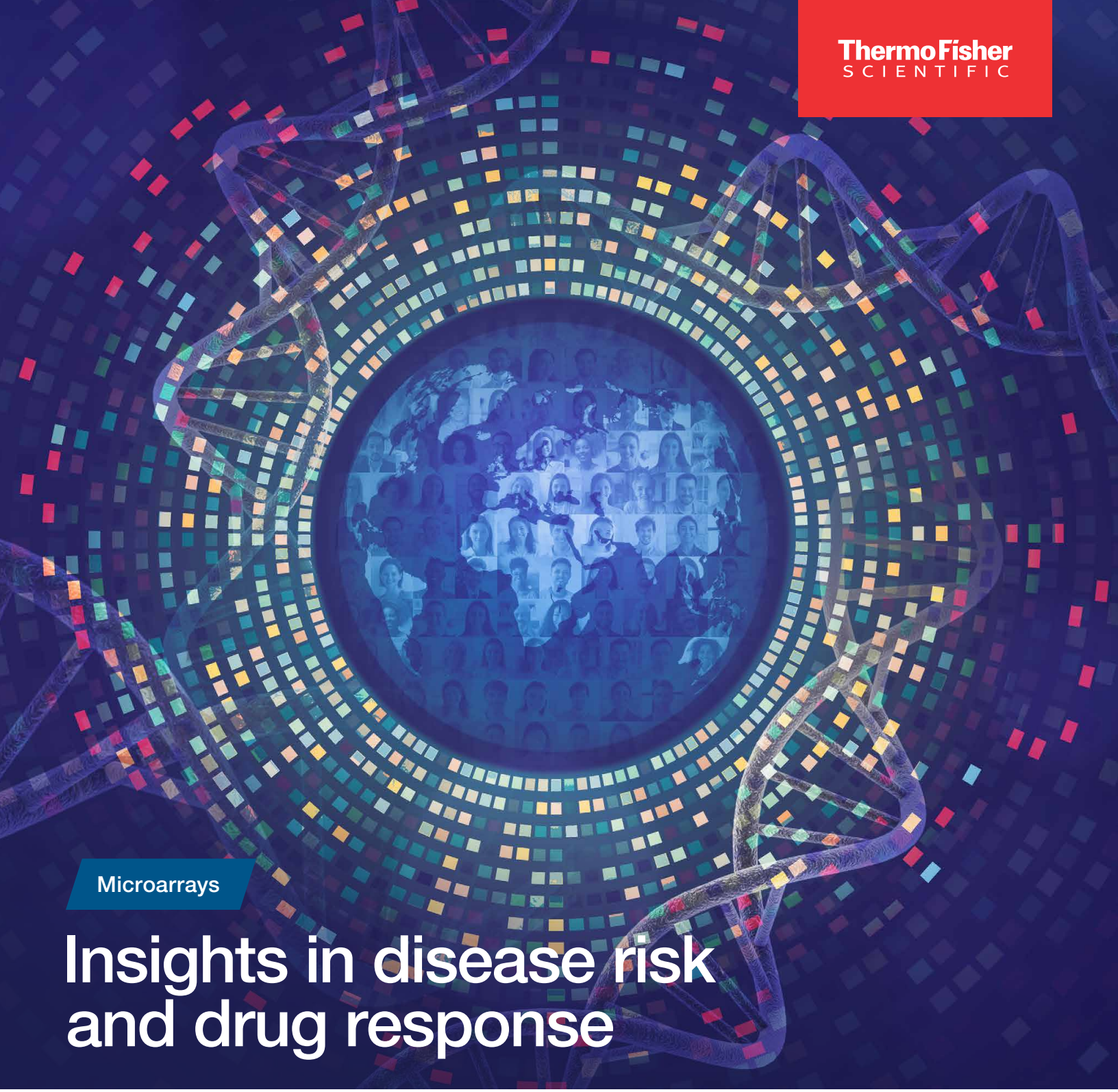


Microarrays

Insights in disease risk and drug response

Axiom genotyping arrays for human studies

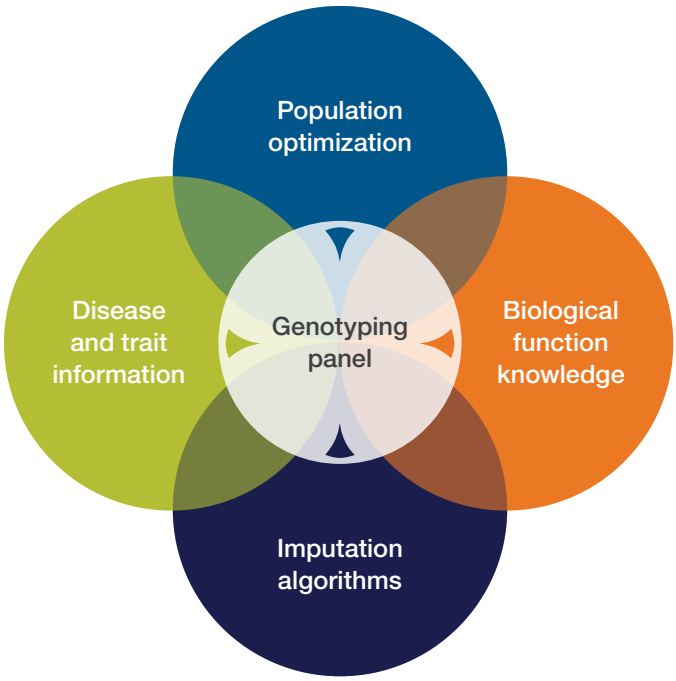
applied biosystems

The power of genotyping today

The translation of biomarkers from discovery to routine clinical applications is vital to the future of precision medicine. Yet, applying these discoveries and associating population-specific variants to individual traits, diseases, and potential downstream treatment options remains one of the most challenging barriers to personalized genomics.

While large population cohort studies and private sequencing initiatives have revealed thousands of single-nucleotide polymorphisms (SNPs) and structural variants that are implicated in nearly 2,000 human diseases and traits, many of these variants are not within genes and have no known biological function.

A better understanding of the role of these variants requires studies that are optimized for population and imputation. These studies must integrate knowledge of diseases, traits, and biological functions using meticulously designed genotyping panels and a reliable, fast, and flexible genotyping platform.



The Axiom advantage

Genotyping beyond your expectations

Maximize imputation effectiveness and accuracy

Whole-genome genotyping within large population cohorts requires great accuracy and coverage to capture population-specific mutations. While next-generation sequencing can provide novel discovery capabilities, it can be challenging due to the cost and time required to run and analyze experiments.

Imputation is the preferred solution to overcome these challenges. Applied Biosystems™ Axiom™ genotyping arrays use unique imputation algorithms that enable:

- Selection of the most appropriate markers to maximize the power of imputation
- Ability to leverage sequence data from private initiatives or the 1000 Genomes Project Phase 3 to build reference panels and array content
- Very high accuracy and coverage for one or more populations

Advanced designs for complex markers

SNPs have long been known to be a major source of genetic variation in human populations. Studies now indicate that copy number variants (CNVs), insertion and deletion polymorphisms (indels), and other structural variants also contribute toward substantial variation.

Axiom genotyping arrays interrogate SNPs and indels, while also using advanced design methods to detect even the most complex markers, including:

- SNPs in complex polymorphic regions
- CNVs
- Presence–absence variations (PAVs)
- Repetitive elements
- Markers with high or low GC content

Every SNP, on every array, in every batch

Unlike bead-based technologies, our unique photolithographic manufacturing process helps ensure that all Axiom genotyping arrays retain every marker, on every array, and in every batch, exactly as they were originally designed.

