Nº1 FERTILITY

PATIENT INFORMATION

Preimplantation Genetic Testing for Aneuploidy (PGT-A)

Formerly known as Preimplantation Genetic Screening (PGS)

What is PGT-A?

Preimplantation genetic testing for aneuploidy (PGT-A), previously known as preimplantation genetic screening (PGS), screens embryos for their chromosome quantity. The term 'aneuploid' describes an abnormal or unbalanced quantity of chromosomes. The term 'euploid' describes a normal or balanced quantity of chromosomes. PGT-A involves taking a small sample of cells from an embryo to determine the quantity of chromosomes contained in that sample.

Including PGT-A in your treatment is your choice, and we encourage patients to contact the Nº1 Genetics Department with any questions about considering PGT-A in their treatment.

What are chromosomes and why are they important?

We are all made of billions of cells, and inside each cell is DNA. DNA is packaged into larger structures called chromosomes, that each contain thousands of genes. Genes provide specific instructions for our body to grow, develop and function.

Chromosomes come in pairs and are numbered by size from 1 through to 22. The 23rd pair are the sex chromosomes that typically determine our biological sex. Usually, two X chromosomes (XX) are found in biological females, and one X and one Y (XY) are found in biological males. We inherit one copy of each chromosome from an egg and one from a sperm, to total 23 pairs (46 total) of chromosomes. Most cells in our body contain a full set of 23 pairs of chromosomes, and each chromosome contains genes essential for our growth, development, and healthy function.

Chromosome imbalances or abnormalities can occur during egg or sperm development, or during early embryo development. Embryos with an incorrect or unbalanced quantity of chromosomes are more likely to result in implantation failure and miscarriage. Approximately 50% of all miscarriages are considered to be the result of abnormal, unbalanced chromosomes. Chromosome abnormalities can occur in any embryo, at any age, however the chance of a chromosomal abnormality in an embryo increases with maternal age (i.e. the age of eggs).

What is the purpose of PGT-A?

The purpose of PGT-A is to determine if an embryo has the normal, balanced quantity of chromosomes, or an abnormal, unbalanced quantity of chromosomes that could impact development. This allows selection and transfer of 'normal' embryos to maximise the chance of a successful pregnancy. At Nº1 Fertility, PGT-A is used to help identify embryos with the best chance of success to potentially reduce the number of IVF cycles and time needed to achieve a pregnancy.

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What is the PGT-A process?

After an egg collection, eggs are fertilised with sperm and grown in the Nº1 IVF lab until the embryo(s) reach hatching blastocyst stage. Embryos typically reach this stage on day 5 or day 6 of development. The Nº1 Embryology Team will contact you with updates regarding the development of your embryos and update you regarding how many embryos were able to be biopsied and frozen.

Embryo biopsies are performed by our experienced Nº1 Embryologists. The procedure involves using a fine glass pipette to take a small sample of cells (3-5) from the embryo. Crucially, the cells are selected from the trophectoderm, a part of the embryo that goes on to form the placenta. All embryos that undergo PGT-A are frozen and stored safely in the Nº1 Fertility Laboratory for potential future transfer. Embryos tolerate the biopsy and freezing procedures well. The biopsied cells are frozen and sent to our external genetic testing laboratory CooperGenomics in the UK for analysis. PGT-A results are available within 2-4 weeks from the date of embryo biopsy.

Importantly, not all embryos are suitable for biopsy. This is due to a variety of factors such as: embryo quality (grade), stage of embryo development, hatching status, and the way in which an embryo is hatching. We encourage you to discuss the biopsy process with the Nº1 Embryology Team for additional, specific information about your embryo.

How does PGT-A work?

The embryo biopsy samples are sent to our external genetic testing laboratory CooperGenomics in London, England. The DNA is extracted from the sampled cells, then copied millions of times for chromosome analysis. Sensitive DNA testing techniques called Next Generation Sequencing (NGS) are used to develop a profile of the chromosomes in the sample. The chromosome profile of the embryo is compared to a standardised set of reference chromosomes to identify if there is an imbalance in the quantity of chromosomes. Using this technology, PGT-A results are reported as ‘normal’ (euploid), ‘abnormal’ (aneuploid), ‘mosaic’ (please see *Patient Information: PGT-A mosaic result* for further information), or ‘no result’ (please see *Patient Information: PGT no result* for further information).

Should I consider PGT-A?

Your Fertility Specialist may recommend PGT-A, or you may request it as part of your treatment plan. Importantly, including PGT-A in your treatment is your decision. At Nº1 Fertility, we recommend consideration of PGT-A for women who are 35 and older because the chance of chromosome abnormalities or imbalances increases with maternal age, however many younger people also choose PGT-A. For example, for eggs used from an individual who is 35, each embryo has a 65% chance of being normal (euploid). In comparison, for eggs used from an individual who is 42, each embryo has a 25% chance of being normal.

