
# Deliver premium PGT-A analysis

## Introducing SNP-based quality controls for preimplantation genetic testing for aneuploidy

Next-generation sequencing (NGS) offers exceptional resolution and throughput for preimplantation genetic testing for aneuploidy (PGT-A) and is highly effective at identifying whole-chromosome aneuploidies, segmental aneuploidies, and small structural rearrangements. Now, single-nucleotide polymorphism (SNP)–based quality control (QC) features are available for Ion ReproSeq™ PGS kits for premium PGT-A analysis.

### Prioritize embryo samples with SNP-based quality controls

SNPs are the nucleotide variations that make individuals unique; and while they vary from person to person, these polymorphisms are inherited and can be directly linked to parental DNA. SNPs can inform PGT-A analysis by offering additional insights on triploidy 69,XXX, contamination, gamete contributor QC, and sibling QC to help improve embryo prioritization for research on *in vitro* fertilization and intracytoplasmic sperm injection.

### Introducing the Ion AmpliSeq Polyploidy SNP QC Panel Kit

The Ion AmpliSeq™ Polyploidy SNP QC Panel Kit enables germline SNP detection from embryo biopsy samples and is available as an optional add-on to PGT-A analysis with Ion ReproSeq PGS kits. Genetic insights are provided by 74 microhaplotype amplicons for human identification and 368 single SNP amplicons for population-wide representation derived from minor allele frequencies in the 1000 Genomes Project. With comprehensive coverage of >500 SNP sites, the Ion AmpliSeq Polyploidy SNP QC Panel Kit can unlock genomic variations unique to each sample for premium PGT-A analysis.

### Gain more genetic insights for better outcomes

Aneuploidy analysis provides valuable information for embryo prioritization research. Now you can boost PGT-A analysis with embryo-specific insights that are available with SNP detection. Additional SNP-based QC features available with the Ion AmpliSeq Polyploidy SNP QC Panel Kit include:

- **Triploidy (69,XXX) identification**—tri-allelic microhaplotype sites and allele frequency distribution can help identify samples with lethal triploidy 69,XXX—an abnormality that is indistinguishable from 46,XX on conventional PGT-A plots [1]
- **Contamination risk assessment**—tri-allelic microhaplotypes can help assess contamination risk from maternal DNA and sample handling to prioritize samples with low risk of altered calls
- **Sibling embryo tracking**—SNP clustering can help confirm that tested samples come from full genetic siblings
- **Gamete contributor analysis**—verify each embryo research sample is associated with intended oocyte and sperm gamete contributors, confirming biological consistency

# Gain more genetic insights for future reproductive decisions by adding the Ion AmpliSeq Polyploidy SNP QC Panel Kit to your PGT-A analysis

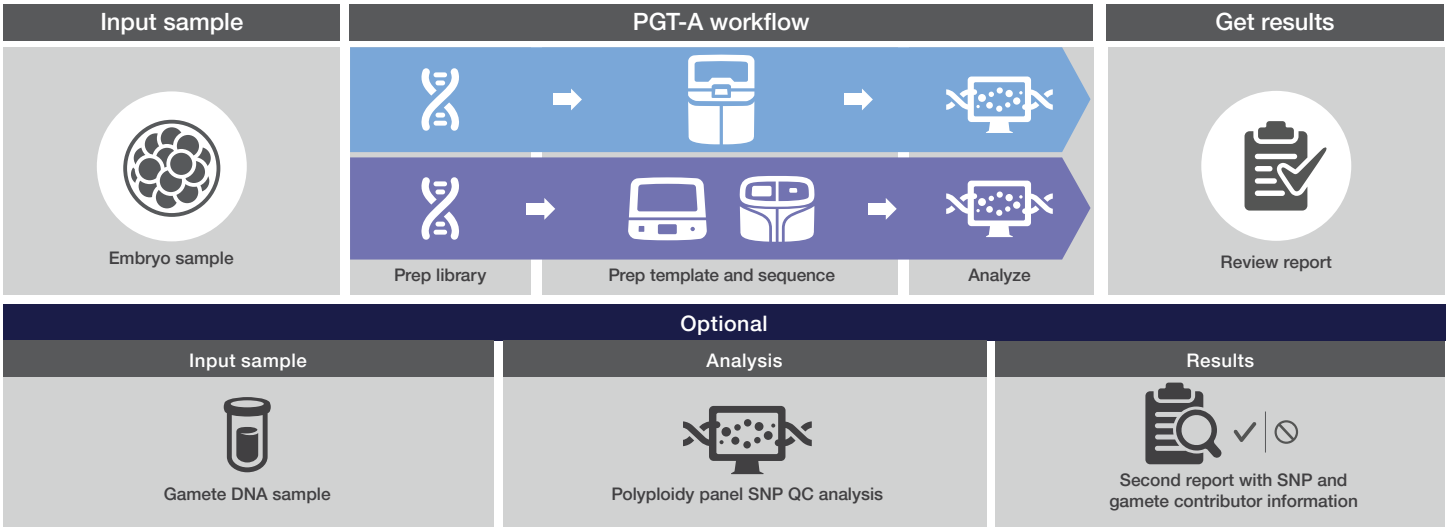
## Complete PGT-A solution on our NGS systems

