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# Genotyping beyond your expectations

## Axiom genotyping arrays for human studies

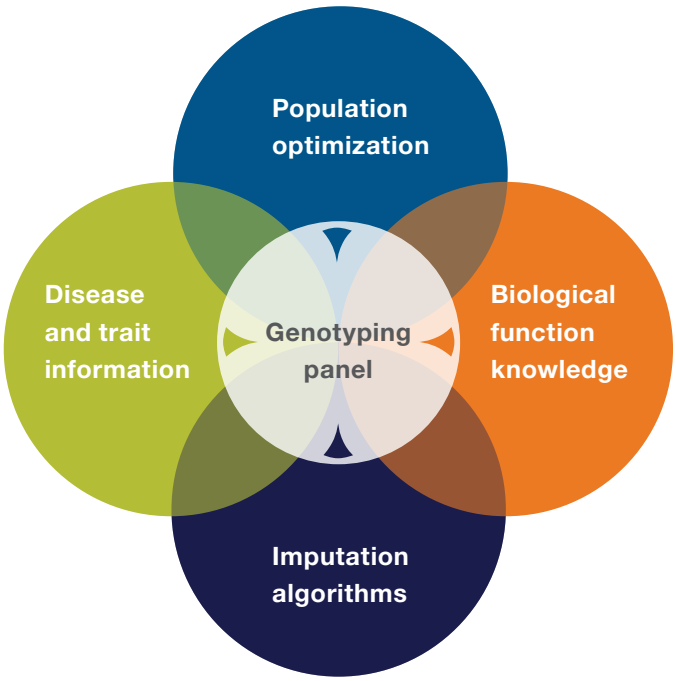
ThermoFisher SCIENTIFIC

# 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.

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 1,000 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 ensures that all Axiom genotyping arrays retain every marker, on every array, and in every batch, exactly as they were originally designed. This helps to ensure:

- Data continuity and no revalidation between batches
- No loss of key markers from your genotyping panel
- Better 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
- Direct-to-consumer applications such as ancestry and wellness



**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™ Multi-Channel (MC) Instrument, and free data analysis software.

# 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 Custom Genotyping Arrays offer extensive customization options with fast turnaround times for individual researchers or consortia who need 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.

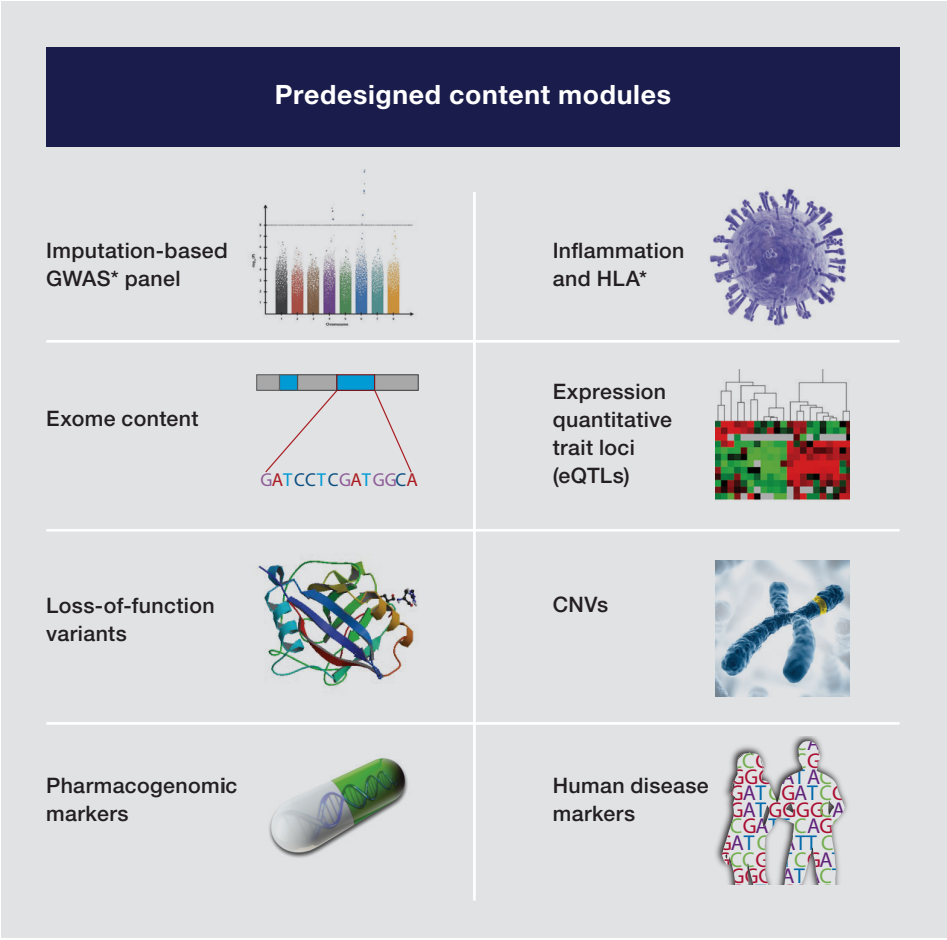



**Figure 2. Identify critical genomic associations by selecting markers ideally suited for your study from a range of predesigned content modules.**

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


Figure 3. Our streamlined array-design workflow delivers your Axiom myDesign Custom Genotyping Array within six weeks of final design approval.

