

**Centre for
Preimplantation
Genetic
Diagnosis**

Genetic testing
Excellence
Support
Advice
PGD
Care

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The PGD Team Members



We are here to offer couples at risk of having children with a genetic disorder, the chance to have a healthy child.

PGD involves creating embryos for a couple and testing those embryos before they form a pregnancy in the womb.

Over 100 babies have been born following PGD in our centre and we look forward to meeting you to discuss the treatment we offer.

Winners of Hospital Doctor Awards 2006

Innovations Team of the Year

THE JUDGES'S COMMENT

'This first-class team of scientists and clinicians has received international acclaim for developing a technique that allows more couples whose children would be at risk of genetic diseases to opt for unaffected pregnancies.

Pre-implantation genetic haplotyping reduces test time, improves reliability and spares unaffected males in sex-linked disorders'

Welcome to the Centre for Preimplantation Genetic Diagnosis

Thank you for your interest in Guy's and St Thomas' Centre for Preimplantation Genetic Diagnosis. This booklet has been designed to provide you with information about preimplantation genetic diagnosis (PGD) and how it may help you to achieve a pregnancy, avoiding the genetic condition known in your family. You will also find a supplementary leaflet in the back of the booklet that has more information about PGD for your particular genetic condition.

We expect that you will already know much about the condition in your family and how it is inherited, so this booklet focuses on what PGD is, and the important issues relating to it. The amount of information that we can put in a booklet is limited, but there will be ample opportunity for further discussion during your consultations in our clinic.

We understand that many of the words used in this booklet may be new to you. There is a glossary at the back to explain words you might not know.

We will tell you if there are any changes to the information provided in this booklet. We welcome your comments on the booklet and any part of our service, so please feel free to speak to a member of the PGD team.

The PGD team

We are a team of medical and scientific specialists dedicated to giving a comprehensive and caring PGD service of the highest standard. We believe it is essential that couples who choose PGD have rapid access to expert advice, given in a sympathetic manner and which takes into account individual factors about each couple.

Most letters of referral to our Centre come from clinical geneticists. We believe it important that you have already been seen in your local Genetics Department before attending our clinic. This is to ensure that you have had an opportunity to discuss the genetic basis of your condition, that all necessary laboratory work has been completed and that you are aware of all the options available to you when you consider having a baby.

A good PGD service must have the back up of a successful Assisted Conception Unit (ACU). Our results for fertility patients compare favourably with the national success rates for in vitro fertilisation (IVF). Success rates are published by the Human Fertilisation and Embryology Authority (HFEA). The HFEA licenses us to practise a variety of assisted conception techniques. We continually aim to improve our success rates through research and regular review of our service. However, we also feel that it is important to be honest and realistic with our patients and we acknowledge that, sadly, we will not be able to achieve a pregnancy for many couples that undergo treatment with us. Whatever the outcome, we hope that the experience will be a positive one and we will try to give you as much support as possible both during and after your treatment.

The PGD team works within the Assisted Conception Unit (ACU) with the help of a large team that provides care for all patients needing to use these specialised services. A number of the ACU staff will usually be involved in your care, and we work together to give a comprehensive service.

Administration team

The PGD coordinator will probably be the first person you meet when you come to the Unit. They are responsible for keeping your records, answering initial queries and making sure a doctor or nurse responds to your questions when appropriate.

Genetic counsellors

You will meet a senior PGD genetic counsellor and sometimes an ACU nurse or doctor on your first visit. The clinical genetics service works closely with the ACU to ensure that you receive the most comprehensive information available.

Nurses

The PGD team works closely with the ACU nurses who plan your treatment schedule and teach you about any medications used in your treatment. The nurses perform ultrasound scans, blood tests and assist in egg collection and embryo replacement procedures.

Doctors

You will meet a PGD doctor on your second visit to us. Our team of doctors will oversee all aspects of your treatment in the Unit and perform ultrasound scans, egg collection and embryo replacement.

Embryologists

The embryologists are responsible for looking after your eggs, sperm and embryos whilst they are in the laboratory. After egg collection, they prepare the eggs and sperm for fertilisation and look after the embryos. They also perform intracytoplasmic sperm injection (ICSI) where it is needed and embryo biopsy.

Scientists

A team of scientists in the laboratory perform the important chromosome or genetic testing for PGD cases. You are likely to meet them during a treatment cycle before embryo transfer.

Counselling

We have specialist counsellors within the team and all couples can see them if they wish. We appreciate that trying to conceive through PGD can be stressful. Counselling provides support from a trained professional who understands what is involved in your treatment and can offer you the time to talk over your options or concerns. Most people find that having someone to discuss this with makes all the difference between feeling stressed and worried to feeling more able to cope.

To make an appointment with a counsellor, please telephone the administration team on 020 7188 2300. All counselling is confidential and free of charge.



What is PGD?

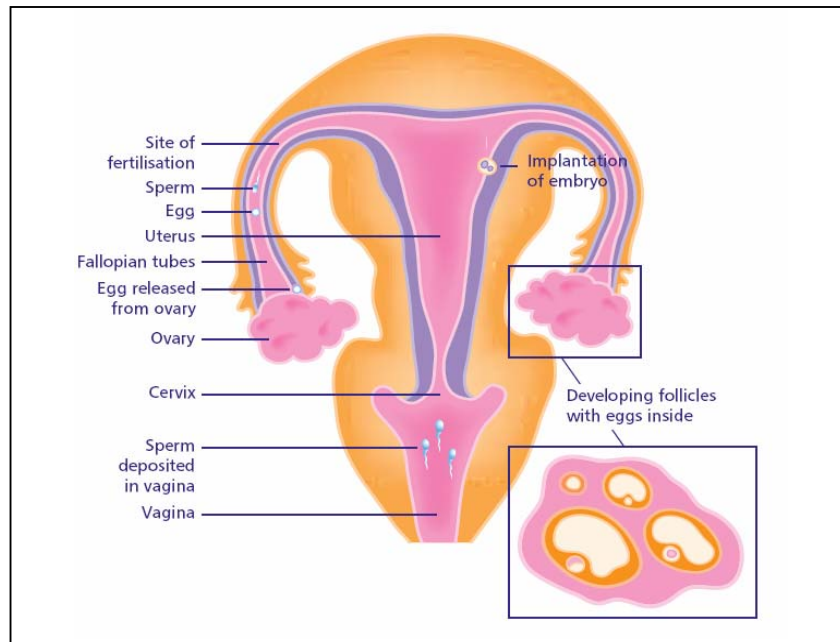
PGD is available to couples that are at risk of having a child with a specific genetic or chromosome disorder. It prevents the birth of an affected child by testing an embryo before it is implanted in the uterus (womb).

