Sequencing

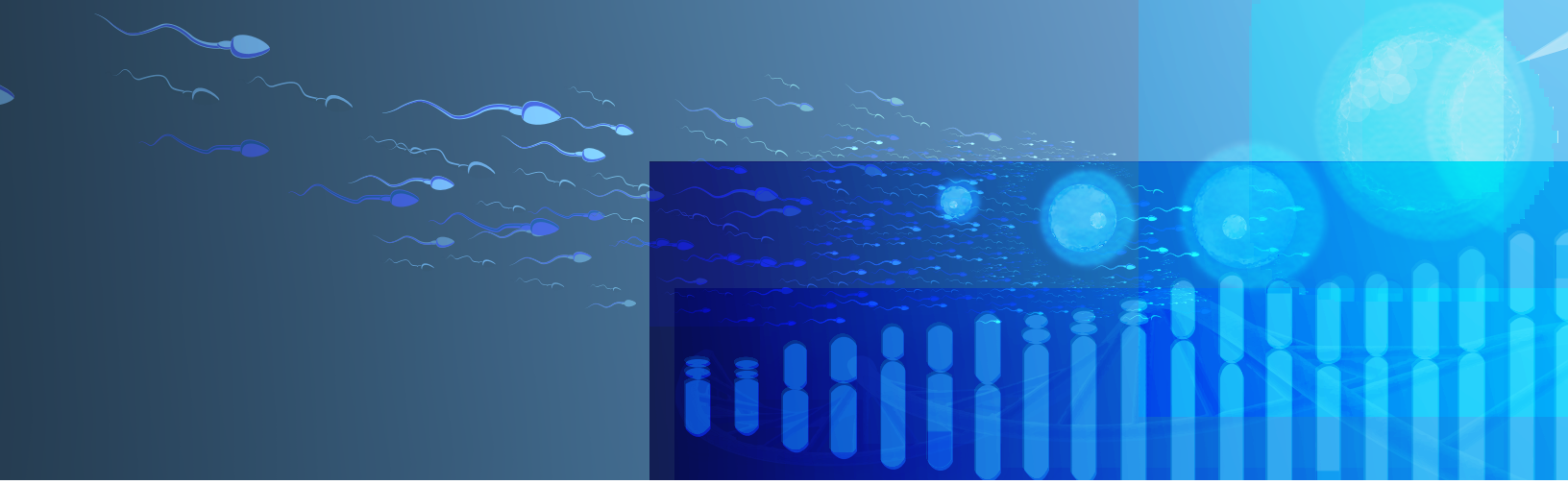
Next-generation decisions empowered by next-generation technologies

Reproductive genetic testing with Ion Torrent NGS solutions

Contents

“We have been using Ion Torrent NGS now for about 3 years. The results are clear. We don’t have many ambiguous or questionable results. The scalability of the system is good, and as our experience with the system increases, so does our confidence in the results we obtain. I’m sure that confidence is evident when we discuss results.”

