Lothian NHS Board

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Date: 28/05/2026
Our Ref: 11432
Enquiries to loth.freedomofinfomation@nhs.scot

Dear Ms Allen

FREEDOM OF INFORMATION – PREIMPLANTATION GENETIC TESTING (PGT).

I write in response to your request for information in relation to preimplantation genetic testing:

Question 1. What information resources (e.g. leaflets, websites, videos) are provided to patients about preimplantation genetic testing (PGT)?

Answer: NHS Lothian provides the following information resources for patients regarding Preimplantation Genetic Testing (PGT):

'Preimplantation Genetic Testing - information for patients'. Leaflet produced by SE Scotland Clinical Genetics Service (attached).

'Preimplantation Genetic Testing - information Sheet'. Sheet produced by Edinburgh Fertility Centre (attached).

In addition, Edinburgh Fertility Centre provide their own videos/leaflets about the IVF part of the process.

Question 2. Are PGT information resources available in accessible formats (e.g. easy read, translated materials)?

Answer: The leaflets are written for patients and are regularly reviewed and updated as necessary but are not specifically easy read or translated.

Question 3. What resources do you have for providing emotional or psychological support to couples during the PGT process?

Answer: Couples would have access to their own Clinical Geneticist/Genetic Counsellor as well as the PGT Genetic Counsellor. In addition, there is a Clinical Psychologist in Clinical Genetics, where required, as well as Fertility Counsellors who are part of the Fertility Clinic team, and the IVF team.

Question 4. What support is offered to patients or couples navigating PGT decision making and/or following PGT outcomes?

Answer: Couples would have access to their own Clinical Geneticist/Genetic Counsellor as well as the PGT Genetic Counsellor. In addition, there is a Clinical Psychologist in Clinical Genetics, where required, as well as Fertility Counsellors who are part of the Fertility Clinic team, and the IVF team.

Question 5. Are patients signposted to any external organisations or charities relating to PGT, if so who and what resources are these?

Answer: HFEA website.

Question 6. Are there any internal guidelines or standard operating procedures (SOPs) for staff on how to discuss PGT with patients?

Answer: We use a 'PGT Clinic Checklist' (attached).

Question 7. What educational resources or training do you have to support staff on discussing PGT with patients?

Answer: Genetics trainees can observe PGT clinics as part of their training, with prior consent from the couples.

I hope the information provided helps with your request.

If you are unhappy with our response to your request, you do have the right to request us to review it. Your request should be made within 40 working days of receipt of this letter, and we will reply within 20 working days of receipt. If our decision is unchanged following a review and you remain dissatisfied with this, you then have the right to make a formal complaint to the Scottish Information Commissioner within 6 months of receipt of our review response. You can do this by using the Scottish Information Commissioner's Office online appeals service at https://www.foi.scot/appeal. If you remain dissatisfied with the Commissioner's response you then have the option to appeal to the Court of Session on a point of law.

If you require a review of our decision to be carried out, please write to the reviewer at the address at the top of this letter. The review will be undertaken by a Reviewer who was not involved in the original decision-making process.

FOI responses (subject to redaction of personal information) may appear on NHS Lothian's Freedom of Information website at: https://org.nhslothian.scot/FOI

Yours sincerely

ALISON MACDONALD
Executive Director of Nursing Midwifery and AHPs
Cc: Chief Executive

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/

PGT Checklist 1 to be completed at Genetics appt PED =

Label (female) Female age = Label (male)

AMH (if known) =

Date taken =

Age when tested=

Date of clinic appt: Seen by: MPOR / SMOR /

Condition in family: HFEA Licence available? Y / N / Being considered
OMIM:
Date checked:

Variant(s): Report obtained? Y / N

Does the variant need reviewed? N / Y, _____

Agree to treat? Y / N / Not yet _____ Local Genetics clinician _____

Preparatory notes

☐ If X-linked condition, discuss their decision if only female carriers are available for transfer?

Length of relationship (years) _____

☐ Pedigree and risk discussed ☐ Other reproductive options

☐ Any fertility concerns? Not known / N / Y, details = _____

☐ Overview of IVF - Timescale, risks, affect of smoking, BMI, affect on sibs

☐ Overview of PGD - Drug tx through to biopsy & testing (haplotyping, markers)
- Misdiag risk _____% per embryo replaced
- PND is offered

Will NEWBORN CONFIRMATION be offered? Yes / No / Other _____

- Trophectoderm biopsy used for all new couples fm Oct 2018 (Day 5/6)
-All tested embryos will be frozen
-Some couples may not get any blastocysts to test
- Limitations of analysis - only check for condn in family
- poor quality / no result embryos
- Attrition of embryos throughout process

☐ Molecular work-up on their family required before starting tx. Timescale _____

☐ All authorised conditions are automatically published on the HFEA website and are therefore in the public domain (patient names are not listed)

☐ One cycle offered then review situation (looking at their response / age / AMH)

☐ Up to 3 cycles offered on NHS

☐ Success rate 40%. Ovarian scan and AMH level only give an indication of success

☐ Long waiting list

☐ Current contraception _____

☐ Barrier contraception essential

☐ Foreign travel to hot country in last 6 months or planned in next year? N / Y

If Yes, give details _____

Samples required at this appt

☐ ☐ Work-up bloods from individual or couple

☐ AMH from female (if not performed yet)

☐ Obtain DNA from relative(s) Who? _____

_____

_____

_____

☐ Saliva or Blood packs/forms given out?

(Need DNA from both sets of parents for de novo cases).

☐ PGD consent form explained and given to couple to take home

Female BMI = W (kg) / H^2 (m) _____

Plan of action after appt

SMOR to refer couple to EFREC? _____

Request that SA, MMR and smear test done locally before next appt _____

Stress that medical, pre-pregnancy &/or anaesthetic assessment will be required before decision about PGD can be made, especially if one partner is affected with a genetic condition eg CF, Myotonic, VHL

_____

Other work to be completed? _____

Work up to be requested? Lab to get reports of family members when they request DNA samples

_____

Any other comments? _____

OK to cc GP and referrer?

Name of clinician

Health of couple including Folic Acid discussion:

Signature of clinician

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