Briefly, it involves the use of Assisted Reproductive Technology (ART), which in other circumstances is offered to couples with fertility problems. The aim is to obtain and fertilise a number of eggs. Once fertilised, the embryos develop for three days and then one cell is removed from each embryo. The genetic material (DNA or chromosomes) within each cell is then tested for the altered gene or chromosome abnormality. Up to two unaffected embryos are then transferred to the uterus with the hope that they will implant and form a pregnancy. If successful, a pregnancy should be unaffected. PGD is a lengthy and complex process. The stages of PGD treatment are described later in this booklet.

Why is assisted conception needed for PGD?

Usually each month, an egg develops in one of the ovaries in a small sac of fluid called a follicle. The egg is released from the ovary and passes down one of the fallopian tubes, which are attached to the uterus. For a pregnancy to occur, sperm, which has been deposited in the vagina during intercourse, has to swim towards the egg in the fallopian tube. This is where fertilisation normally occurs.

The fertilised egg begins to divide as it travels through the fallopian tube towards the uterus. After about five days, the fertilised egg (embryo) hatches out of its outer coating (zona pellucida) and buries itself in the thick lining of the uterus where it begins to grow (the process of implantation). Rarely, an embryo implants in the fallopian tube. This is called an ectopic pregnancy.



We know that a number of your embryos are likely to be affected by the altered gene or chromosome abnormality. Therefore, we need to create a large number of embryos for the best chances of success in a PGD cycle. It is important that we start by obtaining a good number of eggs from a woman. This means that we artificially take control and alter a normal menstrual cycle.

Success rate of PGD

There are several factors that can affect the success rate. The most important is a woman's age. The chances of success decrease as she becomes older, especially over 39. Overall, the chance of a couple having a baby born following treatment is around 20% per cycle (one in five). Your chances increase if you progress successfully through each step of the treatment process. Your chances of a clinical pregnancy (pregnancy with a heartbeat seen on ultrasound scan) are:

- one in five, when you start a cycle
- one in three, if you have embryo(s) replaced.

Starting a treatment cycle is no guarantee that there will be suitable embryos to transfer. Sometimes we may be left with no embryos to transfer for one or more of several reasons:

- there may not be many eggs to collect;
- some eggs may not have fertilised;
- all embryos may have been affected;
- some may not have survived the biopsy procedure.

If you have a positive pregnancy test, there is still a risk that you can miscarry (30%) or have an ectopic pregnancy (2%). Understandably, this can be disappointing and upsetting. As many couples requesting PGD are normally fertile, this may be one of the most difficult aspects of the treatment.

What can affect the chances of success?

Weight (female partner)

If you are overweight:

- you might not respond as well to the medication used to stimulate your ovaries and as a result produce fewer eggs;
- it is more difficult to obtain a clear scan picture during egg collection;
- the medication we give you for pain relief and sedation during this procedure can slow your rate of breathing and this can cause complications;
- if you do become pregnant, the chance of miscarriage might be higher and there can be more complications in pregnancy, in particular with high blood pressure and sugar tolerance (gestational diabetes).

For all these reasons, we recommend that patients who are very overweight should lose weight before starting treatment. We will not be able to offer you treatment unless you are within the correct weight limit. The table below gives the maximum weight acceptable for your height. If you exceed the recommended weight, we can refer you to a dietitian or you can speak to your GP about local weight loss programmes. Group weight loss programmes have been shown to be more effective than individual dieting.

Height		Weight	
feet / inches	metres	stones – pounds	kilograms (kg)
4' 11"	1.50	10-7	68
5' 0"	1.52	11-0	70
5' 1"	1.55	11-3	72
5' 2"	1.57	11-6	74
5' 3"	1.60	12-1	77
5' 4"	1.62	12-4	79
5' 5"	1.65	12-8	82
5' 6"	1.67	13-2	84
5' 7"	1.70	13-5	86
5' 8"	1.72	13-10	89
5' 9"	1.75	14-4	92
5' 10"	1.77	14-8	94
6' 0"	1.82	15-6	99

For example, if you are 5' 6" and weigh more than 13 stone 2 pounds (84kg), we recommend you lose weight before starting treatment.

Smoking (both partners)

We recommend, if you smoke, to cut down or, preferably, stop. Smoking has been shown to reduce the chances of conceiving naturally and lowers the success rates of IVF. Cannabis seriously affects sperm quality and reduces the chance of fertilisation. If you want help to try and stop smoking, please speak to your GP. You might also want to call the NHS Smoking Helpline on 0800 169 0169.

Alcohol

Excessive intake of alcohol, especially 'binge' drinking, can decrease sperm production and the number of moving sperm. Sperm take 72 days to form so the effects of damage take a long time to reverse. It is recommended that you drink less than two units of alcohol per day. There is no clear evidence that alcohol does or does not affect fertility in women. However as women are advised not to drink alcohol during pregnancy preparation for PGD may be a good time to review and limit alcohol intake.