—Dr. Colin Lee Soon Soo
Alpha Fertility Centre



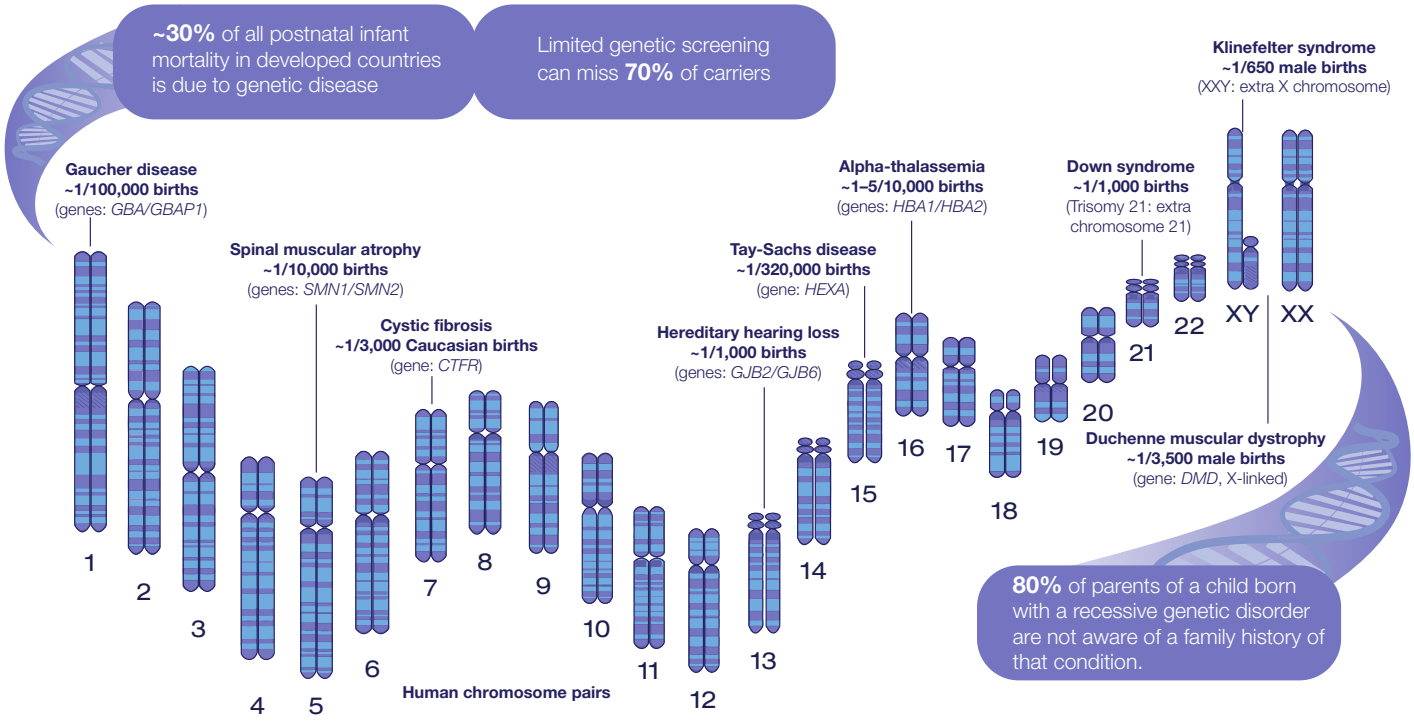
Next-generation sequencing advances reproductive health research

Reproductive genetic research is fast becoming a desired source of education for family planning, regardless of maternal age or family ancestry. 80% of parents with a child born with a recessive disorder are not aware of a family history of that condition,[1] but labs implementing next-generation sequencing (NGS) for inherited disease research are expanding our ability to identify and understand life-impacting genetic variants.

NGS technology is revolutionizing the field of reproductive health with fast, accurate, and comprehensive detection of a broad spectrum of genetic variants known to cause inherited disorders. NGS interrogates hundreds to thousands more genes than preceding technologies. It enables research and analysis of various genomic features from a single sequencing run, including single-nucleotide variants (SNVs), insertion–deletion variants (indels), copy number variations (CNVs), and structural variants. **Genetic findings for reproductive purposes guide decisions toward faster time to pregnancy and healthier future offspring.**

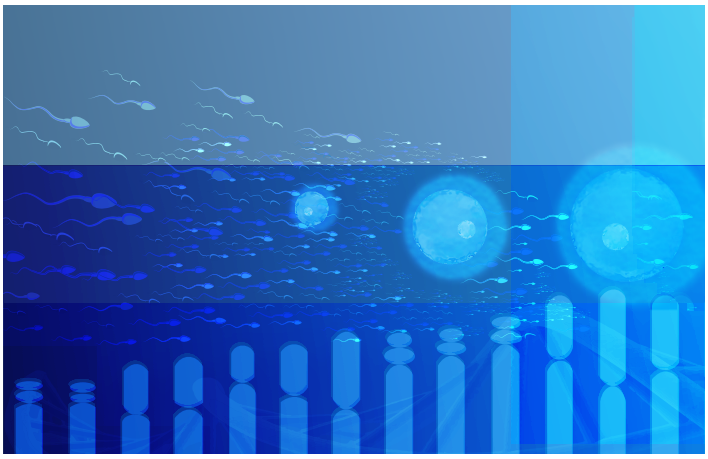
Ion GeneStudio™ S5 systems and Ion Torrent™ NGS technology can help **expand your menu of genetic testing services**. Sample-to-result workflows for a broad offering of genetic tests and seamless scaling to throughput changes enable sample throughput flexibility and the expansion of service offerings, with a single investment. Add intuitive Ion Torrent™ software, for simple analysis and reporting, to make **NGS accessible to reproductive genetics researchers and specialists, regardless of their level of bioinformatics expertise**.

