

Pre-implantation Genetic Screening (PGS)



Fertility Facts

By selecting an embryo with a normal number of chromosomes, PGS:

- 1. Reduces the chance of miscarriage**, because many miscarriages are due to the loss of aneuploid embryos.
- 2. Provides a higher chance of a healthy birth per embryo transfer**, because the embryo transferred has the correct number of chromosomes. The chance of a live birth after transfer of a single chromosomally normal embryo can be as high as 65-70%.

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Chromosomes

Human cells contain 46 chromosomes – a set of 23 chromosomes inherited from the mother's egg and a set of 23 chromosomes from the father's sperm. The chromosomes are numbered 1 to 22 in decreasing order of size when looked at under a microscope, plus X and Y for the two sex chromosomes.

Sperm and egg production both involve the reproductive cells halving their number of chromosomes to get back to 23 chromosomes each for the next generation of eggs and sperm. Sometimes this process malfunctions, especially during egg maturation and in women aged 35 and older. This leads to eggs, sperm and embryos having the wrong number of chromosomes, which is called 'aneuploidy'.

Aneuploidy is usually incompatible with embryo development, so that aneuploid embryos either fail to implant in the uterus or they stop developing after implantation leading to a miscarriage. Some types of aneuploidy are compatible with fetal development, the most well known is Down Syndrome where a child has 3 copies of chromosome 21.

Embryo development and chromosome number

Unfortunately, many embryos with the wrong number of chromosomes have a normal appearance when assessed by the embryologist.

PGS offers a powerful method of selecting which embryos have a better chance of progressing to a baby

by identifying those embryos with the wrong number of chromosomes. About 30-40% of all blastocysts in younger women are aneuploid, and this rises to around 80-90% by the age of 43-45.

How PGS works

PGS starts with an IVF cycle. Each egg is fertilized using a single sperm using intra cytoplasmic sperm injection (ICSI) so that there are no extra sperm attached to the Zona Pellucida (the soft outer shell of the embryo) that could unknowingly be included in the biopsy and contribute DNA to the PGS sample. A small hole is made in the Zona Pellucida with a laser on day 3 of embryo development.

If the embryo progresses to become a blastocyst on day 5 or 6, then some of its trophectoderm layer will start to protrude through the hole in the Zona Pellucida. Using a fine glass pipette and laser, a small amount of this layer is removed. This procedure is called embryo biopsy. >>

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PGS, continued...



The trophoctoderm layer will later form the placenta; removal of a few cells at this stage does not affect the embryo or the chance of pregnancy.

The cells are placed in a labeled tube, and the tubes sent to the genetics laboratory for testing. Meanwhile, the biopsied embryos are frozen.

At the genetics laboratory, DNA from each embryo is amplified about one million times, and placed on a microarray. The microarray has thousands of segments of DNA from the 22 chromosomes plus the X and Y chromosomes.

The DNA from the embryo sample and reference DNA compete to bind to the DNA on the microarray. The embryo DNA and the reference DNA are each labeled with different coloured fluorescent dyes, so the ratio of the two colours can be used to determine the number of each chromosome in the embryo sample.

Giving the results and planning embryo transfer

Samples are stored until there are enough to fill the spaces on the microarray – this keeps down the cost of PGS. Results are usually available within 6 weeks. We will tell you the results as soon as they are known so you can plan an embryo transfer cycle if you have normal embryos.

The benefits of PGS

By selecting an embryo with a normal number of chromosomes, PGS:

1. **Reduces the chance of miscarriage**, because many miscarriages are due to the loss of aneuploid embryos
2. **Provides a higher chance of a healthy birth per embryo transfer**, because the embryo transferred has the correct number of chromosomes. The chance of a live birth after transfer of a single chromosomally normal embryo can be as high as 65-70%.

Important to know

PGS does not increase the overall chance of a baby from an IVF cycle – it improves embryo selection for transfer by identifying abnormal embryos.

- In some IVF cycles there will not be any embryos suitable for testing because none will have developed to the blastocyst stage. Only about 40-50% of all fertilized eggs develop into blastocysts.
- Embryos need to be frozen after biopsy while testing takes place, and you will have to wait about 6 weeks to find out whether you have any normal embryos.
- Some blastocysts may not survive the freezing and thawing process between embryo biopsy and planned embryo transfer.
- Some people will have all their embryos diagnosed as aneuploid and therefore none of the embryos will be suitable for transfer.

Accuracy and reliability of PGS

The chance of making the correct diagnosis about whether an embryo has the correct number of chromosomes is 96% or higher. Most errors arise when an embryo is mosaic, which means some of its cells have the correct number of chromosomes and some have an abnormal number therefore the result may not be representative.

Sometimes there is no result for an embryo or the result is unclear. This can happen for a variety of reasons.

- **Inconclusive result** This occurs when the test results are ambiguous and the laboratory cannot decide whether an embryo is aneuploid or not.
- **Failure to amplify DNA** This occurs when the DNA in the cells taken by biopsy is not amplified enough to be loaded on the microarray for testing. In some cases, the embryo can be thawed, biopsied a second time, and refrozen for a second attempt. This can be done at no charge.
- **Array-based signal failure** This occurs when the DNA from the sample does not combine with the DNA on the microarray properly, so the test cannot be completed.

The cost of PGS

The fees for using PGS are covered in our fee sheet. The PGS fee covers the embryo biopsy and testing of all suitable embryos from the IVF cycle. To do PGS, you need to use ICSI as part of the IVF cycle, and you will need to have frozen embryo transfers to use suitable embryos. The costs of these are separate to the cost of the PGS testing.