- Helps with data continuity
- No revalidation between batches
- Eliminates loss of key markers from your genotyping panel
- Enhances your ability to detect important associations, especially throughout lengthy studies or across large cohorts

Your complete human genotyping workflow

The Applied Biosystems™ Axiom™ genotyping solution is ideal for applications ranging from genome-wide analysis to fine-mapping studies. Each array design is based on specifically curated content optimized for in-depth analysis of both common and rare variants enabling a wide variety of applications, including:

- Large-scale genotyping biobank and precision medicine initiative research
- Targeted and clinical research studies including pharmacogenomics
- Cross-application utility—expand your capabilities in agrigenomics, reproductive health, SARS-CoV-2, or microbiome research

Figure 1. The Axiom genotyping solution includes arrays with genotype-tested content from the Applied Biosystems™ Axiom™ Genomic Database or *de novo* markers that are relevant to your studies. The complete solution comprises arrays, reagents, target preparation options, the Applied Biosystems™ GeneTitan™ MC Fast Scan Instrument, and free data analysis software.

For more on the GeneTitan MC Fast Scan Instrument or additional companion software modules, visit **thermofisher.com/genetitan**

Genotyping designed for you

Customized, modular designs

Applied Biosystems™ Axiom™ myDesign™ custom genotyping arrays

Designing the best genotyping panel for your studies doesn't need to be challenging or time-consuming. Axiom myDesign arrays can be tailored for specific research aims, including the discovery of new associations, confirmation and refinement of previous discoveries, and monitoring or surveillance of important known variants.

Our customization process is completely personalized to each study with rapid turnaround time for individual researchers or consortia targeting specialized content. Capture population-specific mutations by starting with our predesigned content modules, selecting from more than 13 million wet lab–tested markers in the Axiom Genomic Database or using markers from your own sequencing initiatives.

Based on the research workflow of the Axiom genotyping solution, Axiom myDesign arrays offer unprecedented scale and flexibility to design the most optimal array for your unique interests. Semi- or fully customized arrays containing 1,500 to 2.6 million markers can be designed and available in as little as six weeks after SNP lockdown.

Experience the Axiom myDesign array difference:

- Highly reliable wet lab–tested library
- Enhanced level of support in design of your array
- Ease of scalability—no project is too big or too small
- Trusted bioinformatics expertise
- Up-to-date knowledge of research guidelines including ACMG, CPIC, ClinVar, and Pharmacogenomics Knowledge Base (PharmGKB™)
- Over 30 years of microarray expertise partnering with some of the largest population studies in history

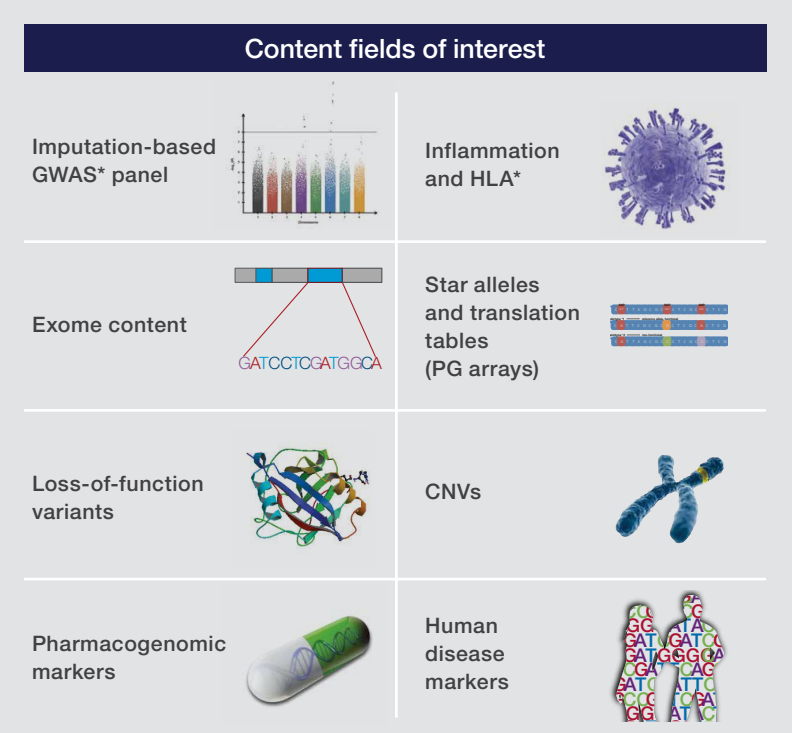


Figure 2. Identify critical genomic associations by selecting markers ideally suited for your study from a range of predesigned content modules.

