genomics

applied biosystems ion torrent

Human genotyping solutions

Genotyping technologies to suit your needs

ThermoFisher SCIENTIFIC

Robust and reliable human genotyping solutions tailored for you

Research into the genetic variation of humans has expanded our understanding of evolution, provided insight into common and rare diseases, accelerated the pace of drug development, and paved the way toward the future of precision medicine. Scientists employing genotyping analysis are aiming to untangle the often complex relationships between genotype and phenotype in studies focusing on single-nucleotide polymorphisms (SNPs), insertion or deletion polymorphisms (indels), and copy number variants (CNVs).

We offer a comprehensive portfolio of genotyping solutions for SNP, indel, and CNV analysis that support all stages of your workflow, from large-scale discovery to targeted use in routine applications. We have a proven track record of partnering with scientists in order to understand their specific needs and recommend the best product and content for their project.

Don’t be limited by your technology—we’re here to help you find the best genotyping tool for your study, no matter the number of markers or number of samples.

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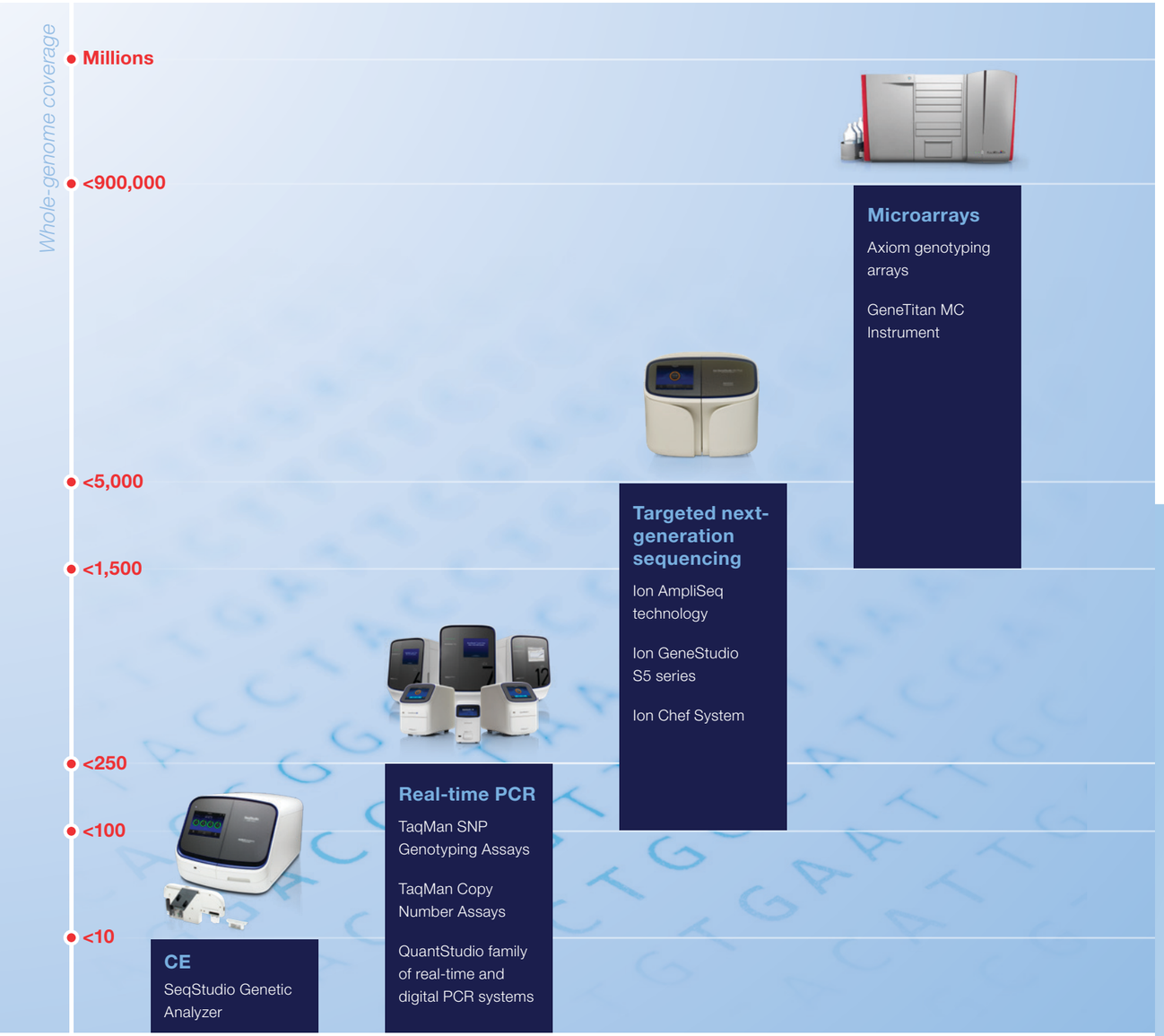
Our comprehensive portfolio of human genotyping solutions



