genea
WORLD LEADING FERTILITY

PGT-SR

Pre-implantation Genetic Testing for Structural Rearrangements

What is Pre-implantation Genetic Testing (PGT)?

Genea's PGT scientists have helped hundreds of families maximise the potential of having a healthy baby. Chromosome screening of embryos was launched in 1996, with testing for inherited single gene disorders introduced in 1998. Since then, our PGT techniques have continued to be world leading. Genea is one of the very few centres in Australia that has both IVF and genetics facilities onsite to perform these sophisticated tests successfully.

Embryos are first created via IVF and then Genea's PGT assays are used to select those free from chromosomal issues (such as unbalanced translocations) or familial single gene disorders (eg. cystic fibrosis, spinal muscular atrophy). By choosing embryos which have had genetic screening, you will increase the chances of a healthy ongoing pregnancy and birth of an unaffected child.

Structural Rearrangements can result in an unbalanced number of chromosomes

Chromosomes are the structures in our cells that contain our genes. Having too many or not enough chromosomes can lead to health issues. Some individuals have the correct number of chromosomes, but in a slightly different arrangement. This may be referred to as a balanced translocation or chromosomal inversion. People with balanced chromosomal rearrangements are almost always healthy, as they have the standard amount of genetic information. However, for either male and female patients, having a balanced chromosome rearrangement often results in infertility.

The primary reason why individuals with a balanced chromosomal rearrangement can encounter fertility issues is because they have a higher chance of passing on an unbalanced amount of chromosome information to their embryos. Many unbalanced embryos will not develop properly or fail to implant. They can also establish pregnancies that lead to complications, miscarriage, or birth of a child with health and developmental issues.

If you have been diagnosed with a chromosomal rearrangement, PGT-SR may be an appropriate option for you.

Pre-implantation Genetic Testing for Structural Rearrangements (PGT-SR)

PGT-SR is a technique where embryos are tested to identify whether they have the balanced, standard number of chromosomes. PGT-SR maximises the chance of healthy ongoing pregnancies for carriers of balanced chromosome rearrangements by identifying only chromosomally balanced embryos for transfer. PGT-SR can also often be used for people who have a familial deletion or duplication, which is where a small part of one chromosome pair is either missing or duplicated.

PGT-SR will also detect general aneuploidy, which is an imbalance in the number of chromosomes due to a random error. Aneuploidy risk is particularly associated with nearly all patients, including those without a pre-existing chromosome rearrangement, have some embryos that are aneuploid. The risk of aneuploidy increases with age.

Leading technology to select the best embryo

Every time an IVF cycle produces more than one embryo, embryologists must make a choice about which embryo will be transferred.

Standard IVF
Selection based on appearance of the embryo.

Genea PGT-SR
Selection based on genetic make-up with the appearance of the embryo used as a secondary criteria.

Breakpoint testing: Determining balanced translocation carrier embryos vs non translocation carrier embryos

Some patients would like to know whether their embryos have the same balanced translocation that they have, with the goal to not pass it on to their children. Genea's PGT Scientists can use techniques to identify not only if an embryo has the balanced number of chromosomes, but also if the embryo is free from carrying the balanced translocation. Embryos that do not carry the balanced translocation can then be the first choice for transfer, such that the next generation can avoid the potential reproductive issues associated with balanced translocation carriers.

Uniparental disomy

For certain chromosomes, it is important that an embryo receives one copy from the egg and one copy from the sperm (a paternal and maternal copy). If both copies are inherited from the same parent, this is called Uniparental disomy (UPD) and can be associated with health issues. For carriers of a balanced translocation involving chromosomes where there is a risk of UPD, Genea is able to determine if the embryo has inherited a copy of each chromosome from each parent. Balanced translocation carriers have an elevated risk of producing embryos with UPD.

Your Genea Pre-implantation Genetic Testing (PGT) Treatment Timeline

What are the steps involved to do PGT-SR?

