merrion
FERTILITY CLINIC

PGT-SR

Preimplantation Genetic Testing for Chromosomal Structural Rearrangements

Phone: +353 (0)1 5567900

Introduction

PGT-SR, also known as Preimplantation Genetic Testing for Chromosomal Structural Rearrangements is a genetic test performed on embryos. These embryos are usually created through ICSI (Intracytoplasmic sperm injection). PGT-SR can be performed for people with a chromosomal rearrangement to improve the chance of establishing a healthy pregnancy.

Chromosomal rearrangements are changes from the normal size or arrangement of chromosomes, which are structures that hold our genetic material. People with chromosome rearrangements are at an increased risk of producing embryos with the incorrect amount of genetic material, which typically do not lead to a successful pregnancy.

Who is PGT-SR for?

PGT-SR is appropriate for people who have a chromosome rearrangement, and thus are at risk of creating embryos with the incorrect chromosome number or structure. You may consider PGT-SR for chromosome rearrangements if you had a child or pregnancy with a chromosome rearrangement or if you or your partner are a carrier of an:

- Inversion
- Reciprocal translocation
- Robertsonian translocation

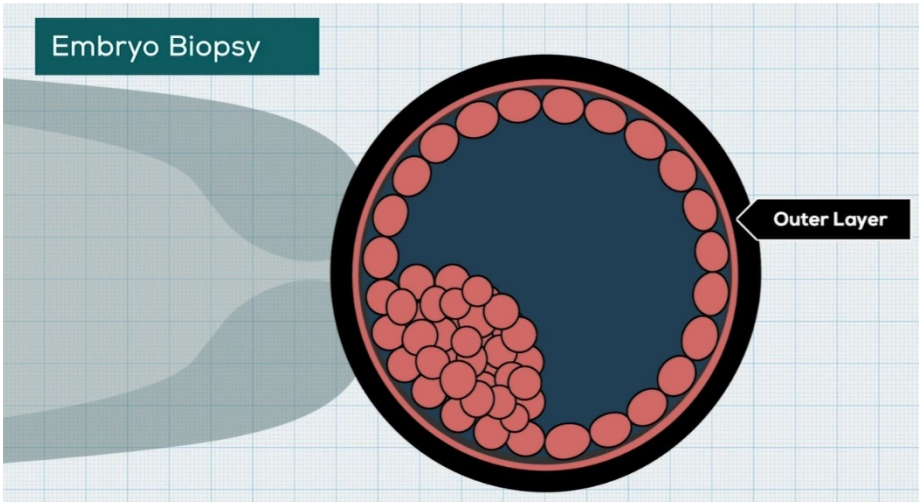
How it works

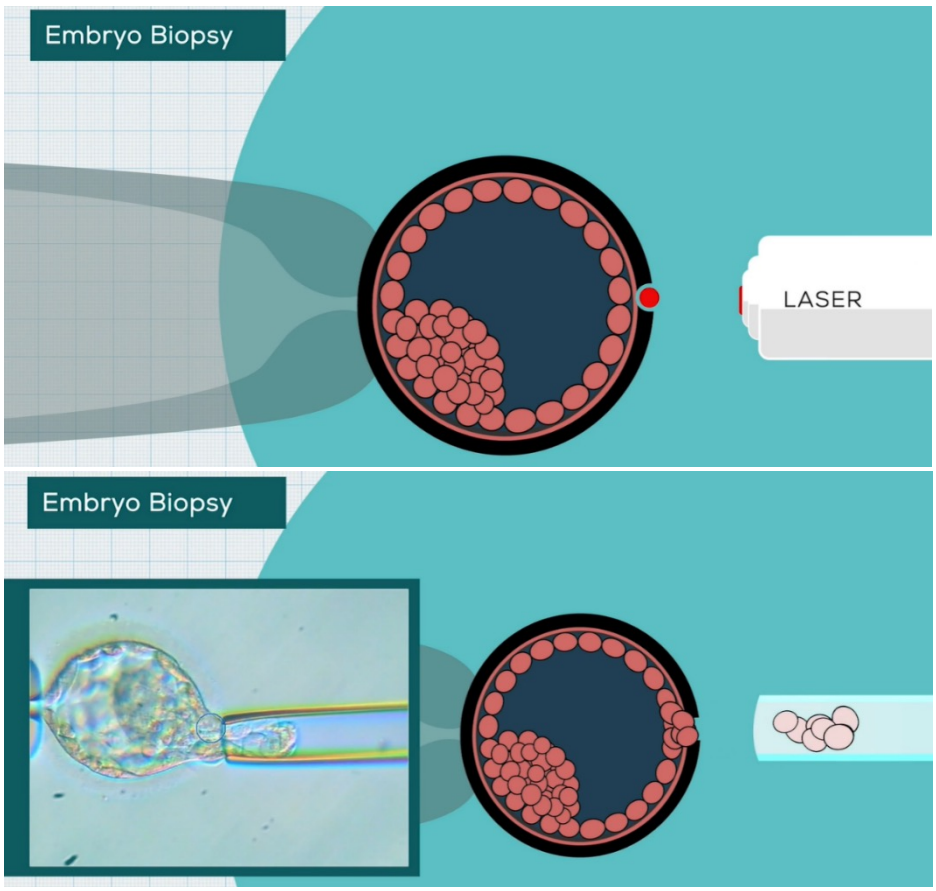
Chromosome rearrangements can be inherited or can happen spontaneously. Many carriers of balanced chromosome rearrangements are healthy and are unaware of their carrier status until they try to have children.