We offer a wide range of accurate and robust technologies to facilitate human genotyping research. Whether you are starting a large-scale genetic epidemiology study, or simply looking to confirm the identity of a few markers, our expertise and comprehensive portfolio of tools can help you get there quickly and efficiently.

Genotyping microarrays

Axiom genotyping solution

Used in many of the world's largest genetic studies, our portfolio of array-based genotyping solutions are ideal for everything from genome-wide analysis to routine screening of complex genetic traits. Applied Biosystems™ Axiom™ genotyping arrays offer high imputation accuracy and flexible modular designs, and they assure 100% consistency of content.

Choose from a variety of predesigned arrays, modify an existing design, or create a completely custom array with exactly the content you need. We partner with you to help ensure you have the best genotyping panel for your studies.

Key features:

- Ability to interrogate indels and candidate SNPs that have high GC content or interfering SNPs in the flanking sequences
- Designs that offer better imputed genotype accuracy
- 100% consistent manufacturing process, ensuring every SNP is on every array, every time
- Scalable, high-throughput formats for medium-to-high-density genotyping applications
- Fully automated workflow that can process up to 8 array plates per week with a single Applied Biosystems™ GeneTitan™ Multi-Channel (MC) Instrument
- Powerful yet simple data analysis software included at no extra cost

Axiom genotyping solution includes arrays with genotype-tested content from the Applied Biosystems™ Axiom™ Genomic Database or *de novo* markers important to you. The full solution comprises arrays, reagents, automated and manual workflows, and simple, free data analysis software.

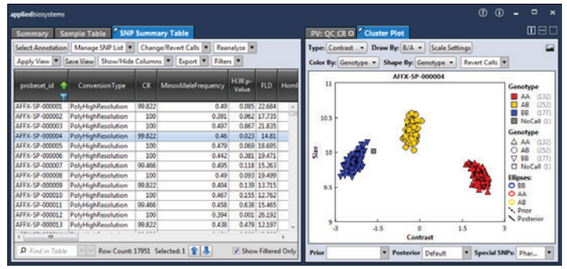
Axiom myDesign Custom Genotyping Arrays

With hundreds of millions of human SNPs and CNVs to choose from, building a bioinformatics pipeline and picking the right markers for your array design can be a challenge; but it doesn't have to be.

The Applied Biosystems™ Axiom™ myDesign™ Custom Genotyping Array program offers a fast, affordable way to create custom arrays without a large upfront order commitment. At last, custom genotyping arrays are accessible to individual researchers as well as large consortia. Partner with our bioinformatics team to design arrays with relevant content for your specific studies.

