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 female partner 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 survive until Day 5 of development are then tested for the specific genetic disorder in that family. Then, all embryos of sufficient quality will be frozen. 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.

PGT gives such couples an alternative option to prenatal diagnosis (PND), which involves testing cells from the foetus during a natural pregnancy. PND can be performed by chorion villus sampling (CVS) at 12 weeks' gestation and carries a 0.5% miscarriage risk, or by amniocentesis at 16 weeks' gestation, and carries a 0.5% miscarriage risk. Couples having PND 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. Female hormone level (anti mullerian hormone) of 7.5 pmol/L or greater (or an adequate ovarian scan).
4. Female age < 39 years.
5. Female 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 Joo Thong, Consultant in IVF
- Dr Maya Chetty, Consultant in IVF
- Ms Alison Whitecross and Ms Laura Wood, IVF Staff Nurses
- 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. For the latest waiting times please visit our website (www.nhslothian.scot.nhs.uk/edinburghivf).

What are the risks of PGT?

There are risks involved with PGT including the risk of ovarian hyperstimulation (OHSS) or alternatively the risk of poor response to the fertility drugs. As this test requires the biopsy of embryos, there is a small risk that the biopsy may not be successful which may require a further biopsy or the embryo may become unusable for treatment. After genetic testing is complete, there is a risk that none of the 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.

What are the success rates of PGT?

The Edinburgh Fertility Centre has been providing patients with PGT for over 15 years. With the advent of trophectoderm (TE) biopsy, reporting success rates has become more complex. A TE biopsy means you will have a frozen embryo transfer (FET) following the results of the genetics test. For 2019 we had a clinical pregnancy rate (a foetal heart detected) of 53.8% for couples who had an embryo suitable for transfer with a subsequent FET.

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.