* GWAS = genome-wide association study; HLA = human leukocyte antigen.

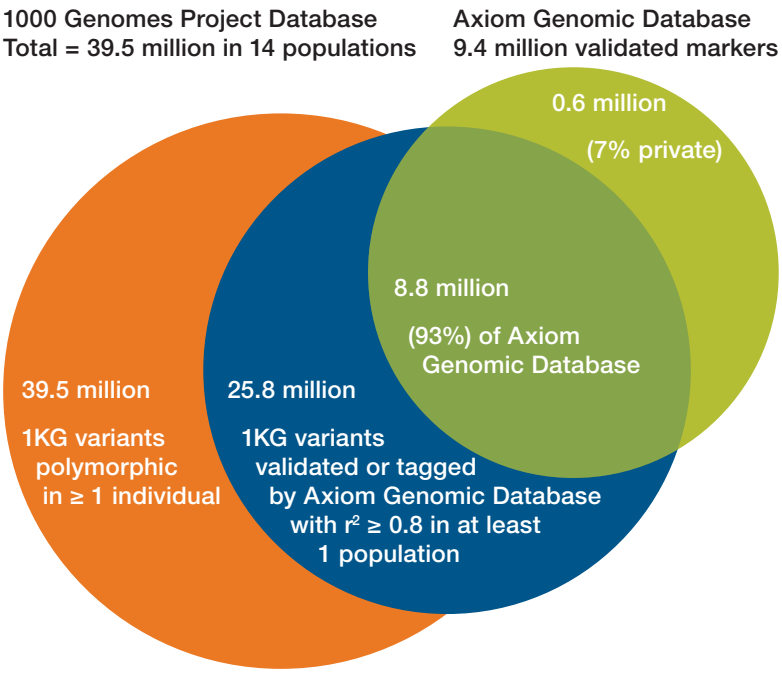


Figure 3. Graphic representation of Axiom Genomic Database.

Microarray Research Services Laboratory

If you are seeking additional support in processing your sample volume, look no further than the Applied Biosystems™ Microarray Research Services Laboratory (MRSL). Our microarray research lab offers affordable genotyping services for large- and small-scale microarray studies. With one of the largest microarray capacities in the world, the MRSL generates high-quality data and enables academic and research institutions like yours to complete your population studies without increasing your lab footprint, hiring or training personnel, or purchasing additional instruments. Our quality data analysis helps provide better results, supporting publication and grant submission.

Beyond basic sample processing, the MRSL acts as an extension or replacement for your own microarray testing facility, taking the same care and attention to detail that you would. It actively detects potential issues in your samples through a multistage sample QC process, adjusting and retesting to optimize your results, at no cost to you. With a behemoth of historical testing data and frequent review by bioinformatics professionals, we deliver reliable and repeatable results with the capability to reanalyze and deliver your data as needed.

If you would like to place a human or animal genotyping order with the Microarray Research Service Lab, please contact us via email at bioinformaticsservices@thermofisher.com.