Prevalence of inherited genetic disorders fuels the need for reproductive genetics research



1. Gao Z et al. (2015) An estimate of the average number of recessive lethal mutations carried by humans. *Genetics* 199:1243–1254.

Comprehensive pan-ethnic carrier screening research



Traditional carrier screening research relies on ethnicity or known family history to guide the disease areas of focus. However, as pan-ethnic and multicultural societies become more prevalent and gene variants become more widespread and less specific, standard predictors of disease risk become less reliable as research tools.

NGS boosts carrier screening research by targeting an expansive panel of known variants, to identify and determine carrier status related to a broad range of disease conditions. **Findings from expanded carrier screening research provide scientific insights to help families reduce risk for future pregnancies and increase probability for healthy offspring**.

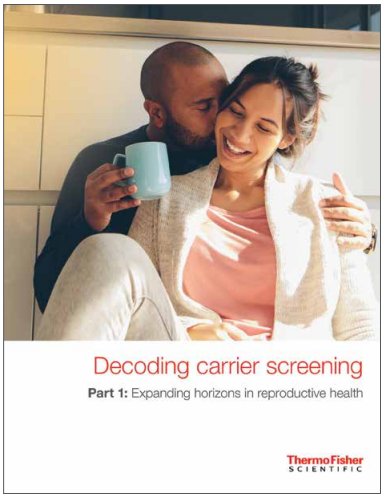
Ion Torrent™ CarrierSeq™ expanded carrier screening (ECS) kits

This single, consolidated NGS assay targets difficult-to-detect variants, due to pseudogenes (*GBA* and *GBAP1* for Gaucher disease), paralogues (*SMN1* and *SMN2* for spinal muscular atrophy), or targets within a locus (*HBA1* and *HBA2* for alpha-thalassemia).

As an end-to-end solution for simple adoption and implementation in your lab, CarrierSeq ECS kits include reagents for library preparation, template preparation, and sequencing, plus software for data analysis and reporting. Leveraging Ion AmpliSeq™ technology, these optimized assay kits streamline implementation and help ensure reliable and consistent results.

A 420-gene panel targeting the full coding region of all genes enables the analysis of >36,000 nonbenign ClinVar variants for SNVs, indels, and CNVs by NGS.

~14,000 amplicons	**418** inherited disorders
420 genes	**33** genes with single exon–level resolution
	>36,000 SNVs, indels, and CNVs



Download ebook

CarrierSeq ECS Complete Gene Panel

Analytic verification of a CarrierSeq ECS kit for the Ion GeneStudio S5 System

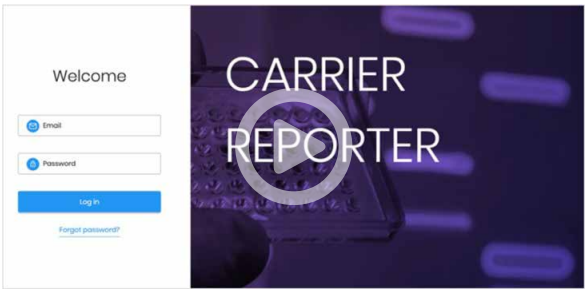
Carrier Reporter Software

Carrier Reporter Software streamlines sample management, variant classification, and case review without the need for bioinformatics assistance. Variant interpretation and classification are quickly accomplished using multiple sources including ClinVar, evidence-based American College of Medical Genetics and Genomics (ACMG) rules, and proprietary databases.

Classification of variants can be configured based on user-defined criteria and variant review. Variant review in Carrier Reporter Software includes links to major variant interpretation sources, allowing further investigation of variant significance. All classification changes are documented and saved by the software.

Analysis can be customized to include or remove genes, variants, and the reporting rules for the different variant classes. Further, for a single sample, Carrier Reporter Software accepts variant-called data from multiple sources to produce a customized, comprehensive final report. Publish reports for individuals or paired results with the option to include reproductive risk calculations.

Additionally, Carrier Reporter Software offers the option to export genetic findings, genomic coordinates, prediction evidence, and many other sample-specific data points for integration into a local laboratory information management system (LIMS).



