



# Obtain PGT-A and PGT-M results with a single NGS workflow

Comprehensive preimplantation genetic testing (PGT) research is now accessible to any lab, regardless of expertise in next-generation sequencing (NGS) or informatics. The Ion GeneStudio™ S5 System enables simultaneous research of chromosomal abnormalities and monogenic disorders with the **Ion ReproSeq™ kit for PGT-A and the PGD-SEQ™ kit for PGT-M**. These integrated sequencing solutions deliver a complete sample-to-results workflow, including simple analysis tools for aneuploidy detection and comprehensive familial carrier status.

Previously, PGT-M required expertise to design and verify informative genetic markers to interrogate each specific disorder. Now, the PGD-SEQ kit offers more than 200 Ion AmpliSeq™ panels targeting informative single-nucleotide variants (SNVs) for research of common and rare monogenic (single-gene) diseases, plus options for customization. Combined with the Ion ReproSeq PGS kits, labs can obtain both PGT-A and PGT-M results in the same run from a single sample—consolidating precious biopsy samples and reducing hands-on time and time-to-results.

## The Ion ReproSeq kit for preimplantation genetic testing-aneuploidy (PGT-A)

- Used for detection of aneuploidies, or chromosomal abnormalities, across all 24 chromosomes (22 autosomes and the X and Y chromosomes)
- Examples include trisomy 21 (Down syndrome) and monosomy X (Turner syndrome)

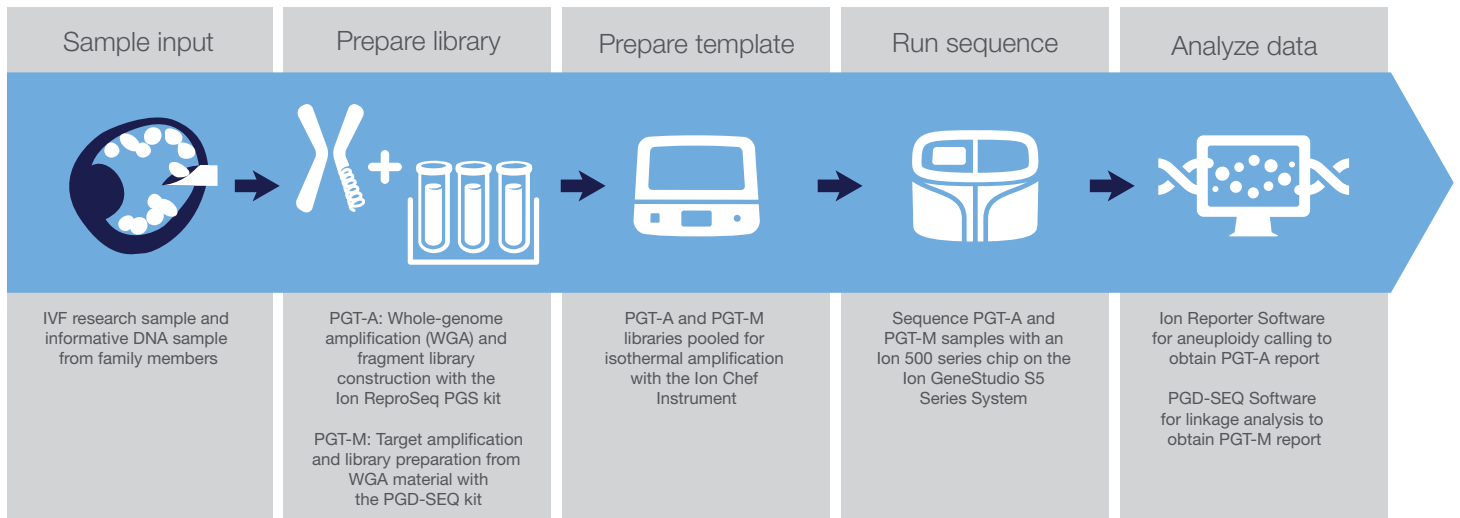
## The PGD-SEQ kit for preimplantation genetic testing-monogenic (PGT-M)

- Linkage analysis identification of disease alleles for single-gene disorders within the family
- Research examples include sickle cell anemia, cystic fibrosis, Huntington's disease, fragile X syndrome, and spinal muscular atrophy

Request a consultation with a reproductive health technical specialist at [thermofisher.com/pgt](https://thermofisher.com/pgt)

## Integrated NGS workflow for comprehensive PGT—sample to results in less than 12 hours\*

PGT-A and PGT-M libraries are prepared manually, then pooled libraries are placed in the Ion Chef™ Instrument for isothermal amplification, enrichment, and loading onto an Ion 500 series chip. Chips are then placed in the Ion GeneStudio S5 System for sequencing. Data analysis is performed by Ion Reporter™ Software for aneuploidy calling and PGD-SEQ™ Software for linkage analysis.



### Benefits of using Ion Torrent™ NGS systems for comprehensive PGT research:

- Obtain PGT-A and PGT-M results in the same NGS workflow from a single sample
- Set-up-and-go workflow allows new users to get up and running quickly with less training
- Utilize the simple, integrated analysis tools
- Get an economic in-house PGT research solution that grows with your business
- Take advantage of a rapid workflow for time-sensitive samples

\* Requires two operators for the library prep stage.

## Ordering information

Description	Unit size	Cat. No.
<b>DNA amplification to sequencing</b>		
Ion ReproSeq PGS Kit with Ion 510 Chips (16 samples per run)	64 samples	A34899
Ion ReproSeq PGS Kit with Ion 520 Chips (24 samples per run)	96 samples	A34900
Ion ReproSeq PGS Kit with Ion 530 Chips (96 samples per run)	384 samples	A34901
PGD-SEQ SMN1 Panel and Reagent Kit (spinal muscular atrophy)		BA03784
PGD-SEQ CFTR Panel and Reagent Kit (cystic fibrosis)		BA03800
PGD-SEQ COL1A1 Panel and Reagent Kit (osteogenesis imperfecta)		BA03803
PGD-SEQ FMR1 Panel and Reagent Kit (fragile X syndrome)		BA03816
PGD-SEQ HBB Panel and Reagent Kit (beta thalassemia)		BA03820
PGD-SEQ HBA1/HBA2 Panel and Reagent Kit (alpha thalassemia)		BA03864
Review the complete PGD-SEQ kit panel menu, a growing list of over 200 panels for research of common and rare monogenic (single-gene) diseases, at <a href="https://thermofisher.com/pgdseq">thermofisher.com/pgdseq</a>		
<b>Sequencing</b>		
Ion GeneStudio S5 System		A38194
Ion GeneStudio S5 Plus System		A38195
Ion GeneStudio S5 Prime System		A38196
Ion GeneStudio S5 System + SmartStart		A38405
Ion GeneStudio S5 Plus System + SmartStart		A38408
Ion GeneStudio Prime System + SmartStart		A38411
<b>Data analysis</b>		
Ion Reporter Software Server System		4487118

Get more information about PGT-A and PGT-M solutions or request a consultation with a technical specialist at [thermofisher.com/pgt](https://thermofisher.com/pgt)