

Yorkshire Regional Genetics Service

Pre-implantation genetic testing (PGT)

Information for patients



Yorkshire Regional
Genetics Service

Pre-implantation genetic testing (PGT) is a technique that is designed to help couples who are at risk of having a child with a serious genetic condition.

PGT can also, sometimes help couples who have had recurrent miscarriages because of a genetic condition. This leaflet will provide you with some general information about PGT, in order to help you reach a decision about whether or not you would like to pursue this option.

What is PGT?

PGT involves using IVF (in vitro fertilisation) to create embryos in the laboratory from the eggs and sperm of a couple. Each embryo is then tested for the particular genetic disorder they are at risk of (this is called embryo biopsy). One unaffected embryo is then transferred into the womb, in the hope that a pregnancy will occur.

Who can have PGT?

PGT is currently only offered in a few centres in the UK. One of the largest and most active PGT centres is based at Guy's Hospital.

The Yorkshire Regional Genetics Service and the Leeds Centre for Reproductive Medicine work closely with Guy's hospital to provide PGT to couples in the North of England. Couples still need to travel to London for some of their treatment.

To be able to offer PGT for a particular genetic condition, it must first have been licensed by the Human Fertilisation and Embryology Authority (HFEA).

The HFEA website lists the conditions that have been licensed at www.hfea.gov.uk/pgt-m-conditions

If the condition you are looking for is not on the list, we can still apply for a license for you as part of the PGT work-up. Applying for a license can take several months.

Please note that PGT-M refers to PGT for monogenic disorders (ie conditions caused by a change in a single gene). For those with at risk of having a child with an unbalanced translocation, individual licenses are usually not required as they are covered by a specific license for PGT-SR (structural rearrangements).

What does PGT involve?

Even though most couples who are going to have PGT are able to become pregnant through natural conception themselves, they will need to undergo IVF to produce embryos for testing.

First, the ovaries of the female partner are stimulated to hopefully produce several eggs. This is achieved by using a combination of fertility drugs. This IVF treatment will be discussed in more detail in the initial appointment with the fertility team.

Once the eggs are mature, they are collected from the ovaries under ultrasound guidance. A fine needle is passed through the vaginal wall to collect the eggs one by one. The procedure is carried out under sedation and usually takes around 20 minutes.

A semen sample is produced by the male partner on the same day.

The sperm are then used to fertilise the eggs in the laboratory. A technique called intra-cytoplasmic sperm injection (ICSI) is used.

Eggs which are successfully fertilised begin to grow and divide. They are now called embryos.

A few days later, once the embryos have grown, a small sample (containing cells) is taken from each embryo and DNA is extracted. Usually, the embryos are then rapidly frozen while the genetic testing takes place.

This DNA is tested for the particular genetic condition. One unaffected embryo is then placed into the womb using a fine tube or catheter. Several days later, a pregnancy test is carried out to see whether the PGT treatment has been successful.

What are the chances of success with PGT?

PGT involves using IVF, therefore the success rate is relatively low compared to the chances of conceiving naturally. Of all patients who start PGT treatment, approximately one in three couples will have a baby. If the couple have embryos suitable for transfer, then approximately one in two embryo transfers will be successful.

How much does PGT cost?

The cost of a cycle of PGT can be around £12,000; however, NHS funding is available to couples for up to three cycles if they meet a set of criteria. To meet these criteria, both partners need to be non-smokers. The woman must be under the age of 40 and have a BMI (body mass index) of over 19 and under 30. The couple must also not have any unaffected children together. There are some other, more complex, criteria that can be discussed with you.

What else is there to consider?

- Before PGT can be offered to a couple, preliminary laboratory work must be undertaken to ensure that an accurate genetic test can be carried out on any embryos produced. This process requires DNA samples from the couple and usually other family members; the exact requirements will vary with each couple. If an accurate test cannot be designed, PGT cannot be offered.
- There are risks associated with having PGT treatment e.g. hyperstimulation of the ovaries where the ovaries become very large and fluid may accumulate which may also cause abdominal swelling. This can lead to admission to hospital.
- Some women have a poor response to the fertility drugs and therefore, there are no eggs for collection. There is the possibility that no embryos will grow or there may be no unaffected embryos.
- There is a small risk of an error occurring in the testing. This will be discussed with you in greater depth if you pursue PGT.
- Previously, babies who have been born using PGT were followed-up after birth to determine if there were any greater or additional risks. There has been no evidence that this is the case, therefore babies are no longer monitored when they have been conceived via PGT.
- PGT can be a very emotionally and financially demanding process to go through.
- PGT can be a lengthy process, taking at least a year for most couples.

How do I get referred to the PGT clinic?

To be referred to the PGT clinic, please ask your geneticist or genetic counsellor to refer you for an initial discussion appointment. This appointment will give you the opportunity to ask questions and find out more about PGT before reaching your decision. This appointment is free of charge.

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Seen in clinic by

For more information:

For more information about PGT please visit:

www.hfea.gov.uk



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© The Leeds Teaching Hospitals NHS Trust • 3rd edition (Ver 1)
Developed by: Department of Clinical Genetics (Chapel Allerton Hospital)
With thanks to The Department of Clinical Genetics at Guy's & St Thomas' Hospital
Produced by: Medical Illustration Services • MID code: 20221207_009/EP

LN002330
Publication date
01/2023
Review date
01/2026