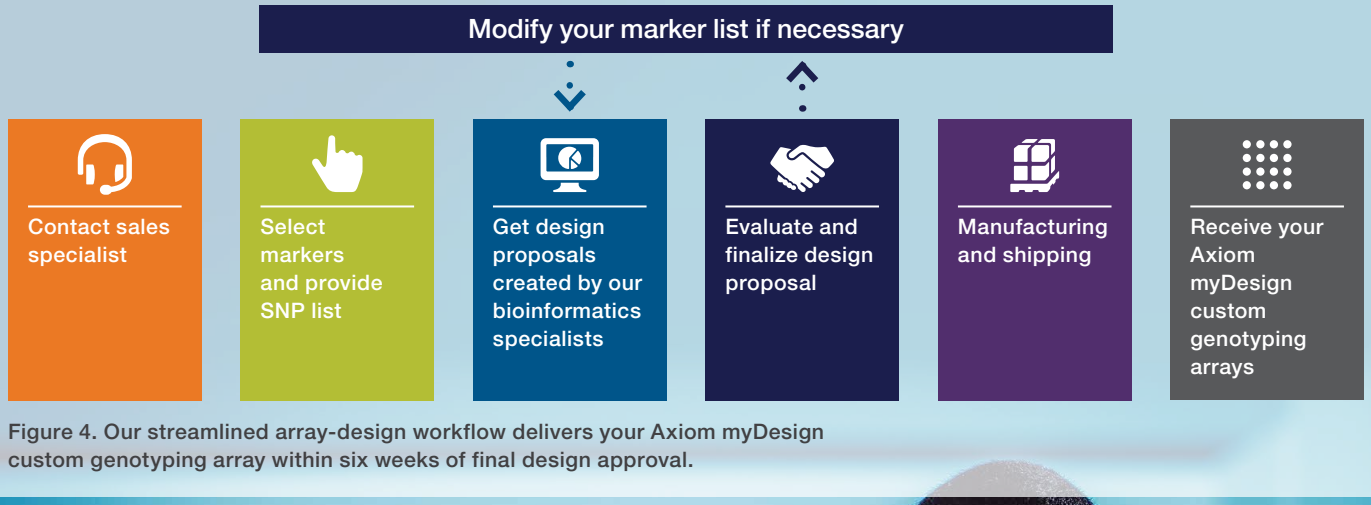
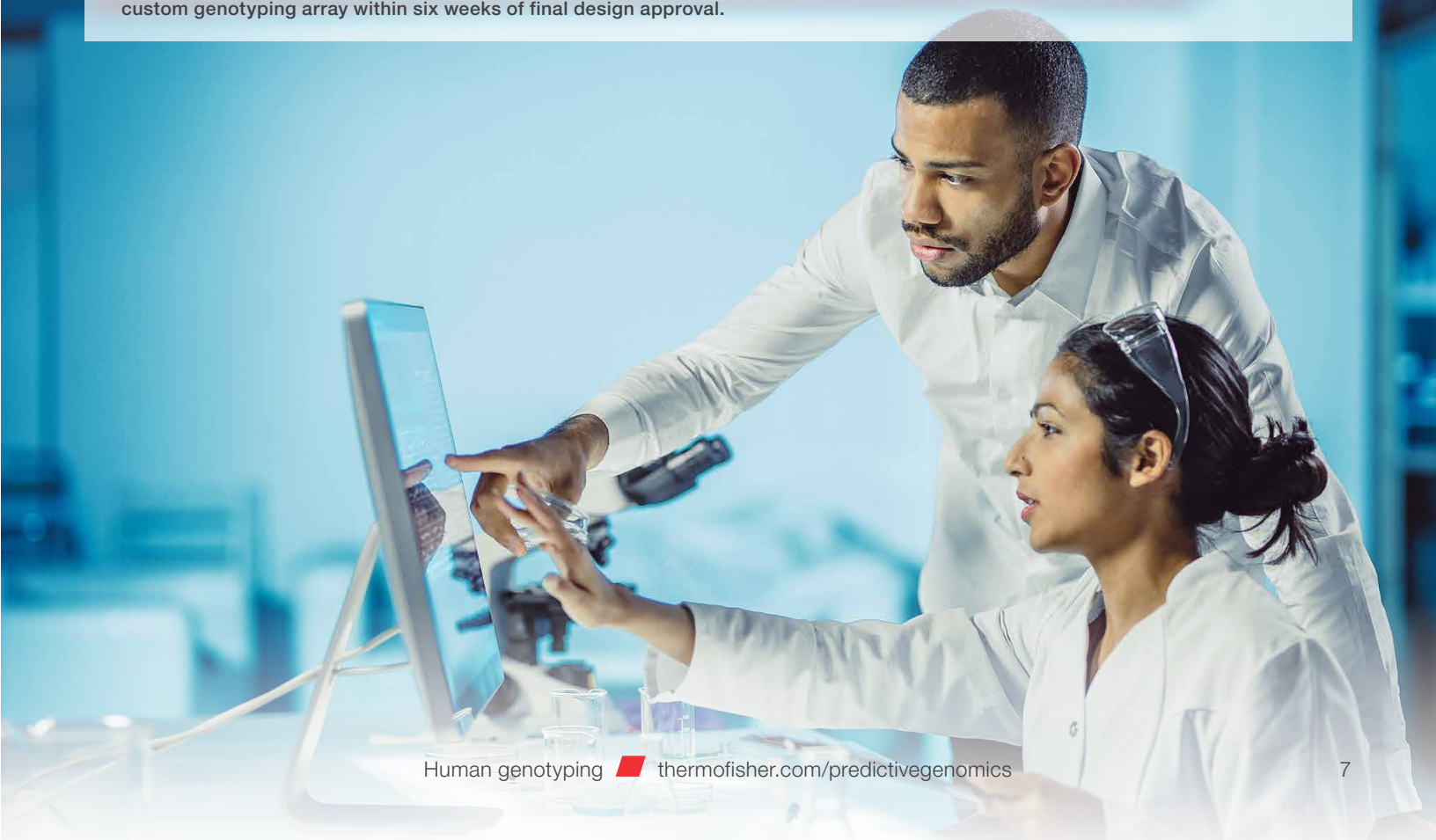