Folic Acid (female partner)

We recommend you take 400 micrograms of folic acid each day. Folic acid is a vitamin, which has been shown to greatly reduce the risk of abnormalities of the brain and spinal cord (anencephaly and spina bifida) in an unborn baby. It has no side-effects and is safe to take during pregnancy.

Folic acid should be started by the beginning of a treatment cycle and continued to the 12th week of pregnancy. It can be bought in pharmacies and supermarkets.

Preparation for PGD treatment

Preparation for a PGD cycle consists of several stages and is the most time consuming part of the process. The preparation time will vary, but on average it will take between 6-8 months after your first visit before you will start treatment.

Referral

After your geneticist or genetic counsellor has referred you to the PGD centre and we have checked that we have a PGD test for use in your case, we will send you an information pack. This contains forms for both of you to complete. We ask you to answer questions about your background, which are relevant to you having PGD. We also ask for your written permission to contact your GP(s) and any other doctors involved in your care.

Criteria for starting PGD treatment

A couple can be considered for a PGD cycle if:

- the female partner is under 39 years of age (usually) and the male partner under 60 years of age;
- they complete and return the questionnaires;
- they are living together and in a stable relationship;
- the female partner's hormone levels are within a range that suggests her ovaries will respond to treatment;
- an accurate test is available and a licence has been obtained from the HFEA (see page 30);
- the PGD team confirms the couple is suitable for treatment;
- there are no concerns about the welfare of any child conceived using our treatment;
- funding is available – either from the NHS or the couple, if they choose to pay for their own treatment.

Funding

Each referral will be considered individually. Your own Strategic Health Authority or Primary Care Trust might have separate criteria, which have to be taken into account before they will consider funding your treatment. They may not, for example, give funding if you already have unaffected children or the female partner is over a certain age and the chances of success are very low. After your first meeting with the team to discuss PGD, before we do anything else, we will make an application for funding.

If you are not eligible for funding you may choose to self-fund your own treatment. For more information about our current charges for funding your own treatment please see the leaflet at the end of this booklet.

The patient information evening

At some stage before starting treatment you may find it helpful to attend a patient information evening. This gives a comprehensive explanation about all aspects of a treatment cycle. It is mainly aimed at couples having treatment for infertility, but there are many areas that overlap with PGD. We appreciate that it can be difficult to attend if you live outside London. If you would like to attend, please call 020 7188 2300.

Consultations

Your first appointment with the PGD team

This appointment usually lasts about one hour. There is a considerable amount of information to discuss at this appointment, so please arrive on time. It is not always possible to see couples that arrive late. During this appointment you are likely to meet the PGD Genetic Counsellor and possibly one of the PGD team doctors or nurses. This appointment provides a good opportunity to answer your questions and for you to tell us about your story and how you feel that PGD can help you. The discussions will include a general introduction to PGD treatment, an explanation of how the treatment works including how we plan to test the embryos, the likelihood of success and the limitations of the procedure. Many of the issues will be covered in this booklet, but this will be your personal appointment and will concentrate on PGD issues that specifically relate to you.

A general health history may be collected from both of you. Women will be asked specific questions about their menstrual cycle, previous pregnancies or pregnancy losses and sexual health. Men will be asked about any previous pregnancies they have fathered and about their sexual health.

It is unusual for any clinical procedures, such as scans or examinations, to be carried out during this appointment, but on some occasions blood tests may be required. These are usually only done at this stage if we have any medical concerns about how effective or safe the treatment will be.

At the end of the first visit, we are usually able to give you some idea of whether we can offer you PGD or whether there are any potential problems that may reduce the chances of PGD success from the outset. Sometimes it is necessary to arrange additional investigations before we can move forward with treatment.

After the appointment you will be sent a letter summarising our discussions. If you decide that you would like to continue, then we ask you to contact us to let us know that you wish to proceed with the next stage of preparation for PGD.

What happens next?

1. Firstly we will make an application for funding.

2. We will then arrange for blood samples to be sent to our laboratory to prepare the PGD test. These samples can usually be collected by the GP or nurse at your local surgery. The laboratories will be working on blood samples from you to ensure the test is possible and accurate (see supplementary leaflet).

You will not be offered a follow on appointment until the laboratory work up is complete and we know whether funding has been agreed.

Subsequent appointments

Once the above is complete you will need to return to our Centre for another appointment before treatment may begin. This appointment will include:

- taking a detailed health history;
- blood tests;
- ultrasound scans;
- consent signing;
- semen analysis.

During this appointment the doctor will take a full medical history from both of you and arrange any preliminary tests that are needed. It is useful to bring with you any information about previous fertility treatment you may have had, if this applies to you, especially IVF or ICSI attempts. The doctor will make sure you are suitable to start PGD treatment and will be happy to answer any questions that you might have.

At this visit, the female partner will have a trans-vaginal (internal) ultrasound scan to check for any problems that might reduce the chances of successful treatment. Problems include cysts in the ovaries, some types of damaged fallopian tubes, or fibroids in the uterus. If a problem is found, further investigations and surgical treatment might be needed before you start treatment. You do not need a full bladder for this scan. A small probe is placed inside the vagina to look at the uterus and both ovaries. The internal scan gives a clearer view than a scan performed through the abdomen (tummy). Most women find this procedure is not as uncomfortable as a smear test.

Obtaining consent

Before you receive any treatment, your doctor will explain what he or she is recommending; the risks involved, and will answer any questions you might have. We want you to have all the information you need to decide whether you want treatment. No treatment is carried out without your consent.

You will be asked to sign consent forms confirming your agreement to treatment which will be sent out to you ahead of time so that you are familiar with the content. You may find the leaflet, **Helping you decide – our consent policy**, useful. Please ask for a copy.

Consent for the storage and use of gametes (eggs, sperm) and embryos

One of the consent forms asks you to confirm what you want to happen to your gametes (eggs or sperm). We need your permission to fertilise your gametes and to use your embryos for your treatment. Before you start an IVF or ICSI cycle, you must decide what will happen to:

- any eggs or sperm which do not produce an embryo;
- any embryos that are not chosen for replacement.

