

## Reproductive health

# Deliver premium PGT-A analysis

## Add 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, maternal contamination, and sibling QC to help improve embryo prioritization for research on *in vitro* fertilization and intracytoplasmic sperm injection.

### The Ion AmpliSeq Polyploidy Panel Kit

The Ion AmpliSeq™ Polyploidy 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 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 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]
- **Maternal contamination risk assessment**—tri-allelic microhaplotypes can help assess contamination risk from oocyte DNA 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

# Gain more genetic insights by adding the Ion AmpliSeq Polyploidy Panel Kit to your PGT-A analysis

## A premium PGT-A solution on Ion Torrent NGS systems

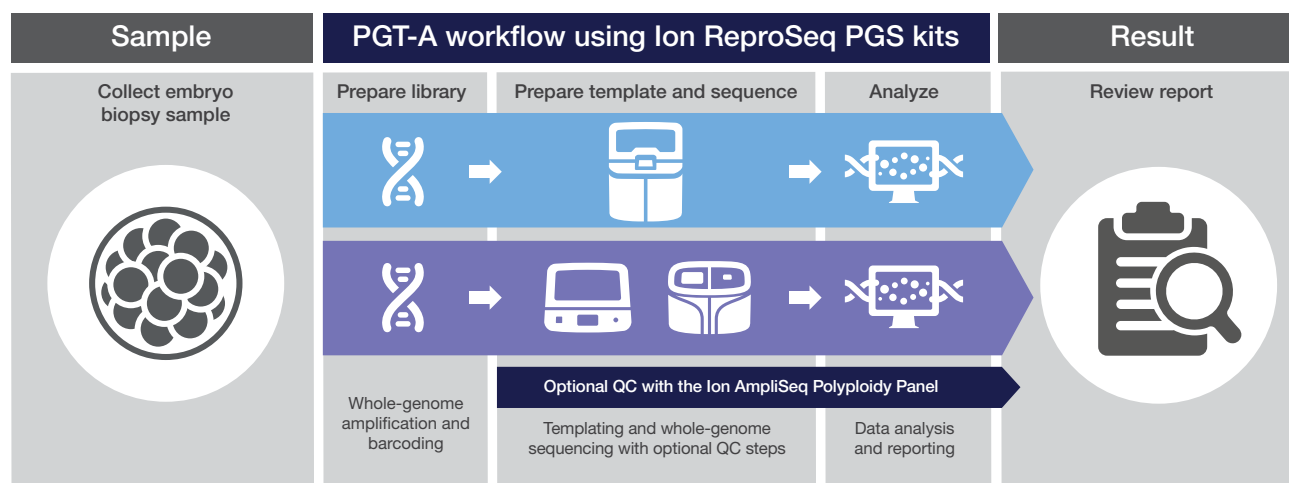
**Ion Torrent™ NGS systems** are part of a complete, sample-to-report workflow for PGT-A with optional SNP-based quality controls. Starting with libraries generated from embryo biopsy samples, the **Ion Torrent™ Genexus™ Integrated Sequencer** or **Ion GeneStudio™ S5 system** easily integrates sequencing and analysis of whole genomes (for PGT-A) and targeted amplification (for QC) in one automated step. Their workflows include automatic data analysis and custom reporting with built-in software, making it easy for any lab to perform NGS, regardless of expertise.

## 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.



A premium PGT-A workflow on the the Genexus sequencer (blue) or Ion GeneStudio S5 systems (purple) with Ion ReproSeq PGS kits and the Ion AmpliSeq Polyploidy Panel. This workflow easily combines aneuploidy analysis with SNP-based QCs by integrating sequencing and analysis in a single platform.

## Ordering information

Description	Quantity	Cat. No.
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 Panel Kit with GX5 Chips	384 samples	GX55689
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 Panel Kit with Ion 520 Chips	80 samples	GS45691
Ion AmpliSeq Polyploidy Panel Kit with Ion 530 Chips	480 samples	GS45690

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.

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