Watch demo

Contact a reproductive health specialist

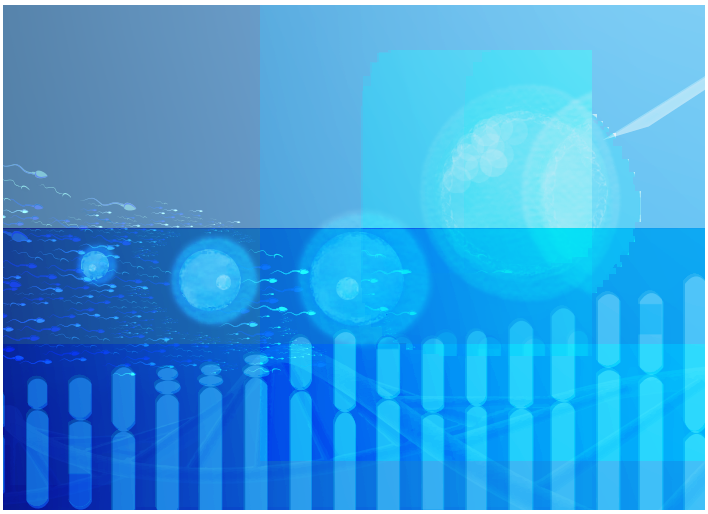
CarrierMax FMR1 Reagent Kit

Fragile X syndrome arises from silencing or genetic disruption of the Fragile X mental retardation gene (*FMR1*). Fragile X is a leading cause of developmental delay, making it a key target for carrier screening research.

The **Applied Biosystems™ CarrierMax™ FMR1 Reagent Kit** reliably analyzes samples to determine the *FMR1* CGG repeat status of both *FMR1* copies for females and the one *FMR1* gene for males.

Combine with CarrierSeq ECS kits for comprehensive pan-ethnic carrier screening research.

Simultaneous PGT-A and PGT-M on a single biopsy



Families may determine that *in vitro* fertilization (IVF) offers the greatest probability for healthy offspring, due to expanded carrier screening findings or other personal or medical knowledge.

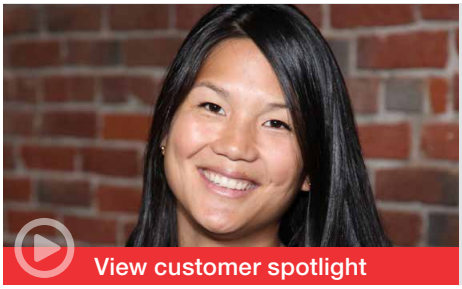
Preimplantation genetic testing (PGT) by NGS delivers genetic insights used for research of embryo prioritization. PGT-A offers swift, accurate identification of aneuploidy (abnormal chromosome number), while PGT-M uncovers presence of monogenic disorders. These genetic findings provide scientific insight that help reduce the time to pregnancy and decrease miscarriage rates.

Thanks to the power of Ion Torrent NGS technology, a single embryo biopsy can be used for both PGT-A and PGT-M investigations within a simple, integrated workflow.

Hear from your peers

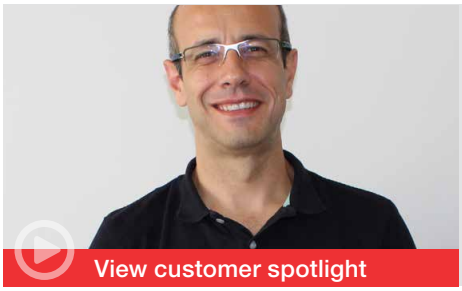
PGT-A simplified—Comparison of leading NGS solutions: Ion ReproSeq™ PGS and VeriSeq™ PGS kits

