

Fact Sheet

Preimplantation Genetic Screening (PGS) with 24 chromosome screening



Key points:

- Error/s in the early development of the sperm, egg or embryo can lead to an abnormal number of chromosomes in the developing embryo (ie: missing or extra chromosomes).
- An abnormal chromosome number can cause implantation failure, miscarriage, or the birth of a child with a chromosome abnormality (eg: Down syndrome).
- Some individuals have an increased risk of producing embryos with an abnormal chromosome number.
- Preimplantation Genetic Screening (PGS) with 24 chromosome screening can be used to screen embryos for abnormalities in chromosome number. Only embryos which are found to be “normal” for the tested chromosomes are considered suitable for transfer to the uterus.
- PGS is NOT 100% accurate. Confirmatory prenatal diagnosis is highly recommended if a pregnancy is achieved following PGS.

What is 24 Chromosome Screening?

An individual's genetic information is packaged into strings of DNA called chromosomes. Normal embryos contain 46 chromosomes, or 23 chromosome pairs. These chromosome pairs are labelled 1 to 22 (the autosomes) and X and Y (the sex chromosomes). Some embryos can have an abnormal number of chromosomes (ie: missing or extra chromosome/s) due to errors in cell division in the developing egg, sperm or embryo. This is known as chromosomal aneuploidy. An aneuploid embryo will fail to implant, miscarry, or result in the birth of an affected child. PGS with 24 chromosome screening can be used to screen for aneuploidy involving any chromosome.

This testing may be appropriate for:

- Individuals with advanced maternal age (>36 years)
- Individuals who have experienced repeated miscarriage
- Individuals who have experienced repeated IVF failure
- Individuals who have previously had a pregnancy with a chromosomal abnormality
- Individuals where one partner has an altered sex chromosome complement (eg: XXY)

PGS with 24 chromosome screening is designed to analyse chromosome copy number. The test does not give any information relating to other genetic conditions or other abnormalities. There is a 3-5% background population risk for birth defects or genetic conditions in any pregnancy. 24 chromosome screening is only designed to detect a majority of the birth defects caused by aneuploidy and not these other risks.

What is involved?

Embryos are created using a fertilisation method called Intracytoplasmic Sperm Injection (ICSI), which involves the injection of a single sperm into the egg. ICSI is specifically used in these cases to reduce the risk of misdiagnosis due to the presence of additional sperm around the egg/embryo. Embryo biopsy is typically performed on day 5/6 (please refer to the “Preimplantation Genetic Diagnosis” fact sheet for further information relating to embryo biopsy).

The biopsied cells are transferred to a small test tube for genetic testing. Due to the time taken to perform the genetic testing, the embryos must be frozen following biopsy. It is important that patients are aware that some embryos will not be considered suitable for biopsy, some embryos will not be considered suitable for freezing, and some embryos may not survive the freeze/thaw process. Final results are usually available 3 weeks after biopsy. A Genetic scientist/Embryologist will discuss the PGS results with the patient at the completion of testing.

If an embryo is found to have the correct chromosome number and is considered genetically suitable for transfer, it can be thawed for use in a frozen embryo transfer cycle. The patient's IVF nurse will organise a pregnancy test to be performed on Day 16 of the frozen embryo transfer cycle. This process should increase the chance of a successful pregnancy and significantly reduce the risk of miscarriage. Surplus “normal” embryos will remain in storage. These embryos may be used in a subsequent cycle. Chromosomally abnormal embryos will be removed from storage and allowed to succumb.

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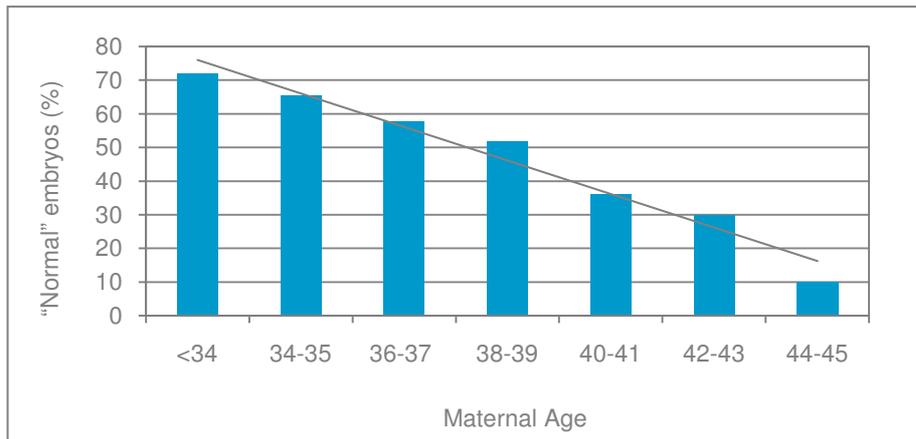
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What are the expected outcomes?

It is well documented that the frequency of chromosomal aneuploidy increases with maternal age. Therefore, older women will be less likely to obtain a “normal” embryo for transfer compared with younger women (Figure 3). Encouragingly, data indicates that once a “normal” embryo is identified for transfer following 24 chromosome screening, the pregnancy rate in older women is not significantly different from that of younger women.

Figure 3: Percentage of day 5/6 embryos diagnosed as chromosomally “normal” following 24 chromosome screening.



Other important information

- Due to the complexity of PGS, it may not be possible to obtain a conclusive result for some or all embryos (or for some chromosomes). In this case, the embryo/s can either be thawed and transferred without a genetic result (or with an incomplete genetic result), thawed and re-biopsied if it/they reach an appropriate stage of development, or thawed and allowed to succumb.
- It is possible that at the completion of the cycle there will be no embryos available for transfer. This may occur as a result of one of the following scenarios:
 - All embryo samples tested during an IVF cycle may be found to be aneuploid, meaning that no embryos are genetically suitable for transfer.
 - Embryos diagnosed as chromosomally “normal” may not survive the freeze/thaw process and therefore may not be suitable for transfer from an embryology perspective.
 - Embryos diagnosed as chromosomally “normal” may survive the freeze/thaw process, but may not continue to develop normally and therefore may not be suitable for transfer from an embryology perspective.
- PGS with 24 chromosome screening is designed to test for aneuploidy involving the whole chromosome. While it can also detect some cases of partial aneuploidy (ie: a portion of a chromosome that is extra or missing), small extra or missing chromosome segments will usually not be detected. The potential significance of any small extra or missing chromosome segments will vary depending on the chromosome region involved.
- There are some rare chromosomal problems that will not or cannot be tested for using 24 chromosome screening.
- This test is only a screening test and therefore cannot provide an absolute guarantee of the chromosome status of the embryo. In some embryos, the biopsied cell/s may not be representative of the whole embryo. While every effort is made to ensure that the PGS test offered has the highest possible accuracy using the currently available technology, results are not 100% accurate. Therefore, **prenatal diagnosis is highly recommended in an ensuing pregnancy.**

What are the costs?

Information relating to the cost of PGS is available from your IVF clinic.