

Predictive genomics

## **Total solution**

# Your partner from sample to insights





# The total solution for predictive genomics

Predictive genomics is changing the way healthcare will be delivered. Population genomics, carrier screening, and preemptive pharmacogenomics research are already beginning to reveal meaningful data that can help predict disease risk and potential drug response. Discoveries are being enabled by sample handling, sample preparation, and analysis technologies that are more powerful, easier to use, and more cost-efficient than ever before.

Advancements in microarray technologies offer opportunities to gain new insights into genetic diseases, traits, and biological functions quickly and cost-effectively for predictive genomics studies of any length or size. The Applied Biosystems<sup>™</sup> genotyping microarray portfolio encompasses a wide range of arrays and variant panels that can be tailored for you to interrogate hundreds-of-thousands of genetic markers that are most relevant for your unique needs in disease risk and pharmacogenomics research. Genotyping data analysis advancements enable you to investigate only the most relevant variants in your population on a single array, without distraction and lost time due to irrelevant data or complex bioinformatics.

Propel your predictive genomics research initiative forward using genotyping microarrays that can increase your confidence in the insights you uncover.

## Contents

Total solution for predictive

Sample collection

Sample storage

Sample preparation

Microarray analysis

Target verification

Laboratory Information Ma

e genomics research	4
	6
	7
	8
	12
	18
nagement System (LIMS)	19







## Total solution for predictive genomics research

Shown here is a sampling of our extensive portfolio of products to support your predictive genomics research. (Not all products may be required for your workflow.)

## Advancing your research goals with laboratory solutions for every step

Thermo Fisher Scientific is unique in the breadth and depth of our portfolio of essential supplies, everyday laboratory equipment, advanced genetics technologies, and expert support to help enable the success of your study objectives, experimental and analysis workflows, and desired experimental outcomes. Accelerate your predictive genomics initiatives with a single expert source to help you select an ideal complement of laboratory products and technologies for each step in your studies.



## Laboratory Information Management System (LIMS)

## Sample collection Maximize sample yield from collection to destination

Uphold your confidence in the integrity, homogeneity, and purity of your samples from the moment they are collected through transport to their final destination.



Easy to use saliva collection kits

Simplify saliva sample collection with the Thermo Scientific<sup>™</sup> SpeciMAX<sup>™</sup> Stabilized Saliva Collection Kit, which is pre-filled with stabilization agent for virus inactivation.

## Secure cryogenic shippers

Help ensure optimal safety and security of your genotyping samples with Thermo Scientific<sup>™</sup> Arctic Express<sup>™</sup> Transport Systems lightweight cryogenic shippers. Integrated liquid nitrogen absorbent prevents spillage.





## Sample storage

## Help ensure sample integrity for longitudinal studies

Safeguard your stored samples for months or even years for continuity in longitudinal and large-scale population genomics studies with dependable, ergonomic, and time-saving cryogenic products.



environmental sustainability.

## Convenient remote monitoring systems

Simplify monitoring of legacy and new instruments, multiple brands, or cold-chain systems from remote locations with a single easy-to-use application. Thermo Scientific™ Smart-Vue™ Pro system communicates via a cloud server for full system access anytime from anywhere.



## High-efficiency cryopreservation systems

#### High-speed sample barcode readers

Accelerate sample management with instant tracking and identification in sample storage The Thermo Scientific<sup>™</sup> VisionMate<sup>™</sup> High Speed Barcode Reader rapidly decodes 2D barcodes on sample tubes and racks simultaneously without user programming.



Versatile and ergonomic tube and vial decappers and cappers Speed sample handling and experimental protocols with automated decapping and capping. The Thermo Scientific<sup>™</sup> Decapper 500 enables hands-free decapping of ANSI-racked Thermo Scientific<sup>™</sup> Nunc<sup>™</sup> and Thermo Scientific<sup>™</sup> Matrix<sup>™</sup> screw-top tubes with Quick Switch Technology for quick switching between cap and rack styles.