Figure 4. Our streamlined array-design workflow delivers your Axiom myDesign custom genotyping array within six weeks of final design approval.



From population screening to targeted disease research

Predesigned genotyping arrays

Imputation-aware arrays

Genotyping panels that maximize imputation accuracy and include population-specific markers with high coverage of both common and rare variants are vital to genotyping studies today. Identify population-specific associations for better understanding of complex diseases with Applied Biosystems™ Axiom™ population-focused arrays.

Human research arrays

Maximize productivity with focused investigation of clinically relevant variants associated with complex disease susceptibility, preemptive pharmacogenomics, wellness, and lifestyle. The Applied Biosystems™ Axiom™ Precision Medicine Diversity Research Array (PMDA) is ideal for studies investigating the relationship between genetics and susceptibility to complex diseases in diverse populations. The Applied Biosystems™ Axiom™ Precision Medicine Research Array (PMRA) provides broad coverage across all major ancestral populations. The Applied Biosystems™ Axiom™ Asia PMRA is curated specifically for coverage of South and East Asia populations.

Axiom PangenomiX Array

The Applied Biosystems™ Axiom™ PangenomiX Array is a comprehensive imputation-aware genotyping resource designed to advance population genomics studies by driving deeper scientific insights into complex disease susceptibility, pharmacogenomics (PGx), and genetic factors underlying wellness and lifestyle in diverse populations. It offers >800,000 markers selected for high genomic coverage from the 1000 Genomes Project Phase 3. Additional gene variants were chosen from broadly referenced public databases, including ClinVar, NHGRI-GWAS catalog, CPIC, PharmGKB, ACMG, and PharmaADME.

The Axiom PangenomiX Array is the most comprehensive and ethnically diverse genotyping microarray in the Axiom portfolio. This array is ideal for biobanking, longitudinal cohort studies in precision medicine initiatives, clinical and translational research, and clinical trials in drug discovery.

The Axiom PangenimiX Array is designed with:

- A high-coverage and high-accuracy imputation grid
- Pathogenic content from publicly annotated databases such as ClinVar and American College of Medical Genetics and Genomics
- Inflammation, blood, and HLA markers
- Immunity markers and SARS-CoV-2 research
- Pharmacogenomics markers from CPIC and PharmGKB
- Fixed-region copy number detection

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

UK Biobank Axiom Array

The Applied Biosystems™ UK Biobank Axiom™ Array was designed in collaboration with the UK Biobank to investigate the contributions of genetic predisposition and environmental exposure to the development of disease. This array is ideal for genotyping large sample cohorts from large collections maintained at biobanks and genome centers.

- Leverage imputation to identify disease-related variants
- Achieve exceptionally high coverage of common and rare variants within specific populations

Clinical research in action: Preemptive pharmacogenomics and carrier screening

Preemptive pharmacogenomics arrays

When genotyping programs evolve into routine clinical research applications, your array requirements may also evolve. Optimizing your genotyping arrays with targeted clinical research content can simplify data analysis and enable more efficient scale-up.