If the eggs do not fertilise within 24 hours, they cannot become embryos. They will be left to perish unless you choose to donate them for research. Similarly, any sperm left over after all the eggs have had a chance to be fertilised, will either be left to perish or can be used for research. Please see page 28 for more information about our research.

The best unaffected embryos are replaced and the remainder are checked for suitability for freezing, if you wish. Please see page 22 for more information

about freezing your embryos. Any embryos not replaced or frozen will be allowed to perish, unless you choose to donate them for research. If you would like us to freeze your sperm or embryos, we have a legal obligation to ask what you would like to happen in the unfortunate event either of you dies or is no longer able to give consent to the use of your sperm or embryos. You will need to decide if you want your partner to:

- continue to be able to use the sperm or embryos;
- have the sperm or embryos thawed out and left to perish;
- donate the sperm or embryos to research.

We understand these are difficult decisions. Please feel free to take time to think about these options and to ask us any questions. You may withdraw or change your consent up until the time the gametes or embryos are used.

Preparatory Investigations

Blood tests

In addition to the blood tests mentioned above needed for genetic testing, a blood test will be taken between day two to day four (day one being first day of a period) of the female partner's menstrual cycle to measure her hormone levels. The result will help us to decide the dose of medication to use for the treatment. It is a national recommendation that all men and women having IVF treatment have a blood test for Hepatitis B and C and HIV. We have decided to extend this recommendation to all couples having any form of treatment in the ACU. This is important because a positive result can impact on:

- your health;
- the health and well being of any child conceived from treatment;
- the likelihood of the virus being passed on.

It is also necessary to do these tests if we are to freeze any extra embryos that are created during treatment.

We may offer treatment to a couple if one partner has a positive result but would recommend you are seen by a specialist doctor for further advice before starting PGD treatment.

Semen analysis

During your second appointment it is usual to examine at least one semen sample in our laboratory, even if you have no fertility problems or have had one done elsewhere. This is because different laboratories use different criteria for assessing sperm. We use World Health Organisation standards and will take responsibility for any treatment recommended, based on our assessment.

Producing a semen sample:

- You should not have intercourse or masturbate (by hand) for two to three days before producing the sample as both of these can affect the quality of the semen.
- You can produce the sample in a room in the Unit, which is specifically for this purpose. Alternatively, you can produce the sample at home if the travelling time to the Unit is less than two hours.

- You produce the semen sample by masturbation.
 - If masturbation is likely to be a problem, we can give you a special condom to produce the sample during intercourse. This condom does not contain a lubricant that kills sperm. The sample must still reach us for analysis within two hours of production.
 - Do not use soap or lubricants immediately before producing the sample, as they are toxic to the sperm.
-
- Collect the whole sample in the container provided.
 - It is extremely important that you label the container with your name, your partner's name, the date, the time the sample was produced and the date of your last ejaculation. All of this information is necessary for safe and accurate identification, processing and interpretation of the result.
 - Do not allow the sample to become too cold if you produce it at home. Place the container in your pocket or your partner's handbag.

If you think you may have difficulty producing a sample, please let us know. We can arrange for your semen to be collected and frozen for future treatment.

Occasionally the male partner of a couple requesting PGD does not have any sperm at all (azoospermia).

A man might have azoospermia because:

- of a blockage in the connecting passages from the testicles – called **obstructive azoospermia**; or
- very few sperm are being produced in the testicles – called **non-obstructive azoospermia**.

As only one sperm is needed per egg for ICSI, we can use a surgical procedure-**Percutaneous Epididymal Sperm Aspiration (PESA)**, **testicular sperm aspiration (TESA)** or **testicular sperm extraction (TESE)** to obtain small numbers of sperm directly from the testicle. These procedures will be further discussed if appropriate to you.

The PGD treatment cycle

The PGD cycle is made up of eight stages:

1. Stimulation of the ovaries
2. Egg collection
3. Insemination
4. Fertilisation
5. Embryo biopsy
6. Embryo testing
7. Embryo transfer
8. Pregnancy test

Starting your PGD cycle

Once we have confirmed that all aspects of preparation for PGD are complete, you may start your treatment. We are usually able to start treatment on your request, but occasionally we may have too many requests at once and will ask you to delay until your next period. Please call the PGD coordinator on the first day of the female partner's period. During Monday to Friday, 8am to 4pm, please call 020 7188 0504. If you call outside these hours, please leave a message stating you want to start treatment and say your name, a contact number and the date of the first day of your period. We will contact you within one to two days and offer you an appointment with the nurses to go through your treatment schedule. If you live far away, we can go over this on the telephone. Please do not worry if we do not return your call immediately. You have three weeks to receive your schedule and your medication, which is delivered by courier.

1. Stimulation of the ovaries

Your ovaries are stimulated with hormones, given to you as either a nasal spray or an injection given under the skin on the thigh or tummy. Buserelin (Suprefact®/Suprecur®) or Nafarelin (Synarel®) is the first medication you will be given. This part of the treatment is known as **downregulation** and:

- stops your ovaries from working temporarily;
- makes sure your ovaries respond better to the hormone injections;
- prevents you from releasing all of the eggs before we have time to collect them.

We will describe this in detail and give you a written schedule to follow nearer the time of treatment. Normally, you will start the first medication 21 days after the first day of your period and continue with it until two nights before your egg collection. We will let you know if you are on a different regime and exactly how this differs.

You might experience side-effects from the medication such as:

- hot flushes;
- night sweats;
- headaches;
- mood swings.

These side-effects can occur because the oestrogen hormone level in your blood drops. You may not experience any or all of these symptoms but, if you do, they will improve once you start your daily injections. An internal

ultrasound scan is performed after two weeks of downregulation. This will check that your ovaries are inactive and contain no large follicles or cysts and that the lining of the uterus (endometrium) is thin. If this is not the case, we will ask you to continue the medication for another week. If there is still an ovarian cyst present after three weeks of downregulation we will drain this using a procedure similar to an egg collection.