Key features:

- Get started easily with a low initial order commitment (as few as 480 samples)
- Get a great array design fast with support from our highly knowledgeable bioinformatics team
- Start sooner with delivery of new custom arrays approximately 6 weeks after final design review
- Reorder custom arrays for as few as 192 samples to complete your study



Axiom Analysis Suite

Applied Biosystems™ Axiom™ Analysis Suite software is a free, easy-to-use software package that integrates SNP genotyping, indel detection, and off-target variant (OTV) calling in an easy-to-use graphical interface.

Key features:

- Automated software generates genotyping calls and quality-control metrics, and filters SNPs into defined classifications
- View your data with customizable visualization tools
- Convert data to long format for seamless integration with current bioinformatics pipelines using the Applied Biosystems™ Axiom™ Long Format Export (AxLE) Tool
- Use optional, application-specific companion modules for further data analysis such as Applied Biosystems™ Axiom™ CNV Summary Tools Software and Axiom™ HLA Analysis Software

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

Targeted genotyping by sequencing

Ion Torrent next-generation sequencing (NGS) systems

The Ion GeneStudio™ S5 series systems comprise a line of NGS systems designed to enable a broad range of targeted NGS applications with superior speed and scalability. Our five-sequencing chip format enables both small and large-scale projects without the need to change platforms. Ion GeneStudio S5 series systems, together with Ion AmpliSeq™ technology for target selection, Ion Chef™ System for automated library and template preparation, and Torrent Suite™ Software and Ion Reporter™ Software for push-button data analysis, enable you to streamline your workflow and focus on finding meaningful insights.

Key features:

- **Scalable**—a single platform with five different sequencing chip types achieves 2–260M reads per day
- **Simple**—DNA to data with less than 45 minutes of hands-on-time using the Ion Chef system
- **Speed**—get results in as little as 24 hours (with 2.5–4 hour sequencer run times) using the Ion GeneStudio S5 Prime System—the fastest turnaround of any benchtop sequencer
- **Small sample input**—as little as 1 ng of FFPE DNA with Ion AmpliSeq technology
- **Simplified data analysis**—end-to-end bioinformatics with Torrent Suite Software and Ion Reporter Software

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

Ion AmpliSeq technology

Since its launch, Ion AmpliSeq technology has empowered researchers by enabling a simple, scalable, and fast targeted NGS workflow for detecting gene targets or hotspots. Based on ultrahigh-multiplex PCR, this unique target-selection technology helps you achieve sequencing success with as little as 1 ng of input DNA, making sequencing of formalin-fixed, paraffin-embedded (FFPE) or highly degraded samples routine on the Ion GeneStudio S5 systems.

Key features:

- Lowest DNA input requirement for targeted NGS
- 1–100s of gene targets in a single run
- Fully customizable to detect all major classes of mutations, including STRs, SNPs, mtDNA, indels, CNVs, and fusions

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

Real-time and digital PCR for genotyping

Genotyping by real-time PCR is a well-known and reliable approach that is widely used in both research and industrial settings for the confirmation of SNPs and CNVs and to screen panels of markers in hundreds or even millions of samples. We offer real-time PCR instruments, Applied Biosystems™ TaqMan® Assays, and reagents that are designed to generate reliable results for validation and screening. TaqMan Assays are available for use with 96-well, 384-well, and Applied Biosystems™ OpenArray™ real-time PCR formats so you can select the workflow that best fits your genotyping needs.

QuantStudio family of real-time and digital PCR systems

With superior flexibility, connectivity, speed, and precision, there is an Applied Biosystems™ QuantStudio™ system that is right for your research. Contact a sales representative to personalize a solution to meet your needs, or use our online product configuration tool to easily configure a system that's best for you.

