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End-to-end solution for PGT-M.

PG-Seq™ Core Panel provides library preparation reagents and data analysis software for smooth implementation of PGT-M on Illumina® and Element Biosciences® sequencing platforms. This research panel analyzes 8 commonly targeted genes in monogenic disorders:

- BRCA1 and BRCA2 genes
- CFTR gene
- DMD gene
- HBB gene
- F8 gene
- FMR1 gene
- GJB2 gene

Take linkage-based PGT-M to the next level

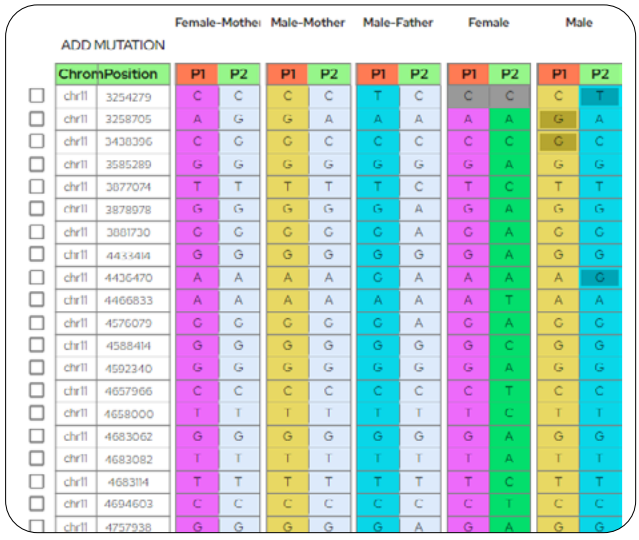
The PG-Seq™ Core Panel analyzes over 200 SNPs located 2 Mb upstream and downstream of the gene or region of interest, covering a total of 4 Mb. Bioinformatics tools are employed to select SNPs with the highest probability of being informative. Additionally, full sequencing of the gene of interest is incorporated to facilitate direct analysis of the variants under study. Most of the pathogenic and likely pathogenic variants in splicing and UTR regions described on ClinVar are included.

Powered by Journey Genomics, the analysis software allows tracking of allele dropouts, recombination events and direct and indirect testing of common variants (SNV or indels) to determine embryo status. It also allows sex determination, analyzing SNPs in X and Y chromosomes, including variants in SRY gene.

JOURNEY GENOMICS

Key features

- Simultaneous screening of 8 frequently studied monogenic disorder genes.
- Enables tracking of allele dropout events
- Generate dense SNP haplotypes
- Easy to interpret software
- Optimized for PG-Seq™ Rapid kit v2 workflow



Cutting-edge workflows for best results

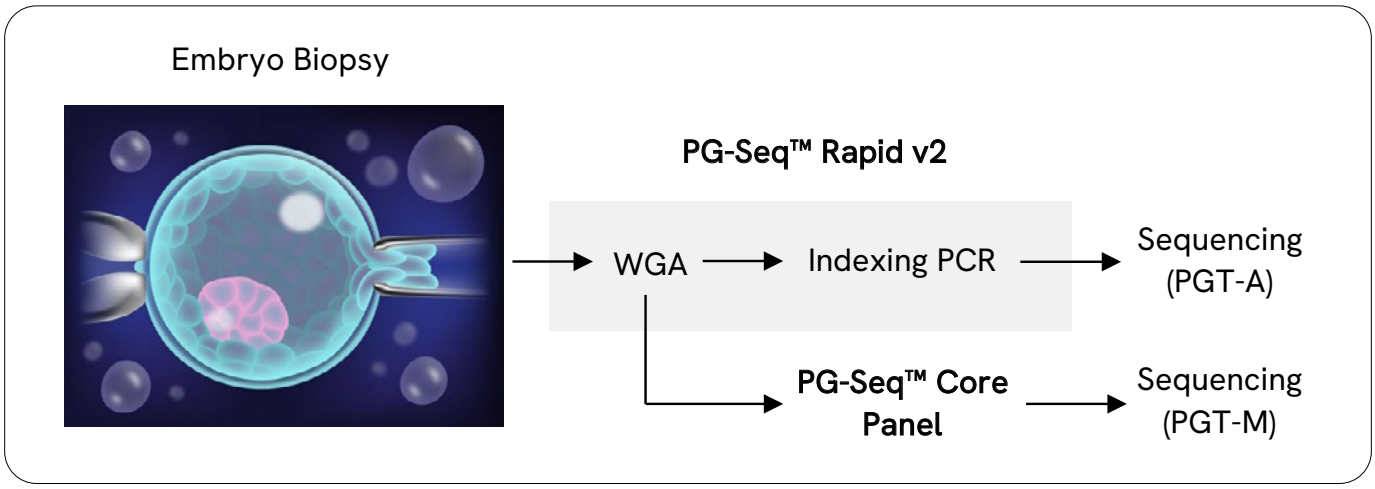


Figure 1: PG-Seq™ Rapid v2 includes all required reagents from cell lysis, whole genome amplification (WGA), indexing along with analysis software for automatic calling of aneuploidies and copy number variants. PG-Seq™ Core Panel uses as input WGA product from PG-Seq™ Rapid v2 or other kits, and includes all necessary reagents, barcodes and software for library preparation and automatic calling of variants. A single biopsy can be used for simultaneous PGT-A and PGT-M research.

References

1. Cost effectiveness of in vitro fertilisation and preimplantation genetic testing to prevent transmission of BRCA1/2 mutations. Doi: 10.1093/humrep/dez203
2. Cystic fibrosis, Duchenne muscular dystrophy and preimplantation genetic diagnosis. Doi: 10.1093/humupd/2.6.531
3. Preconception carrier screening and prenatal diagnosis in thalassemia and hemoglobinopathies: challenges and future perspectives. Doi: 10.1080/14737159.2017.1285701
4. Four Decades of Carrier Detection and Prenatal Diagnosis in Hemophilia A: Historical Overview, State of the Art and Future Directions. Doi: 10.3390/ijms241411846
5. Impact of FMR1 Pre-Mutation Status on Blastocyst Development in Patients Undergoing Pre-Implantation Genetic Diagnosis. Doi: 10.1159/000455849
6. Preimplantation genetic diagnosis (PGD) for nonsyndromic deafness by polar body and blastomere biopsy. Doi: 10.1007/s10815-009-9335-5
7. ESHRE PGT Consortium good practice recommendations for the detection of monogenic disorders. Doi: 10.1093/hropen/hoaa018

Explore the next generation of library prep solutions for PGT

Application	Kit Information
PGT-A	PG-Seq™ Rapid v2
PGT-M	PG-Seq™ Core Panel

Learn more about Journey Genomics

For research use only. Not for use in diagnostic procedures.