Adam Goodman
Director of Preimplantation Genetics, NxGen MDx



IVF and genomics: Positioning for the future with Ion ReproSeq PGS kits

Catherine Welch
Founder and Managing Partner, Sequence46



Simplified preimplantation genetic testing workflow on a single embryo biopsy

Luis Alcaraz
Co-founder, Scientific and Laboratory Director, Bioarray

Ion ReproSeq PGS kits

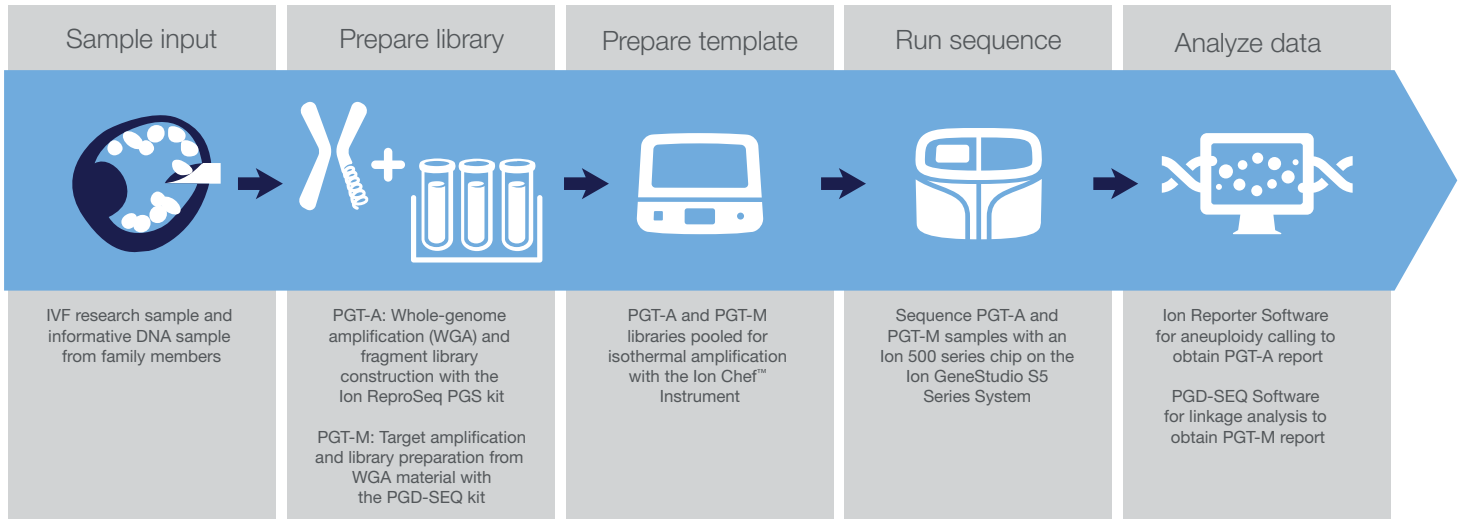
- **Versatile and comprehensive**—detect whole-chromosome, mosaic, and small CNVs, from just 6 pg of DNA from a single cell or multiple cells
- **Scalable throughput**—three kit configurations enable analysis of up to 16, 24, or 96 samples in a single run on a single platform
- **Rapid, end-to-end NGS workflow**—cells to results in less than 10 hours* and minimized user error, with <2 hours* hands-on time
- **Enhanced interpretation of results with Ion Reporter™ Software**—mosaicism detection, gender masking, and easy-to-interpret data plots

PGD-SEQ kits

PGD-SEQ™ kits offer **over 200 gene panels to research embryo status of common and rare monogenic/single-gene disorders (PGT-M) for IVF**. Targets of interest are identified through carrier screening research and/or known family history.

- Complete kit reagents for PGT-M library preparation to analyze up to 15 samples
- Comprehensive familial carrier status using proprietary linkage analysis software
- Compatible with the Ion ReproSeq PGS Kit for combined PGT-M and PGT-A analysis from a single research sample

View our growing list of over 200 PGD-SEQ panels.
Custom panels available upon request.



* Turnaround time for the Ion ReproSeq PGS Kit with Ion 510™ Chips (16 samples/run) is less than 10 hours with less than 2 hours of hands-on time.

Ion Reporter Software

The aneuploidy analysis workflow in Ion Reporter Software makes analyzing and reporting aneuploidy results simple and fast. This low-pass coverage analysis workflow processes samples by comparing them to a built-in bioinformatics control baseline to deliver the ploidy status of each chromosome in a sample. Users can visualize aneuploidy profiles using the customized Integrative Genomics Viewer (IGV) karyotype view and create their own interpretive report.

Contact a reproductive health specialist

Detection of aneuploidies, segmental aneuploidies, and mosaicism using Ion ReproSeq kits

NGS solutions that evolve with your business

Ion GeneStudio S5 systems

The Ion GeneStudio™ S5, S5 Plus, and S5 Prime systems are benchtop sequencers that enable sequencing of panels, small genomes, exomes, and transcriptomes on a single platform. Interchangeable sequencing chips are available for reads ranging 2M to 130M, supporting a growing lab demand.

Built on Ion Torrent semiconductor sequencing technology, the **Ion GeneStudio S5 systems** deliver simple, accessible sample-to-solution workflows with speed, accuracy, and a broad range of gene recognition and scalability for all throughputs.