## Dependable ultra-low-temperature freezers

Rely on uncompromising performance with Thermo Scientific<sup>™</sup> TSX Series ultra-low temperature (ULT) freezers. Our freezers incorporate advancements in responsive temperature control, ergonomics, and ENERGY STAR<sup>™</sup>-certified



Enable efficiency and accessibility in your cryopreservation systems. Thermo Scientific™ CryoExtra<sup>™</sup> Series High-Efficiency Storage Systems are highly versatile with multiple performance options and enhanced ergonomics features. Microprocessor-based liquid nitrogen (LN2) level control facilitates outstanding temperature uniformity.



## Sample preparation Maximize productivity in preparation for genetic analyses

Trust excellence in the everyday instruments and consumables that are the engines behind your predictive genomics research. The performance, reliability, and reproducibility of our sample preparation products can make the difference between excellent and adequate results.





## High-throughput magnetic bead-based extraction systems

Combine yield, quality, and convenience for DNA, RNA, plasmid, and protein extraction and isolation in a single magnetic bead-based system. Thermo Scientific™ Kingfisher<sup>™</sup> Apex instruments provide versatile and user-friendly benchtop operation. Thermo Scientific<sup>™</sup> KingFisher<sup>™</sup> Presto high-throughput magnetic particle processors incorporate automated liquid handling for plate filling in high-throughput laboratories. Thermo Scientific<sup>™</sup> KingFisher<sup>™</sup> Flex Purification Systems offer consistent automated processing of DNA/RNA, protein or cell purification from virtually any source.

## High efficiency magnetic bead-based DNA isolation kits

Simplify genomic DNA isolation from a variety of sample types without compromising yield or purity. The Applied Biosystems<sup>™</sup> MagMAX<sup>™</sup> DNA Multi-Sample Ultra 2.0 Kit uses automatable magnetic bead-based sample preparation technology that combines multiple sample processing steps together into a single step. Applied Biosystems<sup>™</sup> PrepSeq<sup>™</sup> nucleic acid extraction kits yield high-quality DNA from pathogens or residual host cells.





#### High yield nucleic acid extraction and purification spin column kits

Help maximize yield and purity of high-quality nucleic acids extracted from research samples spin-column and automatable magnetic bead-based technologies. Invitrogen™ PureLink™ Total RNA Blood Kit yields total RNA free from non-nucleated erythrocytes in less than one hour. Invitrogen<sup>™</sup> PureLink<sup>™</sup> Genomic DNA Kits simplify extraction of genomic DNA using a single purification kits are tailored for specific nucleic acid types, sources, throughput, and applications.



## Automated Axiom target preparation

the GeneTitan MC Instrument.

## Efficient Axiom sample preparation kit

Simplify Axiom array sample preparation. The Axiom 2.0 Assay prepares your genomic DNA for microarray research analysis. A single kit provides reagents for all steps including DNA amplification, fragmentation, purification, resuspension, hybridization preparation, and stain in preparation for the GeneTitan MC Instrument.



# Highly accurate sample quantification

## Fast RNA isolation blood tubes

Streamline sample collection with immediate stabilization and isolation of RNA and genomic DNA from whole blood drawn into Applied Biosystems<sup>™</sup> Tempus<sup>™</sup> Blood RNA Tubes.



Help maximize performance in your Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> assays with automated sample preparation to help minimize run-to-run variability and hands-on time. The Nimbus Target Preparation Instrument automates the Applied Biosystems™ Axiom<sup>™</sup> 2.0 Assay from start-to-finish in preparation for microarray processing on



Support efficiency with highly accurate quantification of DNA, RNA, and protein. Thermo Scientific<sup>™</sup> NanoDrop<sup>™</sup> One Microvolume UV-Vis Spectrophotometers deliver full-spectral data in seconds to help you optimize use of samples in downstream applications.



## Sample preparation



## Single-source molecular biology kits, enzymes, and reagents

Build efficiency into your predictive genomics studies with a single source of kits and reagents for all your genotyping research workflows including sample extraction and isolation kits; expression, cloning, and gene editing vectors; enzymes; assays; labeling dyes; and general high-grade molecular biology reagents.