# From large populations to targeted studies

## Predesigned genotyping arrays

### Population-focused 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 our population-focused arrays.

**Applied Biosystems™ Axiom™ Precision Medicine Research Array and Axiom Asia Precision Medicine Research Array**

The Axiom Precision Medicine Research Array (PMRA) and Axiom Asia Precision Medicine Research Array (Asia PMRA) are tailored for studies across multiple populations and subpopulations. They are ideal for longitudinal cohort studies in precision medicine research, biobanking, translational research, and drug discovery clinical trials.

- Investigate the relationships between genetics and susceptibility to complex diseases, immune responses, pharmacogenomics, and other disease-related applications
- Leverage imputation for extensive multipopulation coverage
- Investigate common and rare variants
- Customizable with novel content

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

**Applied Biosystems™ UK Biobank Axiom™ Array**

The 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

### Arrays for clinical research

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.

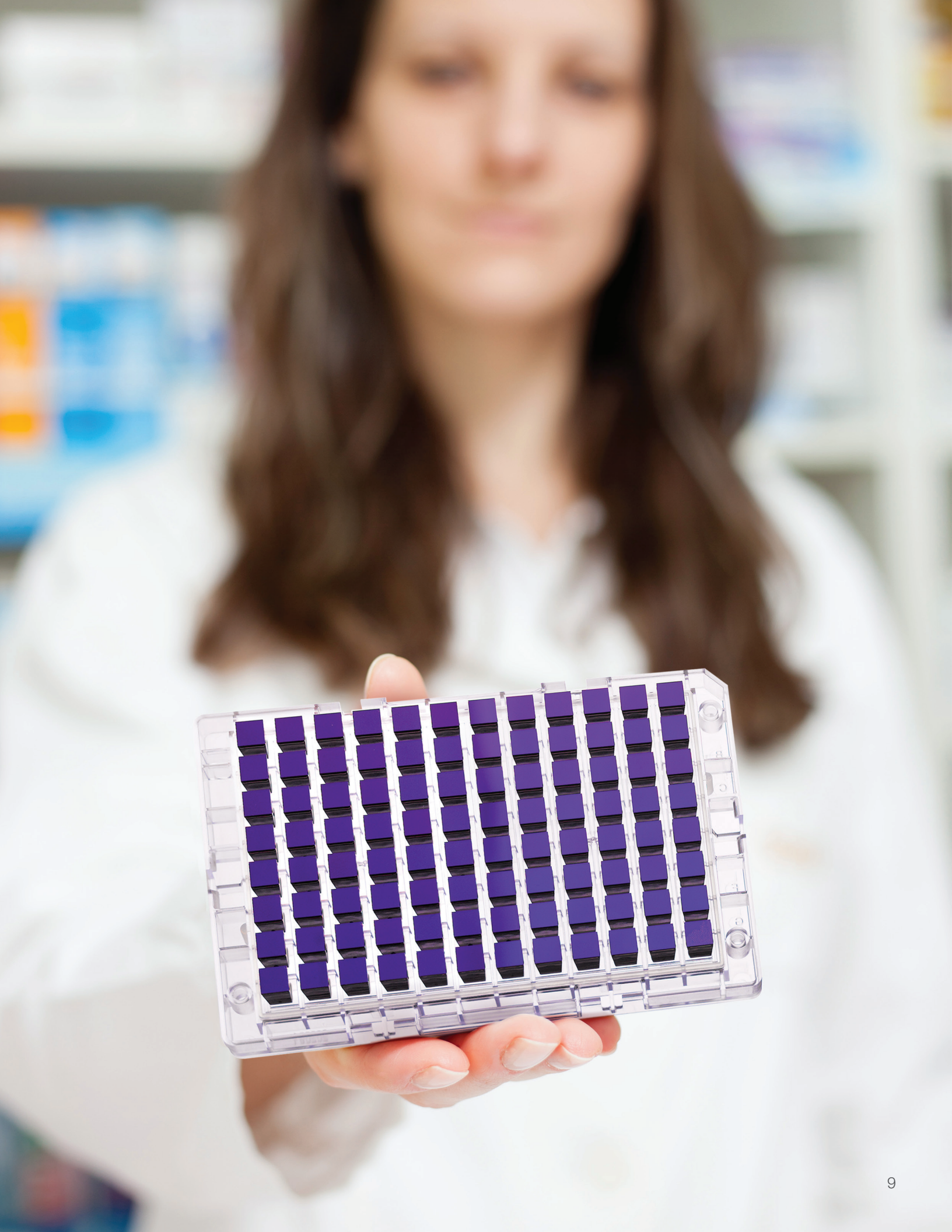
**Applied Biosystems™ PharmacoScan™ Solution**

The PharmacoScan Solution includes all of the known key absorption, distribution, metabolism, and excretion (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 trial populations
- Make convenient use of the star allele and translation tables included in the Axiom Analysis Suite software

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

Find out more about our application-specific predesigned arrays for transplant and microbiome research at **thermofisher.com/microarrays**.



