

# PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

# Information for patients

#### INTRODUCTION

Preimplantation genetic diagnosis (PGD) 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.

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

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

#### WHAT IS PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

PGD 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 PGD?**

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

Leaflet no: 028, Version 5. Due for review: July 2021 Page 1 of 4

## WHAT DOES PGD INVOLVE?

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

The flowchart below describes the process of PGD 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 PGD treatment has been successful.

Leaflet no: 028, Version 5. Due for review: July 2021 Page 2 of 4

#### WHAT ARE THE CHANCES OF SUCCESS WITH PGD?

As PGD 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 PGD COST?**

In Scotland, couples who fulfill the PGD eligibility criteria and have no unaffected children, are eligible for *up to three* NHS-funded PGD cycles. The current cost of a private PGD cycle at the Royal Infirmary of Edinburgh for UK residents is £7,875 including fertility drugs.

#### WHAT ELSE IS THERE TO CONSIDER?

Having PGD is a relatively complicated and lengthy procedure. It takes approximately 6-8 months between your first appointment and the start of treatment, and you would need to attend approximately 8-10 appointments in total.

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 PGD. If a pregnancy is achieved following PGD, 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 PGD is relatively safe, all babies born as a result of PGD are followed up at birth.

Leaflet no: 028, Version 5. Due for review: July 2021 Page 3 of 4

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

## **HOW DO I GET REFERRED TO THE PGD CLINIC?**

To be referred to the PGD 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 PGD before reaching your decision.

## **CONTACTS**

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Leaflet no: 028, Version 5. Due for review: July 2021 Page 4 of 4