#### High-performance microplate readers

Empower genotyping experiment success with accurate quantification of input DNA or RNA. Thermo Scientific<sup>™</sup> Multiskan<sup>™</sup> SkyHigh Microplate Spectrophotometers and Thermo Scientific<sup>™</sup> Varioskan<sup>™</sup> LUX Multimode Readers guantitate up to 16 microvolume samples as well as 96- and 384-well plates with high sensitivity, selectivity, and dynamic range.





#### Precise and fast reagent dispensers

Accelerate experimental protocols with precise and fast high-throughput preparation of microplates for PCR, sequencing, assays, and storage. Thermo Scientific™ Multidrop<sup>™</sup> Combi Reagent Dispensers are easy to use and flexible for a wide range of genomics research applications.

#### Robust and versatile benchtop centrifuges

Rely on consistently high performance run-after-run with our Thermo Scientific™ General Purpose Pro Centrifuges for sample prep and isolation. These centrifuges live up to their name as our most versatile benchtop centrifuges for all stages of predictive genomics research.





## Secure plate sealers thermoplastic elastomer (TPE) cap seals.

# installation and programming.

## High-efficiency biological safety cabinets

Uphold the safety of your laboratory staff and your experiments with a highperformance biological safety cabinet for your applications. Thermo Scientific™ Herasafe<sup>™</sup> 2030i Biological Safety Cabinets with built-in Wi-Fi capability facilitate product, personnel, and environmental safety for sensitive work.



#### High-efficiency vacuum concentrators

Quickly, efficiently, and safely concentrate nucleic acid samples, PCR preparations, and synthetic oligonucleotides. Thermo Scientific<sup>™</sup> Savant SPD1030 and SPD2030 Integrated SpeedVac<sup>™</sup> Systems are fully integrated vacuum concentration systems that combine all necessary components into a single, compact unit designed for fast



Support the purity of samples and reactions for PCR, storage, and other applications. Thermo Scientific plate sealers enable reliable sealing for adhesive, heat, or

## Microarray analysis

Power your predictive genomics research with whole-genome microarray analysis

## Highly efficient genotyping for very large population cohorts

There is a reason many of the world's largest population genotyping, carrier screening, and preemptive pharmacogenomics research initiatives have been established on Axiom microarray platforms. Axiom microarrays are ideal for genotyping of very large populations because they are scalable for any size study. Hundreds-of-thousands of variants can be genotyped for each sample. Imputation from existing genomewide array data can provide millions of additional genotypes. Genotyping using microarrays is highly accurate, even for calling low-freqency or rare variants. Data storage and computing needs are low and data analysis is more straightforward. The result of these benefits is a fast, scalable, customizable, and cost-effective solution to building insights into predictive genomics from research using very large population cohorts.

## Expertise and support when you need it, where you need it Our commitment to your success in predictive genomics research goes beyond

devices and technologies.

## Collaborative genotyping array design and analysis

We collaborate with you from array design through ongoing data analysis so you can be more confident in the insights you gain throughout your studies.

## Expert microarray research services

Help maximize the productivity of your laboratory by strategically outsourcing components of your predictive genomics studies. The Applied Biosystems Microarray Research Services Laboratory and Axiom Genotyping Services offer genotyping and expression services for large-scale, microarray-based studies.

## Comprehensive genotyping knowledge center

Find answers to your questions about genotyping solutions in our Genotyping Support center. Learn how to get started with your population genomics research, read more about genotyping research applications, explore comparison and conversion calculators, and find answers to troubleshooting questions.

## Extensive training portfolio

Keep learning. Gain new population genomics expertise through online, on-site, or training center courses. Our highly skilled research application scientists lead sessions on microarrays, sequencing, and bioinformatics as well as a wide range of other topics.

## Ecosystem of partnerships

Execute your predictive genomics research with the confidence of shared expertise. We continually expand and enrich our relationships with predictive genomics leaders in industry and academia. Leveraging our ecosystem of partnerships from study strategy through therapeutic treatment research is an efficient way to make population genomics predictive.



## Custom and catalog genotyping microarrays

Axiom arrays offer the best of convenience and flexibility

Streamline your genotyping studies with microarrays that are tailored for your specific needs in population genomics, carrier screening, and preemptive pharmacogenomics research.