Gain broader insights into genetic variability associated with drug absorption, distribution, metabolism, and excretion (ADME). The Applied Biosystems™ PharmacoScan™ solution screens for both low- and high-evidence markers to enable more comprehensive genotyping for potential drug response. The Applied Biosystems™ Axiom™ PharmacoFocus™ solution enables targeted genotyping of high-evidence SNP, CNVs, and HLA variants.

PharmacoScan solution

The PharmacoScan solution includes all of the known key ADME genes for comprehensive pharmacogenomics analysis. It is ideal for researchers who are interrogating genetic variations involved in the pharmacokinetics of commonly prescribed medications.

- Preemptively screen for pharmacogenomic risk factors
- Interrogate SNPs, indels, and CNVs in a single assay
- Stratify clinical research trial populations
- Make convenient use of the star allele and translation tables included in the Applied Biosystems™ Axiom™ Analysis Suite software

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

Axiom PharmacoFocus solution

The Axiom PharmacoFocus solution was developed to accelerate pharmacogenomic research in labs, academic hospitals, health care centers, and the pharmaceutical industry. It offers comprehensive coverage of high-evidence functional variants (PharmGKB clinical annotation levels of evidence 1A–2B) that influence ADME of commonly prescribed medications.

- 2,000 markers in 150 ADME genes covering population diversity
- 88% coverage of targeted, high-evidence pharmacogenomic research markers (PharmGKB clinical annotation levels 1A–2B)
- A single array developed and optimized for high-accuracy genotyping, CNV detection, and HLA typing

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

Carrier screening arrays

Accelerate your preconception carrier screening research with high-throughput detection of sequence and structural variations for inherited diseases across a wide range of ethnicities. The Applied Biosystems™ CarrierScan™ 1S Assay Kit consolidates multiple copy number and genotyping tests into a single molecular assay with simple data analysis and reporting software.

Additional predesigned, application-specific Axiom solutions are available, including microbiome, transplant, and SARS-CoV-2 research arrays. Discover more about our time-saving catalog arrays or how you can develop your own research array at **thermofisher.com/microarrays**

Medication management reports

We have partnered with Coriell Life Sciences to integrate genotyping results into Coriell's medication management reporting system. Coriell's Genetic Response Report identifies genetic variants informative for medication efficacy, safety, and dosing. By incorporating into the system a study participant's genetics, lifestyle, and health history, a researcher can be informed of potential drug–drug interactions and treatment options under the appropriate Institutional Review Board (IRB) approval process followed by your institution.

How will you adopt predictive genomics?

Discover what we can do for you

Whatever your needs, you'll find that Thermo Fisher Scientific offers you significant advantages because our technologies are:

- **Population-optimized:** You can optimize our genetic technologies for specific ethnic populations, depending on the participants you want to include
- **Proven:** Many of the world's most significant genomic and biobanking studies have used our genetic technologies
- **Relevant:** You can access relevant genomic content and build a unique research data asset owned by you
- **Scalable:** Whether you need to generate hundreds of thousands of genetic markers or just a few, we have technologies to fit the needs of your initiative
- **Economical:** Our genotyping solutions strike the right balance between content and price point, for initiatives both large and small
- **Accurate:** Enables high-quality, consistent data by processing multiple samples under identical conditions

Speak to one of our specialists for a detailed review of what we can offer you. You can also learn more about our solutions at thermofisher.com/predictive-genomics

Simple, automated data analysis

The Axiom Analysis Suite and Applied Biosystems™ Analysis Power Tools (APT)

The Axiom Analysis Suite is an intuitive, powerful Microsoft™ Windows™ operating system–based software package for analyzing data from all Axiom genotyping arrays. In addition, APT software enables command-line control for multiple operating systems. Both packages integrate SNP and indel genotyping with visual simplification of genotyped markers into recommended categories, so you can easily distill the most informative conclusions from your studies.