Carriers of balanced rearrangements are at risk for producing embryos with the incorrect amount of chromosomal material, which typically do not lead to a successful pregnancy. PGT-SR can help identify embryos with the correct amount of chromosomal material that are most likely to lead to a successful pregnancy and healthy live birth.

Embryo Biopsy

To perform PGT-SR, a small number of cells are removed from the embryo via a biopsy. This sample is sent to a specialised genetics laboratory for analysis. The lab Merrion Fertility Clinic use is Cooper Genomics, which is based in the UK. Embryos are typically biopsied on day 5, day 6 or day 7 after egg retrieval. Only embryos with satisfactory quality that continue developing to this stage are tested. By this time the embryo contains over 100 cells, and several cells can be removed from the outer layer of the embryo (trophectoderm), which is the part of the embryo that will eventually become the placenta. No cells are taken from the inner cell mass, the part of the embryo that will develop to become the foetus.





(Figure 1. Steps involved in an embryo biopsy for PGT)

In order to most effectively remove these cells without damaging the embryo, a highly skilled embryologist will make a small hole in the shell of the embryo and use an instrument that it less than 1/10th the diameter of a human hair to carefully perform the biopsy.

The cells obtained from the biopsy are then washed to remove any potential sources of contamination and transferred into small tubes and sent to the CooperGenomics laboratory for analysis. Only the biopsy sample is sent off site. The embryos remain at Merrion Fertility Clinic and are cryopreserved by until the PGT-SR results are returned.

Thus far, babies born after procedures that include embryo biopsy have not had an increased rate of birth defects. Although data has shown that embryo biopsy has no adverse impact on growth or medical outcomes, there is potential for unknown consequences to live born babies.

An embryo may be damaged during biopsy, which will cause it to not be suitable for transfer. With a skilled embryologist, the risk of damaging an embryo is very low. While it is important to understand the risks involved in the process, embryos have been biopsied for over 25 years and have resulted in tens of thousands of healthy pregnancies.

PGT-SR Analysis

Once the biopsied cells arrive at the genetic testing laboratory, PGT-SR is performed to determine whether the biopsied sample has chromosomal rearrangements.

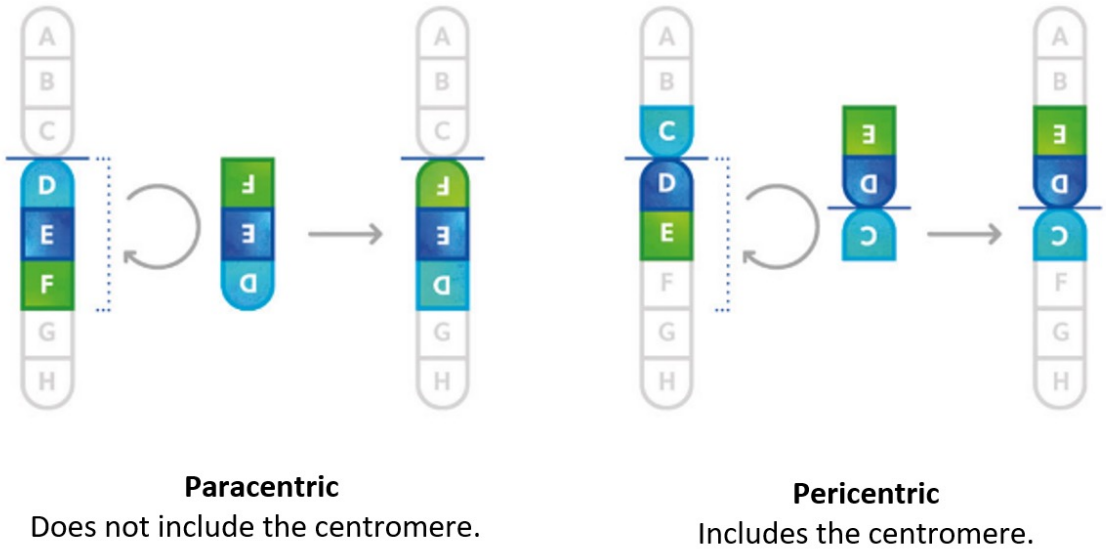
The report from the laboratory will indicate if the embryos could have an optimal chromosomal alignment. These embryos are the most likely to lead to a successful pregnancy and healthy live birth. The results from this test will help the clinical team at Merrion Fertility Clinic determine which embryo to transfer.