#### **Biobanking arrays**

Support productivity in investigation of the genetics of complex diseases with high-throughput genotyping of large sample cohorts. The unique modular content strategy of Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> Biobank Plus Genotyping Arrays enables you to quickly and easily tailor your content for the distribution of variants that interest you. Axiom Biobank arrays are ideal for high-throughput, high-value genotyping of large sample cohorts for studies of genome-wide associations, exome variants, inflammation and HLA, pharmacogenomics, SARS-Cov2, and complex diseases.

#### **Population-optimized arrays**

Expand the breadth of your variant genotyping with genome-wide disease association studies of populations around the world. Axiom population-focused arrays have been curated to support extensive coverage of rare and common variants for a broad range of global ethnic populations. Applied Biosystems<sup>™</sup> Axiom Population-Optimized Arrays are designed for specific targeted populations and Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> World Arrays cover variants for multiple populations and subpopulations.

#### Precision medicine research arrays

Help maximize productivity with focused investigation of variants associated with complex disease susceptibility, preemptive pharmacogenomics, wellness, and lifestyle. The Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> 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<sup>™</sup> Axiom<sup>™</sup> Precision Medicine Research Array (PMRA) provides broad coverage across all major ancestral populations. The Axiom Asia PMRA is curated specifically for coverage of South and East Asia populations.

#### Carrier screening arrays

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

#### Preemptive pharmacogenomics arrays

Gain broader scientific insights into genetic variability associated with drug absorption, distribution, metabolism, and excretion (ADME). The Applied Biosystems<sup>™</sup> PharmacoScan<sup>™</sup> Solution is designed to investigate both low- and high-evidence markers to enable thorough genotyping for the research of potential drug response. The Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> PharmacoFocus<sup>™</sup> Solution enables targeted genotyping of high-evidence SNP, copy number variants (CNVs), and human leukocyte antigen (HLA) variants.



## Axiom arrays for comprehensive and cost-effective predictive genomics research

Axiom arrays are a genotyping solution that enables you to optimize your array designs easily and cost-effectively by choosing from 11 million wet-lab-verified, and fully annotated markers, in our Axiom Genomic Database, Add your own variants if needed. Our artificial-intelligencebased array design and imputation-aware single nucleotide polymorphism (SNP) selection algorithms deliver high accuracy and coverage. Content across the Axiom array portfolio covers an extensive range of rare and common variants for ethnic populations, maximizing the power of predictive genomics studies for diverse and admixed cohorts. Your array content can be modified at any time over the course of a longitudinal study, giving you flexibility to add, remove, or replace markers. Our unique microarray manufacturing technology ensures high SNP fidelity so even your custom variants will remain consistent for large cohort and longitudinal research studies.

#### Axiom custom arrays

Create your own genotyping array designs with Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> myDesign Genotyping Arrays. Take the guesswork out of custom genotyping array design. Whether you need whole-genome or targeted genotyping, Axiom myDesign Genotyping Arrays enable you to create your own array designs quickly, easily, and cost-effectively. The arrays are available in a range of marker coverage and plate formats.





## Microarray products, software and services

Maximize your productivity with exceptionally powerful and flexible microarray technologies

The complete Axiom solution incorporates fast, scalable, customizable, and cost-effective instrumentation, arrays, workflows, and software to streamline your predictive genomics research.





## GeneTitan Multi-Channel (MC) Instrument for hands-free automated array processing

Accelerate your microarray processing with seamless high-throughput automation for a broad range of genotyping research applications including genome-wide association studies (GWAS), replication studies, candidate-gene association, and targeted genotyping. The GeneTitan Multi-Channel (MC) Instrument integrates hybridization, washing, and imaging of Axiom genotyping arrays and any other Applied Biosystems array plates in a single instrument. Applied Biosystems<sup>™</sup> GeneChip<sup>™</sup> Command Console<sup>™</sup> Software provides intuitive instrument control, flexible workflows, and sample registration. Increase productivity with hands-free, unattended plate handling and automatic primary analysis of genotyping data quality.