Please note:

It is normal to have a period during this time. This does not mean that the medication is not working. Do not stop the Buserelin/Naferelin until we ask you to. If you stop the medication too soon, your eggs could be released before we have had a chance to collect them.

Follicle Stimulating Hormone (FSH) injections (Gonal-F® or Puregon® or Menopur®) are given to you as a daily injection, over 10 to 14 days, to stimulate egg development in the ovaries. The injections are given just under the skin on the thigh or tummy. We will teach you or your partner to do this. Alternatively, you can make arrangements with your GP or local hospital to have the injections.

The FSH injection causes the oestrogen hormone level in your blood to rise. This might make you:

- feel bloated;
- feel nauseated;
- have tender breasts;
- feel increasingly emotional.

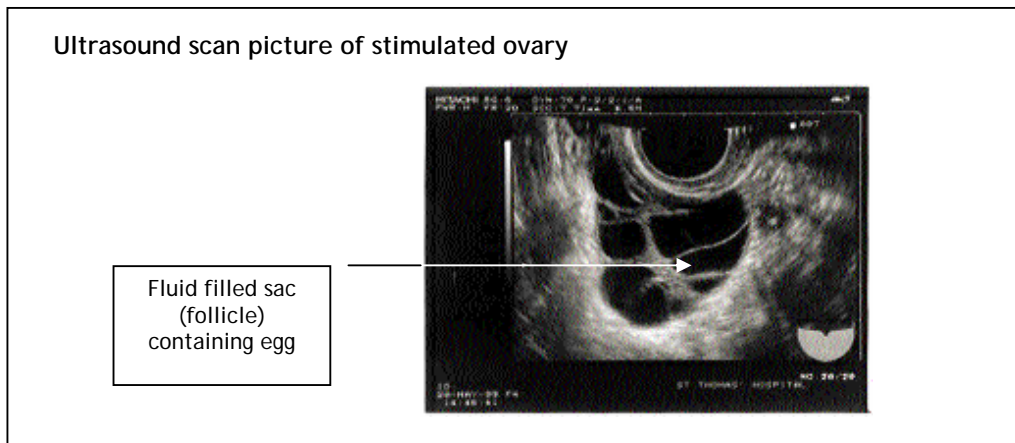
We will monitor your response to the FSH injections using internal scans, which measure the number and size of the follicles within the ovaries. The first scan is usually done nine days after starting the injections. In some cases a further scan is needed, usually 48 hours later.

The decision day

Our team of doctors, embryologists and nurses decides the best day for your egg collection. This decision is based on your medical history, response to the stimulation drugs and ultrasound findings. The cycle may be cancelled before egg collection if you under or over respond to the medication. Late at night and about 36 hours before the egg collection, hCG (Pregnyl® or Ovitrelle®) is given as a 'one off' injection. This is usually on the Wednesday of your scan if the follicles are ready for a collection on the Friday, but often on a Saturday evening, ready for a collection on the Monday. This medication causes the final stages of egg ripening to take place. The time you need to give yourself this injection at home is important and will be explained very clearly to you.

Please note:

- The eggs will not be mature and are unlikely to fertilise if they are collected too soon after the injection.
- If the collection is too long after the late night injection, you will have already ovulated and released all of the eggs before we can collect them.
- You do not need to take any more Buserelin/Naferelin after the hCG (Pregnyl® or Ovitrelle®) injection.



Risk of spontaneous pregnancy

If you have sexual intercourse during a PGD cycle, you must ensure that you use reliable contraception (for example, a condom). Most couples having PGD are fertile and could become pregnant. If this were to occur the pregnancy may be affected by the genetic disorder you carry, as embryos created in this way will not have been tested.

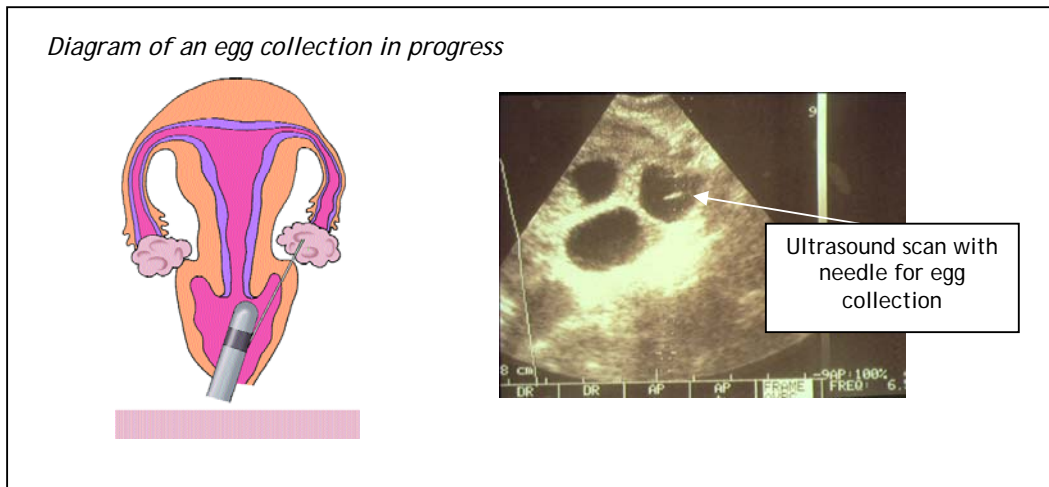
2. Egg collection

How do I prepare for egg collection?

