genea
WORLD LEADING FERTILITY

Pre-implantation Genetic Testing – Aneuploidy

PGT-A embryo screening for chromosomal conditions

Many embryos created either via natural conception or IVF will have chromosomal changes (aneuploidy), which adversely affects the likelihood of having a healthy pregnancy and birth. As well as increasing the likelihood of failed implantation or miscarriage due to the pregnancy being non-viable, aneuploidy can also result in a baby being born with a chromosomal condition which can be associated with health and developmental issues in a child.

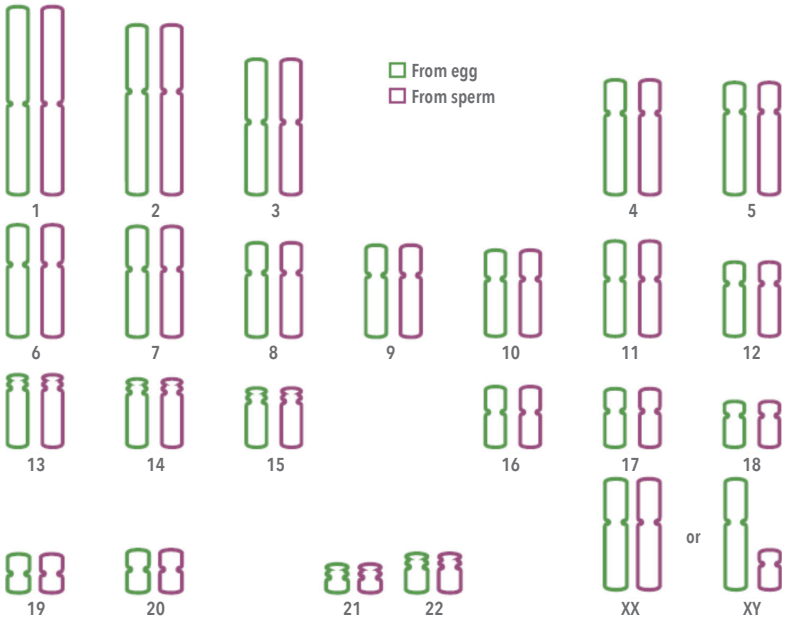
Preimplantation genetic testing for aneuploidy (PGT-A) is a way to check that an embryo has the correct number of chromosomes.

By excluding embryos identified as having the wrong number of chromosomes, PGT-A can maximise the chance of success per embryo transfer and shorten the time it takes to establish a pregnancy.

What are chromosomes?

Chromosomes are the packages of genetic information within our cells. Usually, our cells contain 46 chromosomes for which the scientific term is euploid. We receive half (23) from the egg and half (the other 23) from the sperm. This means we have a total of 23 pairs of chromosomes. The pairs are numbered 1-22, while the last pair are the sex chromosomes (X and Y). Females typically have two X chromosomes (46,XX) and males typically have an X and a Y chromosome (46,XY).

Below is an image showing the typical set of male and female chromosomes.



What is aneuploidy?

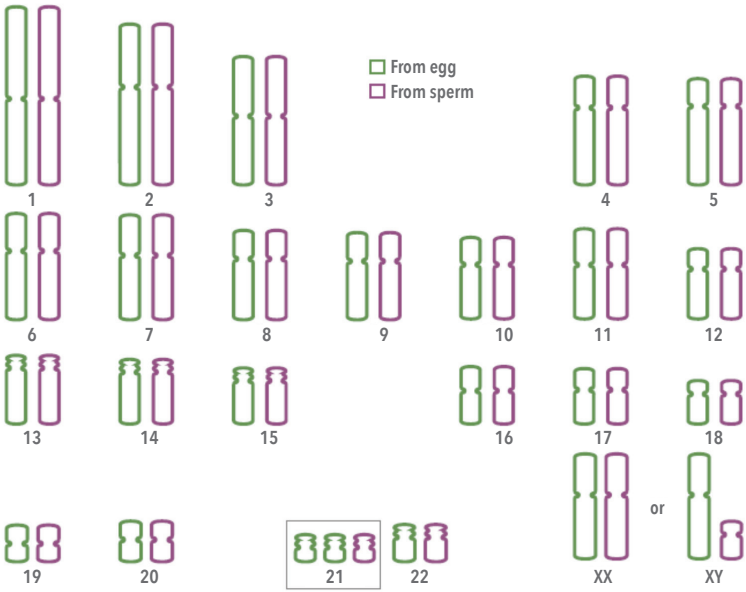
Chromosomes contain our genetic information. Having too many or too few chromosomes means there is the wrong amount of genetic information and this is called aneuploidy. Random errors can result in a baby being born with health and developmental problems, and is the most common reason for embryos to either not implant or result in a miscarriage.

It is estimated that 50 per cent of miscarriages that occur in the first trimester are linked to aneuploidy.