## Streamlined Axiom array data visualization and analysis software

Discover, extract, analyze, and visualize genotyping information for simple and complex genomes in a single graphical interface. Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> Analysis Suite software integrates SNP genotyping, insertion and deletion (indel) detection, multi-allele analysis, and off-target variant analysis for genotyping research applications. Conduct sample and plate quality control, automated genotype calling, and SNP classification in a single step. Additional Axiom array analysis software tools are available for investigations in high-resolution HLA typing, transcriptome analysis, microbial detection, and drug metabolism pathways.

## Faster and cost-effective array processing with Axiom Propel workflow

Scale up your genotyping research with a fast workflow for start-to-finish high-throughput genotyping. The Applied Biosystems<sup>™</sup> Axiom<sup>™</sup> Propel workflow shortens the manual Axiom 2.0 assay to just 72 hours. The Propel workflow is a pre-optimized, modular, and scalable integration of common lab equipment such as shakers, centrifuges, and Multidrop Combi Reagent Dispensers into a microarray processing instrumentation workflow that can help reduce up-front capital investment in automated liquid handlers.





## Leverage our bioinformatics expertise to design your arrays

We provide the array design expertise, so you don't have to. Our microarray bioinformatics team will work with you to create custom array designs tailored to your population of interest. We help you select and prioritize your desired content and annotations. Your array is designed to maximize imputation and ensure your most important markers are covered by multiple probe sets and high redundancy.



Simplified preparation of hybridization-ready Axiom genotyping targets

Reduce processing complexity with Axiom 2.0 Reagent Kits. Produce high-quality, hybridization-ready targets with pre-optimized reagents and empirically verified variants in multiple plate formats. Help maximize productivity with automated Axiom sample preparation to help minimize run-to-run variability and hands-on time. The Nimbus Target Preparation Instrument automates the Axiom 2.0 Assay from start-tofinish in preparation for microarray processing on the GeneTitan MC Instrument.



## **Target verification**

Gain confidence in experimental outcomes with validated variants

There is a Thermo Fisher Scientific solution to verify SNPs, indels, and CNVs for all your array-based genotyping research needs.



Turnkey automation of NGS workflows with the Genexus System Simplify verification of genotyping variants with high-resolution, whole-gene NGS on the Ion Torrent<sup>™</sup> Genexus <sup>™</sup> System. The Genexus System automates nucleic acid extraction and quantification together with library preparation, templating, sequencing, and analysis.

## Precise gene expression analysis with real-time quantitative PCR (RT-qPCR) and digital PCR

Quantify even low-level expression changes of genotyping targets with excellent discrimination. Applied Biosystems<sup>™</sup> QuantStudio 7 Pro Real-Time PCR System enables you to analyze variant expression data from multiple types of RT-gPCR experiments as a single project within minutes. Applied Biosystems<sup>™</sup> QuantStudio<sup>™</sup> Absolute Q<sup>™</sup> Digital PCR System enables exceptionally sensitive quantitation for low-level and rare targets with complete sample preparation on a single plate.





## Fast and flexible fragment analysis and sequence verification with capillary electrophoresis

same plate at the same time. The Applied Biosystems<sup>™</sup> SeqStudio<sup>™</sup> 24 Flex Genetic designed for ease-of-use and flexibility.



## Help maximize research traceability with configurable LIMS data management

Enable safe and secure capture, tracking, documentation, and maintenance of your valuable research data and intellectual property.

## Configurable LIMS helps maximize workflow efficiency

Streamline your predictive genomics research initiatives by using a single Laboratory Information Management System (LIMS) platform to manage study protocols, laboratory processes, and analytical results. Thermo Scientific<sup>™</sup> Core LIMS software is easily configured for research laboratories to collect, share, analyze, and archive scientific data. Core LIMS software runs on Thermo Fisher<sup>™</sup> Platform for Science software to help automate your workflows, manage your samples and data, and integrate with instruments and software from your preferred vendors. Biospecimen registration, secure data storage, and metadata tracking create a complete biospecimen lifecycle and chain of custody record.





Learn more at thermofisher.com/predictive-genomics

## applied biosystems

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