Preimplantation Genetic Testing-Structural Rearrangement [PGT-SR]

Preimplantation genetic testing is performed on embryos created through in-vitro fertilization (IVF). An embryo is created after an egg is fertilized by a sperm. After fertilization, embryos develop from a single cell into a cluster of around 200 cells. In order to perform the genetic analysis, a few cells are biopsied from the embryo and sent to a genetic testing laboratory.

PGT for structural rearrangements (PGT-SR) is specifically designed for individuals who have chromosomal rearrangements and a higher chance of producing embryos with chromosomal differences. The goal of PGT-SR is to identify embryos with the lowest chance of having a chromosomal condition, which helps improve implantation rates, reduce miscarriage rates, and lower the risk of a pregnancy with a chromosomal condition.

What are chromosomes ?

Our bodies rely on about 20,000 genes to guide our growth and development. These genes are packed into structures called chromosomes within our cells. Each chromosome carries around 500-1000 genes. Most people have a total of 46 chromosomes. However, sometimes there can be a missing or extra chromosome, which can cause extra or missing genes. This imbalance may lead to conditions like heart defects or intellectual disabilities.

What is a chromosomal rearrangement?

A chromosomal rearrangement refers to a situation where an individual has the normal amount of chromosomal material required, but the chromosomes are arranged differently than usual. This rearrangement can affect the way chromosomes are distributed during the formation of reproductive cells (eggs or sperm). As a result, when a person with a chromosomal rearrangement attempts to conceive a pregnancy, there is a higher chance of passing on an abnormal number of chromosomes to the embryo.

This increased risk of passing on too many or too few chromosomes can lead to a higher likelihood of miscarriage or the birth of a child with a chromosomal condition. However, preimplantation genetic testing for structural rearrangements (PGT-SR) provides an option to reduce these risks. By using PGT-SR, embryos can be analyzed to identify those with the desired number of chromosomes, thus increasing the chances of a successful pregnancy with a reduced risk of chromosomal conditions.

What are the limitations of PGT-SR?

It's important to note that PGT-SR is a screening test and may have false negative or false positive results. Additionally, the cells analyzed during PGT-SR come from the part of the embryo that would become the placenta and supporting structures, not directly from the future baby. Therefore, testing for chromosomal conditions during pregnancy or after birth may still be necessary to obtain a comprehensive assessment.

Resource: Mahdavi, Manijeh et al. "The Clinical Effectiveness of Preimplantation Genetic Diagnosis for Chromosomal Translocation Carriers: A Meta-analysis." Global medical genetics vol. 7,1 (2020): 14-21. doi:10.1055/s-0040-1712455

For more information, please speak to your healthcare provider.