# Simple, automated data analysis

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

Axiom Analysis Suite is an intuitive, powerful Microsoft™ Windows™ operating system-based software package for analyzing data from all Axiom genotyping arrays. In addition, Axiom Power Tools (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

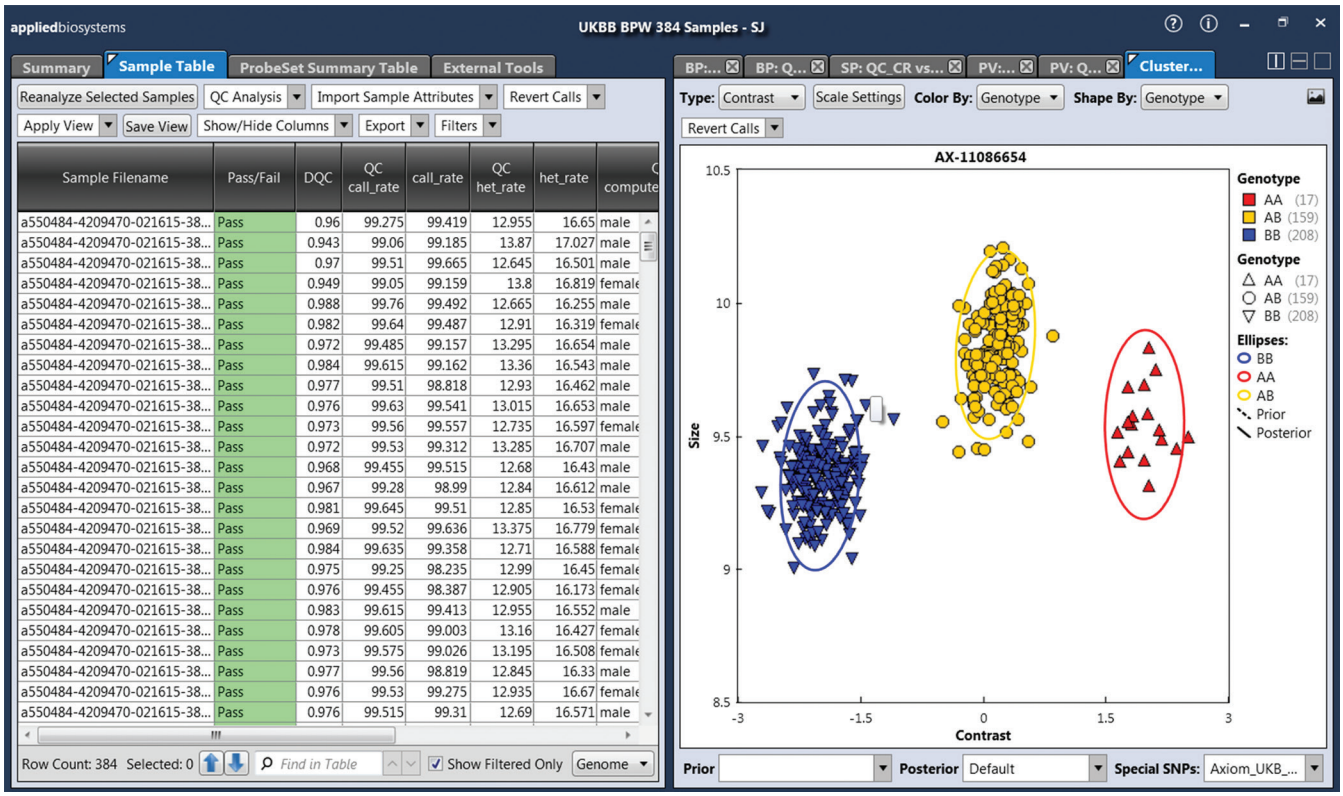



**Figure 4. Easy, quick analysis and visualization of your genotyping data using Axiom Analysis Suite software.**

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

# Increase your productivity and efficiency

## Comprehensive, experienced service and support

### Comprehensive instrument warranty

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

### Service and support plans

We provide complete post-warranty 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 preventative system maintenance
- Optimum workstation performance and the latest software updates
- Lower and more predictable operating costs

### How to reach us

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

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

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## 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 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, are aiming to create a better genetic ancestry product, or 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/genotyping**

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