Types of Structural Rearrangements

INVERSIONS

Inversions are chromosome rearrangements that involve only one chromosome. In an inversion, a segment of a chromosome is flipped and reinserted upside down. People with an inversion may create embryos with missing or duplicated segments of chromosomes.

An inversion that does not include the centromere is called a paracentric (away from the centre) inversion. Both breaks are in the same arm of the chromosome. An inversion that includes the centromere is called a pericentric (around the centre) inversion.



(Figure 2. Paracentric and Pericentric Inversions)

RECIPROCAL TRANSLOCATIONS

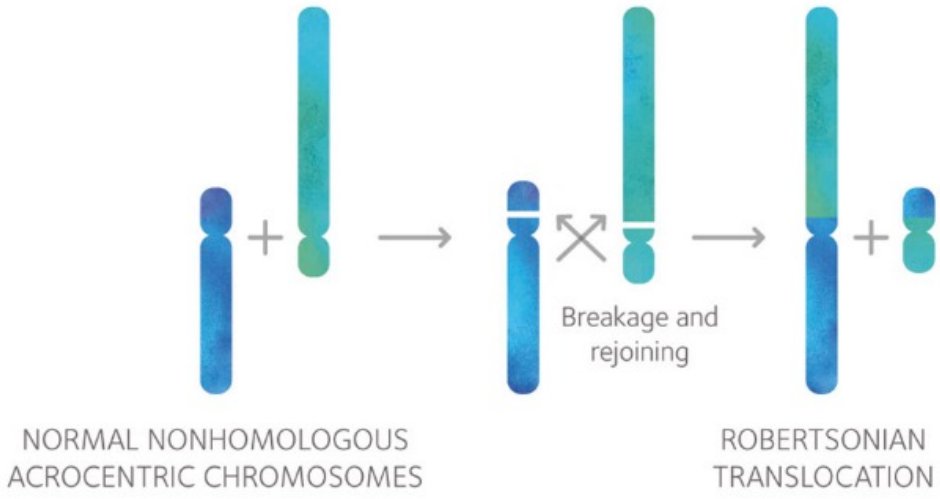
Reciprocal translocations occur when pieces of genetic material break off from two different chromosomes and swap places. People that carry a balanced translocation can create embryos that have either the same balanced translocation, the unbalanced form of the translocation (where there is a gain or loss of chromosomal material), or a completely normal set of chromosomes.

If one parent is a carrier of a reciprocal translocation, a high proportion of embryos may contain an incorrect amount of genetic material depending on the specific reciprocal translocation.

(Figure 3. Reciprocal Translocations)

ROBERTSONIAN TRANSLOCATIONS

Robertsonian translocations occur when two chromosomes join together to form one large chromosome, giving an overall chromosome count of 45 instead of 46. This pairing occurs most commonly between chromosome numbers 13/14 and 14/21, and usually results in conditions such as Translocation Down syndrome, trisomy 13, or uniparental disomy (UPD).



(Figure 4. Robertsonian Translocations)

Embryo Transfer

The benefit of PGT-SR is that by knowing whether an embryo is chromosomally normal or not, you can avoid transferring an abnormal embryo. With the information provided by PGT-SR, it is recommended that a single embryo is transferred as the pregnancy rate per transfer is increased and the miscarriage rate is decreased. With PGT-SR, the risk of an ongoing pregnancy having a chromosomal problem is significantly reduced.

PGT-SR Limitations

PGT-SR cannot guarantee the birth of a chromosomally or genetically normal child and is not a replacement for prenatal testing. In fact, prenatal testing should be considered for all pregnancies resulting from PGT-SR. PGT involves biopsying and testing cells taken from the trophectoderm (the cells that will become the placenta) and not the inner cell mass (the cluster of cells that will become the fetus). Therefore, it is possible that the genetic profile of the cells tested may vary from the cells that will form the fetus.

It's important to understand the benefits, risks, and alternative options, in order to make an informed decision about pursuing PGT-SR. While there is some risk involved, technological advances have made PGT-SR an effective tool for improving IVF success rates.

In extremely rare cases, human error, or natural forces beyond the control of Merrion Fertility Clinic or Cooper Genomics, such as weather and air travel issues, can result in an inability to perform testing and receive results. Remember, even in this rare scenario, only the sample that was biopsied is impacted. Your embryos will still be safe at Merrion Fertility Clinic.

Lastly, sexual intercourse during treatment may result in a spontaneous pregnancy. Sperm from intercourse may result in fertilisation and implantation in addition to, or instead of, fertilisation and implantation from treatment. This would invalidate the results from PGT. Please consult with the Merrion Fertility Clinic team about timing of sexual intercourse during your treatment.

For more information about PGT-SR, please contact Merrion Fertility Clinic and your query will be directed to the team best placed to advise you.