Genetic screening and your individual circumstances should be discussed with your Fertility Specialist before commencing treatment, but as a general guide:

1. Fertility Specialist - After a discussion with a Genea Fertility Specialist, a referral will be sent to the Genea Geneticist and PGT Scientists.

2. Genetics team – An appointment will be booked with a Genea Geneticist or Genetic counsellor after relevant genetic test reports are obtained, including your karyotype results.

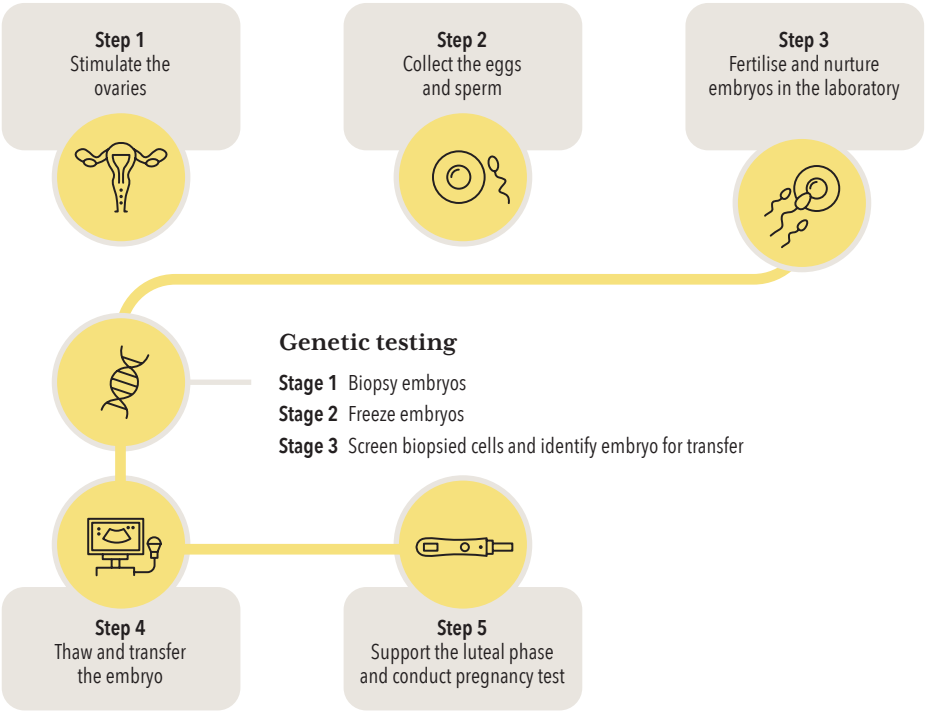
3. Geneticist or Genetic Counselling – The Geneticist or Genetic counsellor will meet with you (via zoom or face to face) to discuss the PGT-SR method and process specifically related to you and your chromosome rearrangement.

4. Nursing Consult* – Your treating Genea clinic will book your nursing consult. Nurses discuss the timeline of your cycle, medications, blood tests and ultrasounds required and what is expected during an egg collection.

5. Start IVF – Your Fertility specialist and treating clinic will let you know the date you can commence an IVF cycle.

*Breakpoint testing and UPD studies (optional): If you elected to have breakpoint testing and/or UPD studies on your embryos, the laboratory may need to design a specific test which may delay the timing of your nursing consult. There are additional fees for UPD and breakpoint testing. You will receive more information about these options during your genetics consult. Most PGT-SR patients will not require a workup.

What is involved in testing of embryos?



Embryos will be grown in the laboratory for 5-6 days until they reach the blastocyst stage. Embryos either start hatching on their own, or require assisted hatching. Assisted hatching is performed either on day 4 or at the time of the biopsy.

The embryology team will assess whether your embryos are suitable for an embryo biopsy and PGT-SR testing and provide you with an update.