Learn how Ion Torrent semiconductor sequencing works

Three simple steps get you quickly from sample to data analysis

Library and template prep

Library prep, template prep, and chip loading with 30 minutes of hands-on time using the Ion Chef system

Sequencing

Complete sequencing runs in as little as 2.5 hours with less than 15 minutes of setup time with Ion GeneStudio S5 instruments

Data analysis

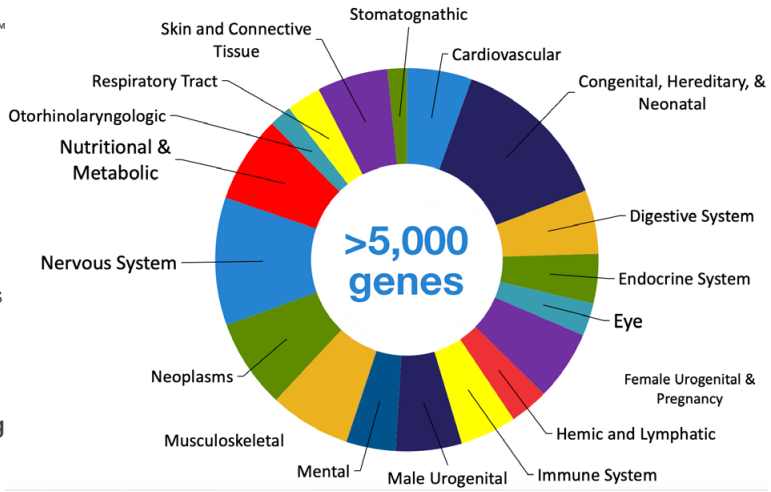
Transform data into insights with in-depth analysis, QC reporting, and variant calling using the Ion Reporter Server

Spend less time thinking about sequencing and more time answering critical reproductive genetics research questions.

Ion AmpliSeq on-demand panels

Newborn genetic screening research analyzes DNA samples against a list of genetic and metabolic disorders. **Ion AmpliSeq™ on-demand panels allow creation of configurable panels from a large collection of pretested genes covering a variety of inherited disease areas such as newborn screening research. Customize Ion AmpliSeq on-demand DNA panels from a catalog of >5,000 lab-tested genes of known variants**. Quickly filter by disease research areas and subcategories of interest to populate genes. Practical pack sizes and customer configurations offer flexibility to scale to your project needs.

Other investigational areas of reproductive health, including preeclampsia or endometrial receptivity, can be explored using Ion AmpliSeq made-to-order panels. Ion AmpliSeq targeted resequencing solutions run on Ion GeneStudio S5 systems create a simple, low-cost path for expanding your reproductive health lab services.



Analytical Validation Consulting Services

Thermo Fisher Scientific Analytical Validation Consulting Services offer fast and cost-effective analytical validation (AV) of NGS-based assays and panels to testing labs that follow industry quality standards and regulatory guidelines. AV packages include both wet-lab and bioinformatics validations.

- Designed to support AV requirements for testing
- Step-by-step support throughout the AV process
- Project management by an AV specialist
- Workflow training and optimization
- Template documentation
- Controls
- Data analysis
- Analytical validation summary template
- Optional: confirmatory orthogonal testing services

We have completed more than 350 successful AV consulting service engagements—learn more at **thermofisher.com/av**

Benefits of AV support:

- Accelerate launch time
- Reduce cost and add transparency for your end-to-end investment
- Achieve lab compliance with templates for documentation

Priority Technical Support Plan

The Priority Technical Support Plan provides your lab with exclusive, priority access to our extensive global service and technical support team, with same-day response from a reproductive health specialist. You can't afford unanticipated downtime. We're committed to keeping your laboratory up and running.

Workflows covered:

- Ion ReproSeq PGS Kit for the Ion GeneStudio S5 System
- CarrierSeq ECS Kit for the Ion GeneStudio S5 System

Learn more at **thermofisher.com/instrumentservices**

Available in North America and EMEA 24 hours, Monday–Friday (excluding holidays).

Product training

Further your knowledge of preimplantation genetic testing, carrier screening, and instrument operation with hands-on workflow training courses. Choose from virtual and in-person classroom instruction, or hands-on learning in your lab, aligned to your schedule, budget, and learning preferences. Training can be used preinstallation, to further or refresh knowledge, or as a post-install follow-up with an application specialist.