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What are the benefits of PGT-A?

PGT-A determines if embryos have the normal, balanced quantity of chromosomes, to therefore select only the 'normal' embryos for transfer and maximise the chance of a successful pregnancy. The potential benefits include:

- Avoiding embryo transfers that would not result in a pregnancy
- Reducing the risk of miscarriage
- Increasing the chance of a live birth from each transfer
- Reducing the time taken to achieve a pregnancy
- Reducing treatment costs with fewer IVF cycles needed to achieve a pregnancy

How accurate is PGT-A?

The accuracy of PGT-A is over 97%. This accuracy rate incorporates technical limitations combined with the understanding that in some cases, the chromosomes in an embryo biopsy sample may not reflect the chromosome complement in the remainder of the embryo. However, most of the time PGT-A results from embryo biopsy are an accurate representation of the chromosomes in the whole embryo.

How are PGT-A results reported?

You will be contacted by a Nº1 Genetic Counsellor to discuss your PGT-A results. PGT-A results are available within 2-4 weeks from the date of embryo biopsy. Results are reported as 'normal' (euploid), 'abnormal' (aneuploid) or 'mosaic' (please see *Patient Information: PGT-A mosaic result* for further information) or 'no result' (please see *Patient Information: PGT no result* for further information).

Why do we use CooperGenomics?

CooperGenomics have multiple genetic testing laboratories in both the USA and UK. They are a global leader in reproductive genetic testing. We have used their services and expertise since the start of Nº1 Fertility and to date, they have performed over 100,000 embryo testing procedures.

What is the cost of PGT-A?

Currently, the cost of PGT-A is $780 per embryo and is capped at $4250 per cycle for 6 or more embryos. For example, if you were able to biopsy 8 embryos in one cycle, it would be capped at $4250.

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What are the risks and limitations of PGT-A?

Every embryo biopsy procedure carries a small risk of damage to the embryo, and in rare circumstances loss of the embryo. Additionally, not all embryos are suitable for biopsy. The experienced Nº1 Embryology team make careful and educated decisions regarding an embryo's suitability for biopsy to preserve the embryo's viability.

Due to testing and process limitations, not all embryo biopsy samples return a PGT-A result. These embryos are reported as 'no result' (please see *Patient Information: PGT no result* for further information).

Embryos with abnormal PGT-A results are unsuitable for transfer.

Importantly, PGT-A is a screening test, not a diagnostic test. This is largely because we are analysing a small number of cells destined to become the placenta from an embryo with approximately 100 cells. This means we cannot clarify the chromosomal composition of a whole embryo. However, PGT-A offers the best estimate currently available. Because PGT-A is a screening test, we would recommend NIPT (non-invasive prenatal testing) for a pregnancy achieved with a PGT-A tested embryo.

What can't PGT-A test for?

PGT-A cannot test for specific genetic conditions. Sometimes individuals can be at risk of having a child with a genetic condition because they are 'silent carriers' for a recessive condition, or they have a family history of a known genetic condition. As PGT-A screens for chromosome quantity in an embryo only, it does not test for specific gene changes associated with genetic diseases. For example, some individuals can be at risk of having a child with a genetic condition because they are carriers for a recessive condition such as cystic fibrosis, Fragile X syndrome and spinal muscular atrophy. Genetic carrier screening is available to determine if you and your partner are 'silent carriers' of any recessive genetic conditions and clarify if you are at risk of having a child with a recessive genetic condition. Some conditions are not caused by genetic factors or may involve a mixture of genetic and non-genetic factors such as spina bifida, autism spectrum disorder and intellectual disability. Such conditions are unable to be tested using PGT-A.

If you are concerned about your family history or specific medical or genetic conditions, please get in touch with Nº1 Genetics Department.

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Does PGT-A replace prenatal testing?

No. While transferring an embryo that has been tested with PGT-A is expected to significantly reduce the risk of having a child with a chromosome abnormality, it does not eliminate this risk. Non-invasive prenatal testing (NIPT) such as *percept NIPT* (please see vcgs.org.au/tests/perceptnipt for further information), is still recommended for pregnancy achieved with a PGT-A tested embryo.

Questions?

If you have read this information and wish to discuss PGT-A further, please contact the Nº1 Genetics Department on 9132 9600 or email genetics@number1fertility.com to organise a time to speak to one of our Genetic Counsellors.

The information provided above is intended for educational purposes only and should not be used as a substitute or replacement for medical advice received from a medical professional. It is important to discuss your individual circumstances and situation with your treating doctor.