You will be asked not to have anything to eat or drink after midnight on the night before your egg collection. This is because you will be very deeply sedated by an intravenous anaesthetic (anaesthetic into a vein in your arm) for the egg retrieval procedure. Any anaesthetic can make you feel sick and we want to reduce the chance of you vomiting, which can lead to complications. You may have a bath or shower on the morning of your egg collection. Please do not wear make-up, nail varnish, perfume or contact lenses because they can interfere with the equipment we use to monitor you during the procedure. As you will be asleep for the whole of the procedure, we do not encourage male partners to be present in the theatre. Your partner will need to provide a fresh sperm sample on the day of egg collection (see page 11).

What happens during the egg collection?

Once you are in the egg collection room you will be introduced to the team involved in the procedure. This will include a gynaecologist, nurse, embryologist, an anaesthetist and their assistant.



As part of our strict witnessing procedures, before you are given an anaesthetic we will ask you to tell us your name and date of birth. We check these details against your medical records and confirm the storage tubes for your eggs are correctly labelled. A small cannula (plastic tube) is put in a vein in your arm and used to give you medication during the procedure. We will give you extra oxygen to breathe and monitor your pulse, blood pressure and breathing throughout the egg collection. An ultrasound probe is placed inside the vagina and a fine needle is passed alongside, within a protective cover. The needle is gently passed through the vaginal wall into the nearest follicle in the ovary. The needle is passed from one follicle to the next until we have emptied all the follicles in one ovary. The needle is then removed and the procedure is repeated in the other ovary.

The embryologist will examine the fluid from each follicle under the microscope and check for any eggs. Each egg is then placed in special fluid and transferred to an incubator. Not every follicle will contain an egg and sometimes no eggs will be found during egg collection. We will discuss this with you before the procedure.

How long will it take?

Egg collection usually takes about 20 minutes, depending on the number of follicles to be drained. After the procedure you will rest on a bed in the recovery area for about one or two hours.

After the egg collection

Your partner or another adult will need to drive you home or accompany you in a taxi. We do not recommend public transport because it is not safe after having sedation. An anaesthetic can affect your reasoning, reflexes, judgement, coordination and skill.

If your partner needs to have a sperm retrieval procedure (PESA or TESA/TESE) on the same day, then he will be unable to drive, due to an anaesthetic. Please arrange for a responsible adult to drive you both home or accompany you in a taxi.

For 24 hours after the procedure please do not:

- stay alone – a responsible adult must stay with you during this time;
- drive any vehicle, including a bicycle;
- operate any machinery;

- attempt to cook, use sharp utensils or pour hot or boiling liquids;
- drink alcohol;
- smoke;
- take sleeping tablets;
- make any important decisions or sign any contracts.

It is not unusual to feel some lower abdominal or pelvic pain after the procedure. It is perfectly safe to take paracetamol for this, without altering your chances of IVF working or damaging a pregnancy. You might have some blood- stained vaginal discharge, which should become darker and stop after a few days. This blood is coming from the site where the needle has passed through the vaginal wall and is not the lining of the uterus breaking down, as in a normal period.

You will be prescribed the hormone progesterone for 17 days following the egg collection to help the lining of the uterus be as receptive as possible to the embryos. Progesterone is usually given as a suppository into the vagina each night, or can be taken as an injection every other day.

3. Insemination / injection of sperm

Your semen sample is prepared by separating the normal and active sperm from the ejaculated fluid. Fertilisation during PGD treatment will be done in one of two ways; either by in vitro fertilisation (IVF) or by Intracytoplasmic Sperm Injection (ICSI).

In Vitro Fertilisation (IVF)

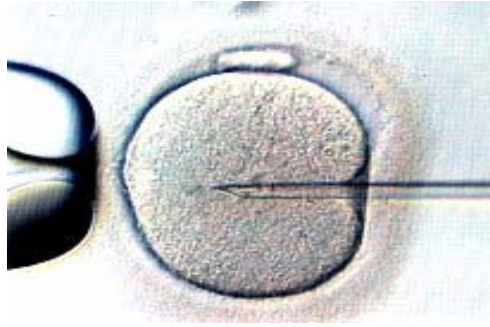
This is used mainly when carrying out PGD treatment for chromosome abnormalities or embryo sexing for X-linked conditions and when the sperm quality is good. During IVF sperm and eggs are placed together in a culture dish to allow fertilisation to occur in the laboratory.



Intracytoplasmic Sperm Injection (ICSI)

ICSI involves injecting a single sperm into the centre of each egg to achieve fertilisation. It was first developed in 1992 by a group of Belgian infertility specialists for use in men who had very poor sperm. It is used in PGD treatment however, for conditions caused by a single faulty gene to reduce the amount of non-embryo DNA (including sperm DNA) which could make the risk of a wrong diagnosis higher.

Single sperm inside pipette injected into egg



For both procedures, the dishes are carefully labelled with your name and unique number. They are left in an incubator in the laboratory overnight so fertilisation can take place.

4. Fertilisation

The next morning the embryologist carefully examines each egg to see if fertilisation has occurred. We will call you to tell you how many eggs have fertilised.

Rarely, about one in 100 times, none of the eggs fertilise and there are no embryos to be replaced. This is obviously very disappointing. We will offer you the earliest available appointment to see a PGD team doctor to discuss the cycle and your future treatment options. You will also be offered an appointment to see one of our counsellors.

Embryo development

Eggs that have fertilised are called embryos. As embryos develop, their cells divide. Two days after fertilisation, the embryo should have two to four cells. Three days after fertilisation, the embryo should have six to ten cells.