Ion Torrent™ Genexus™ and Ion GeneStudio™ systems offer complete, sample-to-report workflows for PGT-A with optional SNP-based quality controls. Starting with libraries generated from embryo biopsy samples, the suite automates templating, sequencing, and analysis of whole genomes (for PGT-A) and targeted amplification (for QC). The workflow includes automatic data analysis and custom reporting, making it easy for labs to perform NGS, regardless of experience.

## Supporting you now and in the future

Thermo Fisher Scientific is here to help you get started and keep you up and running. Our extensive options for ongoing training include virtual instruction, classroom instruction, or hands-on learning in your lab to further your knowledge of PGT-A and instrument operation. You can also receive exclusive priority access to global service and technical support teams with priority technical support.* **Contact a reproductive health specialist** to learn more.

* Priority technical support add-on is only available in selected areas. Contact your service representative for details.




Note: SNP, single-nucleotide polymorphism; QC, quality control

**The complete PGT-A workflow with Ion ReproSeq PGS kits and the Ion AmpliSeq Polyploidy SNP QC Panel. This workflow easily combines aneuploidy analysis with SNP-based quality controls on a single platform.**

## Ordering information

| Description | Quantity | Cat. No. |
|---|---|---|
| **Preimplantation genetic testing** | | |
| Ion ReproSeq PGS Kit with GX5 Chips (24 samples/lane) | 192 samples | GX34902 |
| Ion ReproSeq PGS Kit with GX5 Chips (48 samples/lane) | 384 samples | GX34903 |
| Ion AmpliSeq Polyploidy SNP QC Panel Kit with GX5 Chips | 384 samples | GX55689 |
| Ion SingleSeq Barcode Plate Set GX 101–196 and Reagents Kit | 96 reactions | A50250 |
| Ion SingleSeq Barcode Plate Set GX 201–296 and Reagents Kit | 96 reactions | A50251 |
| Ion SingleSeq Barcode Plate Set GX 301–396 and Reagents Kit | 96 reactions | A50252 |
| Ion SingleSeq Barcode Plate Set GX 401–496 and Reagents Kit | 96 reactions | A50253 |
| 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 |
| Ion AmpliSeq Polyploidy SNP QC Panel | 96 samples | A55688 |
| **Sequencing systems** | | |
| Genexus Integrated Sequencer | | A45727 |
| Ion GeneStudio S5 System | | A384194 |
| Ion GeneStudio S5 Plus System | | A384195 |
| Ion GeneStudio S5 Prime System | | A384196 |
| 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 |
| **Analytical validation services** | | |
| Ion ReproSeq Complete Validation Solution | | A47476 |
| **Priority Technical Support Plan for reproductive health solutions** | | |
| Priority Technical Support Plan for the Genexus Integrated Sequencer | 1 year | ZGLPSCGENEXUS |
| 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** | | |
| Ion ReproSeq Training Package | | TRN00362 |

1. ESHRE PGT-SR/PGT-A Working Group; Coonen E, Rubio C, Christopikou D, et al. ESHRE PGT Consortium good practice recommendations for the detection of structural and numerical chromosomal aberrations. *Hum Reprod Open.* 2020 May 29;2020(3):hoaa017. doi: 10.1093/hropen/hoaa017. PMID: 32500102; PMCID: PMC7257111

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