Preimplantation Genetic Testing (PGT)

Information for patients

Introduction

Preimplantation genetic testing (PGT) is a technique designed to help couples at risk of having a child with a single gene disorder, such as Cystic Fibrosis or Huntington's disease, or for those at risk of transmitting a chromosomal disorder.

PGT for single gene disorders is carried out in Edinburgh and PGT for chromosomal disorders is carried out in Glasgow.

This leaflet provides you with some general information about PGT, to help you decide whether or not you would like to pursue this option.

What is preimplantation genetic testing (PGT)?

PGT involves using *in vitro* fertilisation (IVF) to create embryos in the laboratory from the eggs and sperm of that couple. All the embryos that survive to the 'blastocyst' stage (day 5-6) are then tested for the particular genetic disorder. All embryos are frozen. A few months' later, one 'low risk' embryo is thawed and then transferred into the womb, in the hope that a pregnancy will occur.

Who can have PGT?

In Edinburgh, we offer PGT for a wide variety of conditions including: Cystic Fibrosis, Huntington's disease, BRCA, Duchenne muscular dystrophy, Myotonic dystrophy, and Fragile X syndrome. Other conditions can be considered.

What does PGT involve?

Even though most couples considering PGT are able to become pregnant themselves, they will need to undergo IVF to produce embryos for testing.

The flowchart on the next page describes the process of PGT treatment.

First, the ovaries of the female partner are stimulated to produce several eggs. This is achieved by using a combination of fertility drugs, which are usually taken by daily injection.

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 deep sedation and usually takes around 20 minutes.

The sperm are then used to fertilise the eggs in the laboratory using a technique called intra-cytoplasmic sperm injection (ICSI) where one sperm is injected into each egg.

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

5-6 days later, once the embryo has grown to a blastocyst, a few cells are very carefully removed (this is called the 'trophectoderm biopsy'). The sample is then tested for the particular genetic condition. All the embryos are frozen.

A few months' later, one 'low risk' embryo is thawed and replaced into the womb using a fine tube or 'catheter'.

10-12 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?

As PGT involves using IVF, the success rate is relatively low compared to the chances of conceiving naturally. Our current success rate is 40% per cycle.

How much does PGT cost?

In Scotland, couples who fulfil the PGT eligibility criteria and have no unaffected children, are eligible for **up to three** NHS-funded PGD cycles.

What else is there to consider?

Having PGT is a relatively complicated and lengthy procedure. It takes approximately 18 months between your first appointment and the start of treatment, and you would need to attend several appointments.

There are risks associated with having IVF treatment, for example 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.

A twin pregnancy can result, even when only one embryo is replaced. Multiple pregnancies carry a higher risk of complication for the babies and for the mother.

Some women have a poor response to the fertility drugs and therefore there are no eggs for collection. There is a chance that no embryos will grow or there may be no ‘low risk’ embryos.

There is a small risk of an error or a ‘misdiagnosis’. This will be discussed with you in greater depth if you pursue PGT. If a pregnancy is achieved following PGT, we offer all couples the option of prenatal testing to check that the pregnancy is not affected with the genetic condition.

Whilst we believe that PGT is relatively safe, all babies born as a result of PGT are followed up at birth.

PGT can be a very emotionally demanding process to go through.

How do I get referred to the PGT clinic?

To be referred to the PGT clinic, please ask your local 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.

Contact information

Your local genetics service:

South East of Scotland Clinical Genetic Services
Molecular Medicine Centre
Western General Hospital
Crewe Road South
Edinburgh
EH4 2XU

Telephone: 0131 537 1116