PGT SR Seq

PGT[SR] is a test developed for carriers of chromosome rearrangements

Chromosome rearrangements involve the movement of sections of chromosome from one place to another. For example, one common type of rearrangement, called a 'translocation', occurs when two chromosomes exchange material between themselves.

Approximately 1 in 500 people carries a **chromosome rearrangement.** People who have a balanced chromosome rearrangement in their cells are usually healthy, but they are at increased risk of producing pregnancies where the fetus has pieces of chromosome lost or duplicated. This loss or duplication of parts of a chromosome often leads to miscarriage, or the birth of children with disabilities.

1 IN 500 PEOPLE CARRIES A CHROMOSOME REARRANGMENT

Translocations

Fragments of two chromosomes are broken and exchanged. In this case the exchange does not involve any gene gain or loss for that chromosome, they continue functioning normally.

Fragments of two chromosomes are broken and exchanged. In this case there is a gene loss/gain in the chromosomes resulting from unbalanced chromosome material in the genome.

Robertsonian translocations are the most common type of structural rearrangements wherein a certain type of chromosome (acrocentric) fuses with another such chromosome resulting in a long chromosome. Such translocations may result in a loss or gain of either of the fused chromosomes when dividing in egg or sperm cell.

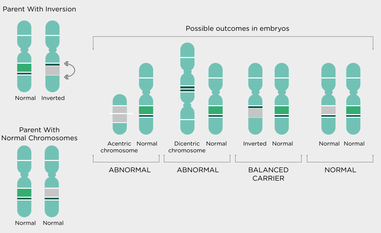
Deletions

A breakage in the DNA sequence can result in a loss or deletion of a chromosome fragment.

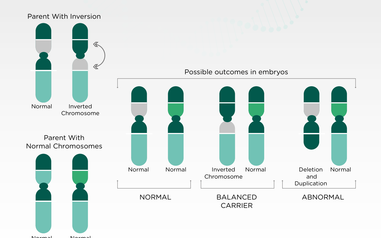
Inversions

When two breaks occur on the same chromosome, and the resulting fragment rotates before inserting back into the chromosome, this leads to 'inversions'. When eggs or sperm are produced, the normal and inverted copies of the chromosome come together and exchange pieces. The presence of the inversion can result in a proportion of the gametes to lose parts of the affected chromosome, while duplicating other areas.

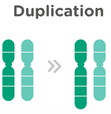
Paracentric inversion



Pericentric inversion



Duplication



Duplication is when a segment of a chromosome is doubled-up, meaning there will be extra copies of the genes in the affected region.

Insertion

Insertion is when a fragment of a chromosome is detached from its original place and inserted into another chromosome.

Who should consider PGT[SR]Seq?

PGT[SR] is for couples where one partner is known to carry a balanced structural rearrangement. Types of rearrangement that can be tested include Robertsonian or reciprocal translocations and pericentric or paracentric inversions.
Juno Genetics' PGT[SR] technology uses next-generation sequencing in order to provide the best possible accuracy rates (exceeding 95%). The highly-validated test assesses thousands of sites on each chromosome, measuring the amount of DNA at each point in order to calculate the number of chromosomes. In many cases, this analysis is supplemented by analyzing variations in the DNA sequence, known as single nucleotide polymorphisms (SNPs), further enhancing the accuracy of the test.

Your partners in genetic health

JUNO GENETICS

supportuk@junogenetics.com