Day 1: fertilised egg



Late day 1: 2 cell embryo



Day 2: 4 cell embryo



Day 3: 8 cell embryo



Day 5-6: blastocyst



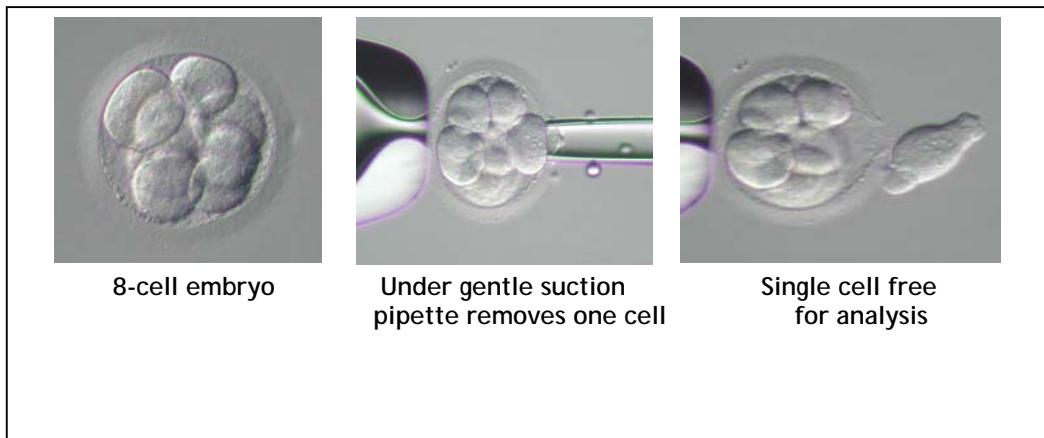
Late day 6: hatched blastocyst

5. Embryo biopsy

When embryos have grown to between six and 10 cells, the embryo biopsy procedure can take place. Each embryo in turn is held in place and a weak acid solution or laser is used to dissolve a small hole in the protective shell around the embryo (known as the zona pellucida). This enables one cell from the embryo to be removed under gentle suction using a fine tube, called a pipette. This technique is highly specialised and can only be carried out by embryologists with this skill who have a special licence from the Human Fertilisation and Embryology Authority (HFEA). It is our usual policy to remove only one cell, but in some circumstances we may have to take two.

The cells are then transferred in a small carefully labelled tube. This is given to the scientists who test the embryos for the DNA or chromosome abnormality. The embryos are then replaced in the incubator to continue to develop, whilst the analysis is completed.

Embryo Biopsy



6. Embryo testing

The types of tests we perform on the embryos will depend upon the reason behind you having PGD. Please refer to the supplementary leaflet inserted in the booklet for details of the testing that will be done on your embryos.

What happens in the laboratory?

We have extremely rigorous checking and witnessing protocols for all laboratory procedures. These are to make sure that the eggs, sperm and embryos used in your treatment belong to you and that the embryos are clearly labelled as affected or unaffected. We will explain these protocols to you before you start any treatment and are always happy to discuss them with you in more depth, at any stage of your treatment.

7. Embryo transfer

During embryo transfer we place the best unaffected embryos into your uterus. This is a much simpler procedure than egg collection and there is no need for an anaesthetic. During the procedure, we use an ultrasound scan through your tummy to help us to transfer the embryos where they have the highest chance of implantation.

How should I prepare for it?

You will need to have a full bladder for this procedure so your womb can be seen on the ultrasound scan. An ACU nurse will give you instructions on how to prepare for this.

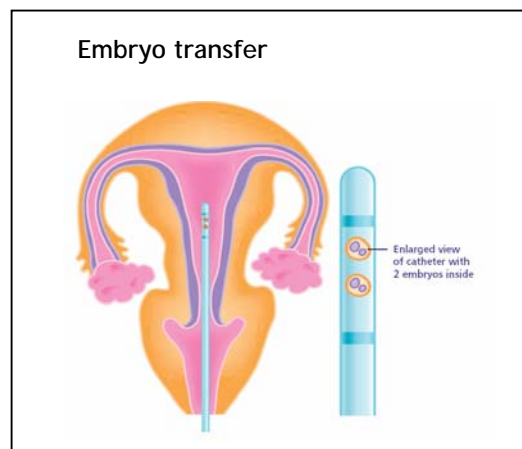
A full bladder makes the procedure technically easier as the bladder lies in front of the uterus and, in four out of five women, the uterus naturally bends forwards. Filling up the bladder will, therefore, 'straighten out' the uterus and make it easier to direct a soft catheter, which contains the embryos. This increases the chances of implantation.

What happens during the embryo transfer?

You and your partner will be asked to state your names and dates of birth before the transfer. The doctor and embryologist will check that the dishes containing your embryos are labelled with your name and unique identity number. The embryologist, together with one of our PGD team, will discuss with you which embryos are safe to be transferred.

A speculum, which is the instrument also used during a smear test, is placed in your vagina to clearly see your cervix (neck of the uterus). The outside of your cervix is cleaned and any mucus from inside your cervical canal is removed. This mucus might prevent the embryos getting to where we want them to be in the uterus.

The soft catheter, which holds the embryos, is inserted into your uterus. When we are happy that the catheter is in the best position, the embryos are gently injected. The catheter is then removed and checked to make sure all of the embryo(s) have been replaced. You will be able to empty your bladder immediately after the transfer, without any risk of losing the embryos. This process takes about 15 minutes. It is generally painless although the main discomfort is that the scan probes pushes down on your bladder which needs to be full for the procedure.



How many embryos will be transferred?

Current HFEA guidelines allow us to transfer a maximum of two embryos in women who are under 40 years of age. We will let you know if these guidelines change.

The number of embryos transferred is limited to lower the risk of a multiple pregnancy. Triplet pregnancies have a significantly increased risk of complications including miscarriage, high blood pressure and premature birth and even twins have more problems with delivery and at birth. Premature

babies have an increased risk of complications, such as a weakened immune system, physical and mental disability and feeding and breathing difficulties.

Blastocyst transfer

By day five after egg collection the embryos most likely to lead to pregnancy have undergone further development to become fluid filled balls of cells called "blastocysts". Recent evidence suggests that for some patients waiting to day five allows us to choose the embryos with the best potential of pregnancy to be transferred. The greatest benefit of blastocyst transfer is for patients who have a good chance of becoming pregnant. We can identify this group of women and they usually include those who have at least four good quality embryos to transfer. However blastocyst transfer is not always possible.