- Automated genotyping calls and quality-control metrics, and filtering of SNPs into defined classifications
- Fine-mapping and post-GWAS causal variant analysis support disease and pathway research
- Customizable data visualization tools
- Ability to convert data to long format for seamless integration with current bioinformatics pipelines using Applied Biosystems™ Axiom™ Long Format Export (AxLE) Tool

Applied Biosystems™ Automated Axiom™ Analysis Suite software

This software package enables analysis of the CEL files upon scan completion of an Axiom array plate without user intervention. Upon completion of plate scanning, a notification is sent to the application server core and sample files are automatically uploaded to a designated folder.

Find out more about our additional companion software modules at thermofisher.com/microarrays

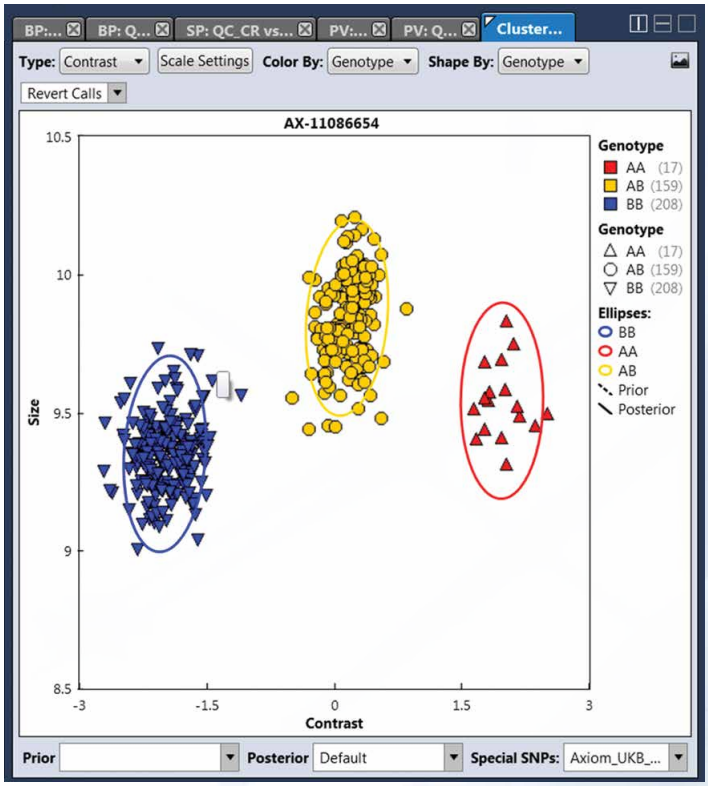


Figure 5. Easy, quick analysis and visualization of your genotyping data using Axiom Analysis Suite software.

Supporting your productivity and efficiency

Comprehensive, experienced service and support

Comprehensive instrument warranty

Our factory-trained and certified field service engineers (FSEs) strive to deliver workmanship of the highest quality. Your warranty covers repair costs, including engineer time and travel, during the entirety of your contract.

Service and support plans

We provide complete postwarranty support with our professional consulting services to help you maintain productivity, maximize the value of your investment, and optimize performance. With a service and support plan,* you can have lower, predictable operating costs and more running time.

- Flexible and configurable support solutions
- Optimum reliability with scheduled preventive system maintenance
- Optimum workstation performance and the latest software updates
- Lower and more predictable operating costs

How to reach us

To find your support or technical support team, go to thermofisher.com/contactus

For product FAQs, protocols, training courses, and webinars, go to thermofisher.com/technicalresources

* Plans vary by region.

Genotyping for the future

As we learn more about the implications of genetic variation in human health, genotyping as a molecular tool in research and clinical research settings is as important as ever. Scientists are advancing the promise of precision medicine by integrating information about genomic variation with phenotypes and other clinical research data in their genotyping studies. The insights that these studies yield already enable more precise genetic profiling of patient risk, prognosis, immune response, and response to therapy.

Whether you are involved in a large-scale biobank or precision medicine–research genotyping studies or you are implementing pharmacogenomic knowledge in your clinical research, the Axiom genotyping solution can deliver exactly the content you need for as long as you need it.

More informative insights today

Go beyond your expectations with a genotyping platform that offers highly accurate imputation, designs for complex markers, long-term content fidelity, and simple analysis.

Find out more about the Axiom genotyping solution at **thermofisher.com/microarray**