The chance of an embryo being aneuploid increases based on:

- maternal age;
- a parental chromosome rearrangement called a balanced translocation;

One of the most common chromosomal conditions is trisomy 21 (caused by an extra, third copy of chromosome 21, as illustrated below). This is also know as Down syndrome.



How does Genea PGT-A reduce the chance of transferring an embryo with aneuploidy?

Through the exclusion of embryos identified with aneuploidy, PGT-A can maximise treatment outcomes per embryo transfer.

In a standard IVF treatment, scientists and Fertility Specialists choose which embryo to transfer. This is based on information from microscopic inspection of the development and appearance of the embryos, to identify those embryos likely to result in a pregnancy. Unfortunately, many aneuploid embryos have a normal appearance and cannot be identified by microscopic inspection.

PGT-A gives them another tool to help Genea's scientists make this decision by examining the chromosomal health of each embryo.

PGT-A helps Genea to exclude embryos that contain an obvious chromosomal change that would make it unlikely to result in an ongoing healthy pregnancy.

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.

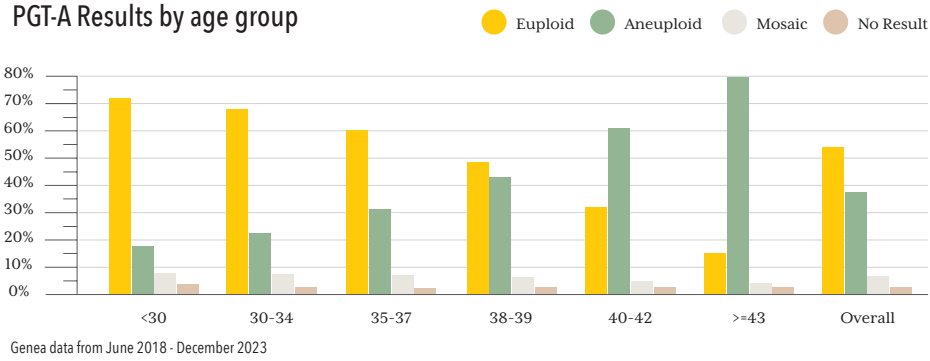
Genea PGT-A
Selection based on genetic make-up with the appearance of the embryo used as a secondary criteria.

What is involved in embryo testing using PGT-A?

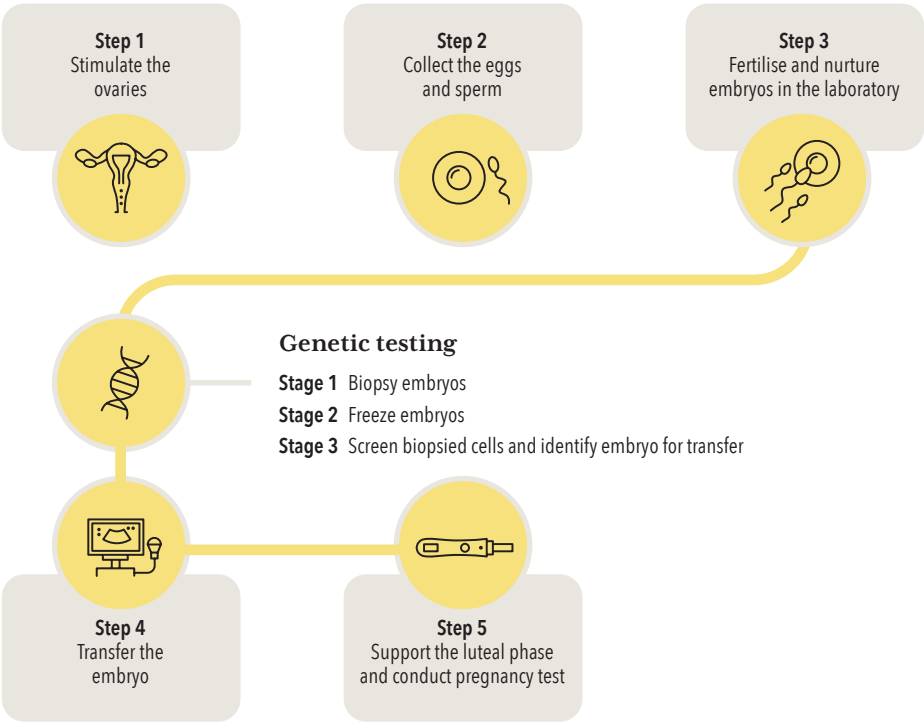
Genetic screening and your individual circumstances should be discussed with your Fertility Specialist before commencing treatment, but as a general guide:

1. After a discussion with your Fertility Specialist, advise them if you wish to have PGT-A performed on your embryos created via IVF.
2. Embryos will be grown in the laboratory for 5-6 days until they become a hatching blastocyst. Any blastocysts that are not already hatching naturally will undergo assisted hatching. To the best of our knowledge, assisted hatching is not harmful and does not increase the risk of twining. The embryology team will then assess whether your embryos are suitable to be biopsied and PGT-A tested.
3. An embryo biopsy is performed by removing a small sample of cells (less than 5 per cent) from the trophectoderm (the structure of the embryo that becomes the placenta in an ongoing pregnancy).
4. All embryos will be frozen after they are biopsied while we perform testing on the biopsy sample.
5. Results will be available in around three weeks, at which time one of our PGT-A scientists or genetic counsellors will call you to discuss the results.