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

Overview of the Applied Biosystems™ workflow for genotyping by qPCR

Prepare samples	Select assays	Set up reactions	Run PCR	Analyze data
We provide multiple solutions to isolate and purify DNA. The Applied Biosystems™ DNA Extract All Reagents Kit provides PCR-ready DNA from a wide variety of samples in about 5 minutes.	Choose from predesigned or custom assays for a wide variety of applications. Assays are available in multiple formats: single tubes, 96- and 384-well plates, 384-well microfluidic cards, and OpenArray plates.	Prepare a reaction plate using TaqMan Assays and TaqMan® ProAmp™ Master Mix, along with your DNA samples. No additional reagents required.	Amplify DNA and achieve allelic discrimination by running the PCR reaction on an Applied Biosystems™ real-time PCR system and/or thermal cycler.	Analyze and interpret real-time PCR data with intuitive software tools including TaqMan® Genotyper Software or our cloud-based Genotyping app available on Thermo Fisher Connect.

Predesigned and custom TaqMan SNP Genotyping Assays

These assays provide a highly flexible technology for the detection of SNPs. Find a predesigned assay or create your own by submitting target sequences to our secure assay design pipeline using the Applied Biosystems™ Custom TaqMan® Assay Design Tool. Our bioinformatics pipeline has successfully generated millions of assay designs by utilizing heuristic rules deduced from both manufacturing and assay performance data.

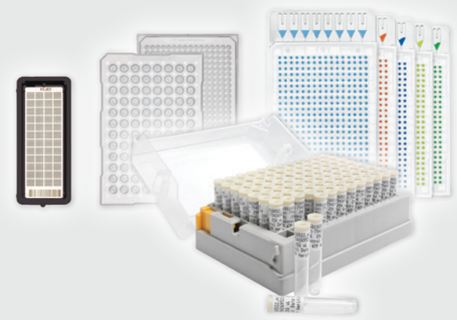
All predesigned TaqMan Assays are covered by the TaqMan Assays qPCR Guarantee.* Find out more at **thermofisher.com/taqmanguarantee**

* Subject to terms and conditions.

Create your own custom assays by submitting confidential target sequences to our secure assay design pipeline at **thermofisher.com/taqmansnpdesign**

Predesigned and custom TaqMan Copy Number Assays

Applied Biosystems™ TaqMan® Copy Number Assays provide specific, reproducible, and easy-to-interpret copy number results. TaqMan Copy Number Assays are an ideal validation tool for microarray or NGS follow-up studies and can be used to find specific targets. The workflow can be automated so that several hundred to thousands of samples can be processed in a single day.



Key features of TaqMan SNP Genotyping and Copy Number Assays:

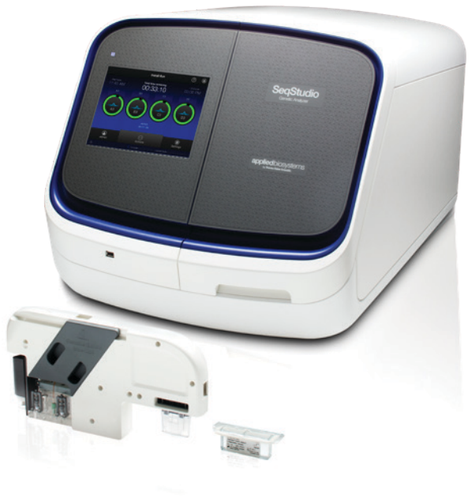
- **Proven**—gold-standard TaqMan chemistry with MGB probes and robust assay designs deliver accurate, reproducible, and reliable results
- **Easy**—convenient single-tube format and simple workflow provide an easy path to trusted results—no optimization required
- **Relevant**—extensive collection of predesigned human assays and our custom assay design tools offer direct access to content that is relevant to your research
- **Tested**—all human TaqMan SNP Genotyping Assays are functionally tested to ensure allelic discrimination

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

Capillary electrophoresis and fragment analysis

SeqStudio Genetic Analyzers

Applied Biosystems™ SeqStudio™ Genetic Analyzers provide the same workflow and trusted technology—now with an innovative, all-in-one cartridge that reduces setup time from hours to minutes. The SeqStudio instrument is a low-throughput, easy-to-use, and convenient benchtop system that delivers gold-standard Sanger sequencing and fragment analysis with just a simple click. It is easily used across a broad range of applications as well as by multiple users.