Learn more at **thermofisher.com/educationservices**

Ordering information

Description	Quantity	Cat. No.
Carrier screening		
Ion AmpliSeq CarrierSeq ECS Panel	24 rxn	A48036
	96 rxn	A43471
CarrierSeq ECS Kit with Ion 530 Chips (3 samples/chip)	24 rxn	A48022
CarrierSeq ECS Kit with Ion 530 Chips (4 samples/chip)	96 rxn	A43585
CarrierSeq ECS Kit with Ion 540 Chips (15 samples/chip)	120 rxn	A48023
CarrierSeq ECS Kit with Ion 540 Chips (16 samples/chip)	384 rxn	A43586
CarrierMax FMR1 Reagent Kit	48 samples	952362
CarrierMax SMN1/SMN2 Reagent Kit	50 samples	952363
Preimplantation genetic testing		
Ion ReproSeq PGS Kit with Ion 510 Chips (16 samples/run)	64 samples	A34899
Ion ReproSeq PGS Kit with Ion 520 Chips (24 samples/run)	96 samples	A34900
Ion ReproSeq PGS Kit with Ion 530 Chips (96 samples/run)	384 samples	A34901
PGD-SEQ SMN1 Panel and Reagent Kit (spinal muscular atrophy)	Up to 15 samples	BA03784
PGD-SEQ CFTR Panel and Reagent Kit (cystic fibrosis)	Up to 15 samples	BA03800
PGD-SEQ COL1A1 Panel and Reagent Kit (osteogenesis imperfecta)	Up to 15 samples	BA03803
PGD-SEQ FMR1 Panel and Reagent Kit (Fragile X syndrome)	Up to 15 samples	BA03816
PGD-SEQ HBB Panel and Reagent Kit (beta-thalassemia)	Up to 15 samples	BA03820
PGD-SEQ HBA1/HBA2 Panel and Reagent Kit (alpha-thalassemia)	Up to 15 samples	BA03864
PGD-SEQ kit panel menu: growing list of >200 panels for research of common and rare monogenic (single-gene) diseases		Visit **thermofisher.com/pgdseq**
Ion AmpliSeq on-demand panels		
Ion AmpliSeq Ready-to-Use Panels		Visit **thermofisher.com**
Ion AmpliSeq Ready-to-Use Panels and Community Panels		Visit **ampliseq.com**
Ion AmpliSeq Made-to-Order Panels and On-Demand Panels		Visit AmpliSeq Designer at **ampliseq.com**
Ion AmpliSeq Library Kit Plus	24 rxns	4488990
	96 rxns	A35907
	384 rxns	A38875
IonCode Barcode Adapters 1–96 Kit	960 rxns	A29747
IonCode Barcode Adapters 1–384 Kit	3,840 rxns	A29751
Ion Xpress Barcode Adapters	1–96 complete set of adapters	4474517
Ion Library Equalizer Kit		4482298
Ion AmpliSeq Sample ID Panel		4479790
Sequencing systems		
Ion GeneStudio S5 System		A38194
Ion GeneStudio S5 Plus System		A38195
Ion GeneStudio S5 Prime System		A38196
Ion GeneStudio S5 System + SmartStart Orientation		A38405
Ion GeneStudio S5 Plus System + SmartStart Orientation		A38408
Ion GeneStudio S5 Prime System + SmartStart Orientation		A38411
Data analysis		
Ion Reporter Software Server System		4487118

Ordering information

Description	Quantity	Cat. No.
Analytical validation services		
ReproSeq Complete Validation Solution		A47476
CarrierSeq Complete Validation Solution		A47477
Priority Technical Support Plan for reproductive health applications		
Priority Technical Support Plan for the Ion GeneStudio S5 System	1 year	ZGLPSCIONS5
Priority Technical Support Plan for the Ion GeneStudio S5 Plus System	1 year	ZGLPSCIONS5PLUS
Priority Technical Support Plan for the Ion GeneStudio S5 Prime System	1 year	ZGLPSCIONS5PRIME
Training packages		
ReproSeq Training Package		TRN00362
CarrierSeq Training Package		TRN00365

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Isolate DNA with the most versatile sample preparation instruments on the market. Integrated software enables simple protocol customization to easily edit, modify, or create new protocols.

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Applied Biosystems™ MagMAX™ Saliva gDNA Isolation Kit

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