Please note, if you want a fresh transfer, only untested embryos can be used for this purpose. You can discuss the pros and cons of a fresh versus frozen embryo transfer with your Fertility Specialist in relation to your personal circumstances.



Genea data from June 2018 - December 2023



Results explained

No aneuploidy detected	No chromosomal changes were detected and this embryo is available for transfer.
Mosaic embryo	This means that some cells in the embryo have been identified to 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.
Abnormal	Aneuploidy was detected in all cells tested and this embryo is not available for transfer and will be discarded.
No result	Sometimes the test fails or is inconclusive. These embryos will remain in storage and can be transferred after you sign a consent form. You may also be asked if you would like to retest these embryos.

What is the accuracy of PGT-A?

PGT-A is a screening tool that assists in choosing an embryo to transfer that has the best likelihood of a successful implantation and ongoing pregnancy.

Whilst PGT-A has a high degree of accuracy, there are limitations to this technology and the small possibility of false positive and false negative results. A Genea PGT Scientist can provide you with up to date information about accuracy at the time of testing.

Screening options in a pregnancy after transfer of a PGT-A tested embryo

As PGT-A is not 100 per cent accurate, standard chromosome screening can be considered in an ongoing pregnancy. Prior to the transfer of a mosaic embryo, any specific recommendations for additional testing will be discussed with you.

Pregnancy screening may include non-invasive prenatal testing (NIPT) from about 10 weeks of pregnancy, or combined first trimester screening, which includes a blood test alongside your standard ultrasound around 12 weeks of pregnancy. Please click here for information about your prenatal screening options or go to www.genetics.edu.au/SitePages/Prenatal-testing-booklet.aspx

What you need to know

1. Testing will reduce the number of embryos available for transfer, but will ensure those that are transferred have the best chance of survival
2. You may have an embryo that is not suitable for testing but can be considered for transfer untested
3. Embryos must be frozen pending test results, which are usually available in 2-3 weeks
4. It is possible that all of your embryos may have an abnormality, resulting in no embryos being suitable for transfer from your IVF cycle
5. Due to technical limitations of the testing process, it is possible that some embryos do not achieve a result
6. An embryo may return a result indicating a mixture of aneuploid and normal cells (referred to as mosaicism) and may be able to be transferred after a discussion with a genetic counsellor
7. The test does not detect all types of genetic abnormalities and is not the same as genetic carrier screening.
8. There is no guarantee a tested embryo will result in a pregnancy.

What you need to do next

Review the following timeline of treatment if you would like to consider having embryos tested using PGT-A.

The timeline for PGT-A

1. Before starting your cycle
   - Read the associated information booklet (Your Fertility Journey) and refer your questions to your Fertility Specialist
   - Complete and return your consent form
2. Day of egg collection
   - Confirm with your embryologist that you are still considering PGT-A as an option
   - Confirm with your embryologist which phone number should be called on Day 3 or Day 4 for an update
3. Day 3 or 4 after your egg collection
   - A scientist will contact you to discuss your embryos and the technical aspects you need to consider before proceeding
   - Your embryos will be hatched in preparation for potential testing on Day 5 or Day 6
4. Day 5 after your egg collection
   - If you have not already provided final confirmation that you wish to proceed with PGT-A, a scientist will call you with an update on the suitability of your embryos for testing and also discuss non-testing options with you.
   - If you make the decision to proceed with testing, your embryos will be biopsied and immediately frozen. If you choose to not proceed with testing, your nursing team will liaise with you and your Fertility Specialist to discuss embryo transfer and freezing as part of standard IVF cycle treatment
   - Please note, in some cases embryos that are potentially healthy and could lead to a viable pregnancy may not be robust enough to allow a biopsy. In this instance you might consider an untested embryo transfer or freezing untested embryos (if they are suitable)
5. Approximately 2-3 weeks later
   - A scientist or genetic counsellor will call you to discuss the outcomes from the PGT-A testing. If any embryos are reported as abnormal/aneuploid, these embryos will not be available for transfer and will be discarded.

     These results will also be made available to your Fertility Specialist, who will discuss with you your treatment options and next steps.

If you have any questions, please do not hesitate to contact a Genea scientist on +61 2 9229 6420 for a detailed discussion.

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