- Convenient and easy-to-use cartridge system includes the capillaries, polymer, and buffers required for each run
- Combine Sanger sequencing and fragment analysis reactions in the same instrument run
- Fast turnaround with a run time of as little as 30 minutes
- Set up and monitor your run; view, manage, and share your data from anywhere, using your phone, tablet, or computer (Apple™ or Windows™ PC)*
- Secondary analysis software package included with system purchase, including Applied Biosystems™ GeneMapper™ 5.0, Minor Variant Finder, and others
- One-year instrument warranty included
- Get up and running quickly with SmartStart™ orientation—in-lab, one-day training for basic setup, cloud enablement, reagent review, and instrument operation and maintenance

* Internet connection required.

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

SNaPshot Multiplex System

SNPs play a critical role in understanding differences in genetic traits, susceptibility to disease, and response to drug therapies. To analyze SNP variants, the Applied Biosystems™ SNaPshot™ Multiplex System was developed to work seamlessly with the SeqStudio Genetic Analyzer with built-in reporting of fragment analysis results of size and peak area. The ability to mix fragment analysis and sequencing reactions on one plate enables investigators to perform SNP profiling and Sanger sequencing on a single run.

Human cell line and sample authentication

The study and development of human diseases relies heavily on the analysis of dissociated human cell lines grown in culture. However, an increasingly acknowledged problem is that cells grown *in vitro* can be misidentified or contaminated with other unrelated cell lines. The identity of cell lines can be verified by the analysis of a highly specific genetic "fingerprint" of highly variable short tandem repeats (STRs). The SeqStudio Genetic Analyzer integrates well with Applied Biosystems™ Identifiler™ Plus, Identifiler™ Direct, and GlobalFiler™ kits.

Nucleic acid preparation

DNA extraction kits

Performance in downstream applications is often influenced by the quality of the starting nucleic acid being analyzed. We offer a broad range of kits for purifying high-quality genomic DNA from a variety of sample types.

Features of recommended Applied Biosystems™ and Invitrogen™ nucleic acid preparation kits.

Product	DNAzol™ Reagent	DNAzol™ BD Reagent	PureLink™ Genomic DNA Mini Kit	PureLink™ *Pro* 96 Genomic DNA Purification Kit	MagMAX™ FFPE DNA/RNA Ultra Kit	MagMAX™ DNA Multi-Sample Ultra Kit 2.0
Sample type	Tissue, cells	Blood	Tissue, blood, cells	Tissue, blood, cells	FFPE & fixed samples	Blood
High-throughput compatible	No	No	No	Yes	Yes	Yes
Isolation method	Organic extraction	Organic extraction	Silica membrane	Filter plate	Magnetic beads	Magnetic beads

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

Reduce hands-on time and user variation by combining Thermo Scientific™ KingFisher™ instruments with Applied Biosystems™ MagMAX™ kits for a complete automated solution.

Instrument	KingFisher Duo Prime	KingFisher Flex	KingFisher Presto
			
Size	Compact benchtop	Benchtop	Integrates with liquid handler
Throughput	Low-medium throughput 6/12/24 samples per run	Medium-high throughput 24–96 samples per run	High-throughput 96 samples per run

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

Service and support

Comprehensive instrument warranty

Our factory-trained and certified field service engineers (FSEs) deliver the highest quality of service and technical expertise. Your warranty covers all 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, more predictable operating costs, enabling more running time on reliable instruments.

- Flexible and configurable support solutions
- Prioritized response based on your business demands
- Optimum reliability via scheduled preventative system maintenance
- Optimum workstation performance and latest software updates
- Discounted optional services and support products (varies by region)
- Predictable operating cost

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**

Find out more about our genotyping solutions at **thermofisher.com/genotyping**

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