Edinburgh Fertility Centre

Preimplantation Genetic Testing (PGT)
Information Sheet

What is Preimplantation Genetic Testing (PGT)?
PGT is a specialised technique that is designed to help couples who are at risk of having a child with a specific genetic condition or chromosomal disorder.

PGT involves the egg provider first taking ovarian stimulation drugs by daily injection. Next, an IVF technique called ICSI is used to create embryos in the laboratory from the eggs and sperm of the couple receiving treatment. Embryos that develop appropriately to day 5/6 (blastocyst stage) are then biopsied, frozen and tested for the specific genetic disorder in that family. One to three months' later, one 'low risk' embryo is transferred into the uterus, with the hope that it will implant and a pregnancy will result. Hence PGT makes it possible for couples to greatly reduce the risk of transmitting a serious genetic disorder to their children.

PGT gives such couples an alternative option to prenatal testing, which involves testing cells from the foetus during a natural pregnancy. Prenatal testing can be performed by chorion villus sampling (CVS) at 12 weeks' gestation and carries a 2% miscarriage risk, or by amniocentesis at 16 weeks' gestation, and carries a 1% miscarriage risk. Couples having prenatal testing might then be faced with the difficult decision of terminating an affected pregnancy.

Who is eligible for PGT in Scotland?
The couple must have:

1. A known genetic condition in the family which conveys a 'significant risk of a serious genetic condition'.
2. No living, unaffected child, or untested child (for an adult-onset disorder) as a couple or, if one partner has no living biological child.
3. Egg provider having an adequate ovarian reserve.
4. Egg provider < 39 years.
5. Egg provider's body mass index < 30.
6. Both partners should be non-smokers for at least 3 months, living at same address for at least 2 years and both must be eligible for NHS treatment.

Individuals who have an unaffected child from PGT are not entitled to any further NHS-funded cycles.

The PGT Team
The PGT team is an expert, multidisciplinary team involving several specialities including Clinical Genetics, Molecular Genetics, IVF and Embryology.

Your first appointment will take place in Clinical Genetics department at the Western General Hospital in Edinburgh. After this, all of your appointments will take place in the Edinburgh Fertility Centre (EFC) which is located in the Royal Infirmary of Edinburgh, Little France Crescent, Edinburgh.

Over the course of your treatment, you may meet:

- Professor Mary Porteous, Consultant Clinical Geneticist
- Ms Sally Morton, PGT Genetic Counsellor
- Dr Brian Brady, Consultant in IVF
- Dr Maya Chetty, Consultant in IVF
- Ms Laura Wood, IVF Deputy Charge Nurse
- Dr Daniel Collins, Consultant Embryologist
- And some of the other IVF doctors and nurses.

Undergoing PGT is a complex process involving attending several appointments, blood tests, scans, completing many consent forms and other tests over several months and it can be both stressful and emotional. Professional support and counselling is available throughout the process and you are welcome to meet with our counsellors and discuss issues, feelings and anxieties before, during and following your treatment. For the latest waiting times please visit our website (www.nhslothian.scot.nhs.uk/edinburghivf).

What does PGT involve?

PGT involves assisted reproduction techniques (ART) to generate embryos *in vitro*, using the insemination method of intra-cytoplasmic sperm injection (ISCI). Suitably developed blastocyst stage embryos then have part of their trophectoderm (TE) biopsied on day 5 or day 6 and the cells are sent for genetic analysis. The biopsied embryos are frozen immediately. If frozen embryos are identified by the genetic test as suitable for transfer, a frozen embryo transfer (FET) can be arranged.

What are the risks of PGT?

There are a number of risks involved with PGT including the risk of ovarian hyperstimulation (OHSS) or alternatively the risk of poor response to the fertility drugs. There may not be suitable embryos developed to be able to biopsy. There is a small risk (under 5%) that the biopsy may not initially be successful which would require a further biopsy and risk that the embryo may become unusable for treatment. After genetic testing is complete, there is a risk that none of the successfully biopsied embryos are suitable for use in treatment. There is also a small risk of a multiple pregnancy (even when only one embryo has been transferred) and the small risk of a 'misdiagnosis'. All of these risks would be discussed in more detail at your appointments. The testing involved in PGT may reveal additional genetic information about embryos and the clinical effect of these findings on subsequent children born through the procedure may not be known.

What are the success rates of PGT?

The Edinburgh Fertility Centre has been providing patients with PGT for over 15 years. In 2023 our success rate was around 50% of FETs demonstrating positive foetal heartbeats for PGT patients who have had an embryo to transfer (54.5%).

Are there alternatives to PGT?

The alternative options are:

- Natural conception with no prenatal testing.
- Natural conception with prenatal testing by chorionic villus sampling (CVS) or amniocentesis.
- Sperm or egg donation.
- Adoption.
- No (further) children.

How do we get referred for PGT?

Eligible couples can be referred by letter by their local Clinical Genetics Service to Professor Mary Porteous, Consultant Clinical Geneticist, South East of Scotland Genetics Service, Western General Hospital, Crewe Road, Edinburgh, EH4 2XU.

Should you wish further genetic counselling, this can be arranged with our PGT co-ordinator, Sally Morton, or through the Genetics centre at the Western General, Edinburgh

Further information, including testimonies of people living with the condition, by contacting the patient support group 'Genetic Alliance UK' at http://www.geneticalliance.org.uk/