PGT-SR

PGT for Chromosomal Structural Rearrangements

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Embryos of couples carrying chromosomal structural rearrangements (resiprocal translocation, Robertsonian translocation or inversion) might be abnormal and infertility/ subfertility problems might be observed in some cases. In these cases, selection of balanced/ normal embryos is possible with PGT-SR using FISH or NGS technology depending on the karyotype of the couple.

In balanced chromosomal rearrangement carriers PGT with NGS not only detects unbalanced chromosomal rearrangements due to chromosomes involved in translocation, but also detects other chromosomal aneuploidies.

> ***"Beware of the interchromosomal effect :***
>
> ***PGT-SR with NGS allows screening of all 24 chromosomes in addition to translocation of interest."***

Process of PGT-SR

Preimplantation Genetic Testing for Chromosomal Structural Rearrangements

1 PATIENT COUNSELING
Case should be registered with the karyotype report of the couple. Geneticists decide on the method (FISH or NGS based PGT-SR) appropriate for each specific case depending on the karyotype report

2 PGT-SR
PGT-SR is performed and results are shared with the IVF center

3 EMBRYO TRANSFER
Euploid embryo transfer is performed and if available, additional euploid embryos are vitrified upon request of the family for future use.

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In preimplantation genetic test of segmental aneuploidies, NGS technology can theoritically detect 10 mb partial deletions and duplications. Using patient tailored bioinformatic algorithms, enriched amplification and deep reading we can detect translocations as small as 2 mb.

As shown in the diagram below, while the small breakage region in chromosome 4 could not be detected using standard bioinformatics evaluation, it could be detected using adjusted algorithms during analysis.

> *"We provide solutions for chromosomal abnormalities; even as small as 2 mb."*