An embryo biopsy is performed by removing a small sample of hatching cells (less than 5 per cent) from the trophectoderm (the outside structure of the embryo that becomes the placenta in an ongoing pregnancy). It is normal for some of your embryos to not develop sufficiently and therefore be unsuitable for testing or freezing.

All biopsied embryos will be frozen to allow sufficient time to perform the PGT tests. Please note, a fresh embryo transfer will not be possible if you want all your embryos to undergo PGT-SR testing.

PGT Results

Results will be available approximately three weeks after your embryo is biopsied, at which time a PGT scientist, genetic counsellor, your Fertility Specialist, or an embryologist will call you to discuss the results.

A copy of these results will also be sent to your Fertility Specialist who will discuss your treatment options and next steps.

Results explained

No Abnormality Detected (NAD)
The embryo is not affected with the condition tested, has a balanced number of chromosomes, and is available for transfer.

Abnormal (ABN) / AFF not relevant for PGT-SR
The embryo is affected with the condition tested and/or has an unbalanced number of chromosomes. It will not be available for transfer.

Mosaic Embryo
This means that some cells in the embryo have the wrong number of chromosomes and some cells have the standard number of chromosomes. Mosaic embryos have a reduced chance of implantation and live birth and higher chance of miscarriage. Outcome data shows that mosaic embryos have the potential to lead to a healthy ongoing pregnancy and Genea allows transfer of mosaic embryos after a detailed discussion with one of our Genetic Counsellors. The vast majority of mosaic embryos are due to random errors, they are not caused by chromosomal rearrangements.

Unknown (UNK)
No result was obtained due to sample or testing issues. You may be asked if you would like to re-biopsy the embryo and try to test again. This will reduce the quality and success rates of transferring this embryo.

Rarely, there may be results where there is uncertainty and further testing or interpretation may be required, which will be discussed with you.

Clinical interpretation provided in the results will be current as of the date provided. Clinical interpretation of test results can change over time with emerging data and/or changes in professional organization guidelines. Genea is under no obligation to update results in response to these changes.

Can I transfer an abnormal embryo if I have no other embryos to transfer?

It is a Genea policy that for an embryo that has been tested and reported as abnormal or affected, this embryo is not available for transfer.

Prenatal screening - Should you have prenatal diagnostic tests done?

Whilst PGT-SR does have a high degree of accuracy, it will never be 100 percent due to various biological factors and technical difficulties that can be experienced. Our geneticist or genetic counsellor can provide more detail on the accuracy based on your individual circumstances. We recommend that prenatal screening and diagnostic tests are discussed with your referring fertility doctor.

What you need to know

- Testing will reduce the number of embryos available for transfer, but will ensure those that are transferred have the best chance of survival and a healthy pregnancy.
- You may have an embryo that is not suitable for testing. Some untestable embryos may be able to be transfered after a discussion with your Fertility Specialist, or frozen for future untested transfer.
- Embryos must be frozen pending test results, which are usually available in 2 weeks.
- It is possible that all your embryos may have unbalanced chromosomes, resulting in no embryos being suitable for transfer from your IVF cycle. Embryos reported as abnormal will not be available for transfer at a Genea clinic and will be discarded as per current local regulations.
- Due to technical limitations of the testing process, it is possible that some embryos do not achieve a result.
- An embryo may return a result indicating a mixture of abnormal and normal cells (referred to as mosaicism) and may be able to be transferred after a discussion with a genetic counsellor.
- The test does not detect all types of genetic abnormalities and standard prenatal screening is recommended in an ongoing pregnancy.
- There is no guarantee a tested embryo will result in a pregnancy.
- Unless specifically requested, abnormal embryos will be discarded as per current local regulations regarding the transfer of affected embryos.

If you have any questions, please do not hesitate to contact a Genea Genetic Counsellor on **(02) 8484 6548**.

For more detailed information about costs, suitable Genea Fertility Specialists, and next steps please contact the Fertility Concierge on **(02) 8484 7666**.

1300 361 795
genea.com.au

GEN08023