What happens if there are spare normal embryos?

It is possible to freeze embryos from a PGD cycle for later use. A frozen embryo, which has been thawed, is less likely to implant into the lining of the uterus than a fresh embryo, especially when it has been biopsied. We are selective about the embryos we choose to freeze but we will attempt to freeze any unaffected embryos that are suitable. Embryos are frozen at an extremely low temperature, which makes sure they do not deteriorate over the number of years they are stored. Even if you get pregnant in your initial attempt at treatment, you may wish to use frozen embryos, if possible, to expand your family at a later date. If you have frozen embryos, it is essential that you keep in touch with us to let us know what you wish to do with them and tell us of any changes of address.

Frozen embryo cycle

In a Frozen Embryo Replacement Cycle (FERC) we thaw your frozen embryos and transfer any that are suitable into your uterus. A PGD doctor or embryologist will discuss with you how many embryos are to be thawed in any one attempt. Although we are very selective about the embryos we freeze, after biopsy has taken place only a small percentage survive after being thawed. An advantage of a frozen cycle is that we do not need to use hormone injections to stimulate the ovaries nor do any further genetic testing of the embryos. For more information about FERC, please speak to a member of our team.

What happens after embryo transfer?

You will know if the treatment has been successful 10-12 days after embryo transfer.

We appreciate that this wait can be difficult for many people. Please do not be tempted to perform a pregnancy test earlier than advised. The hCG injection can stay in your blood stream for eight to 10 days and this will make the test positive, even if you are not pregnant. Please continue to take progesterone by inserting a suppository into the vagina each night or by an injection, every other day. Unfortunately, there is no evidence that anything you do at this stage will increase the chance of you becoming pregnant. We encourage you to return to work but you may prefer to have a few days off around the time of transfer. Having a bath or sexual intercourse are not known to affect the chances of pregnancy.

8. Pregnancy test

All women should perform a pregnancy test 12 days after the embryo transfer, even if they bleed before this time. This is essential because some patients who have bleeding after a cycle could have an ectopic pregnancy. This is uncommon but a potentially serious complication. We feel it is so important you do the test that we give you a pregnancy test kit and ask you to phone the Unit to give us the result.

A positive pregnancy test

This means that one or more embryos has implanted but we will not be able to see this on a scan until you are about six weeks pregnant (four weeks after embryo transfer). We will usually scan you twice to see how many embryos have implanted, whether they have developed a heartbeat and where they have implanted. Sadly, we sometimes diagnose miscarriages and ectopic pregnancies at this stage.

If we find that you have an ongoing pregnancy we will refer you back to your GP to arrange antenatal care at your local hospital. If you have a twin pregnancy we will refer you to a specialist for an early scan. Unfortunately, a number of pregnancies can still miscarry even if these early scans are encouraging.

A negative pregnancy test

Sadly, this means that the treatment has been unsuccessful. You might already have started bleeding but, if not, a period will come in the next few days. This might be heavier than normal due to the medications you have taken, which have made the lining of your uterus thicker than usual. We know this can be a very disappointing time and will offer you an appointment with one of our PGD team doctors to discuss the cycle and possible treatment options for the future. You might also find it helpful to see one of our counsellors.

Other information you need to know about PGD treatment

How will I get my medication?

We will send a prescription to a pharmaceutical company if your treatment is funded by certain Strategic Health Authorities or Primary Care Trusts and they will contact you personally to arrange delivery of the medication in time for treatment.

If you live in certain areas or are paying for your own treatment, we will send the prescription to an independent drug distributor who will liaise with you directly about payment and delivery of the medication. GPs must follow certain guidelines and are unlikely to prescribe or pay for your medication if you are paying for your own treatment.

Please note:

We do not make any profit on prescriptions or medications.

If you are funding your own treatment, we cannot allow you to start any medication until full payment for the treatment cycle is received.

What happens if my treatment cycle is cancelled?

The majority of women respond well enough to stimulation to progress to egg collection. About one in ten cycles has to be cancelled due to under or over response. If your cycle has to be cancelled, we will ask you to stop taking all of the medication. We will arrange a follow-up appointment with a senior ACU doctor, within four to six weeks. At this appointment we will reassess your treatment and plan a further attempt, if appropriate. We realise that it is disappointing to have a cycle cancelled. Please let us know if you would like to speak to one of our counsellors. Having your cycle cancelled does not mean you will never respond appropriately. We may be able to adjust your stimulation programme to achieve a better response. This will give you an improved chance of becoming pregnant in a further treatment cycle.

Why would a treatment cycle be cancelled?

There are two main reasons why your cycle may be cancelled:

- If too few follicles have developed in the ovaries, there are unlikely to be enough eggs for collection;
- If you have produced too many follicles, this means you are at risk of ovarian hyperstimulation syndrome (see page 26). This will be made worse if you become pregnant.

Why do cycles fail?

There are many factors which influence how an embryo will develop once it is replaced in the uterus. These factors are, unfortunately, poorly understood. It is impossible to see what happens after embryos have been replaced and it is often difficult to give a specific reason why a cycle has failed. This is especially difficult for most PGD couples who can conceive themselves.

In the majority of cases, the cause is likely to be that the embryos have stopped dividing before they attach to the uterus. We usually replace the embryos two or three days after fertilisation and in a natural conception they would usually start to attach on day five. Embryos are more likely to stop dividing if they are of poor quality.

If the uterine cavity is irregular due to fibroids or the fallopian tubes are swollen and contain fluid, we may recommend surgery to remove these before your next PGD cycle. If there have been problems with the thickening of the endometrium, we might add in extra medications to try and improve this in any future attempt.

In a successful cycle, what problems can occur in early pregnancy?

Sadly, as with any natural conception, the pregnancy might not progress normally and a miscarriage or ectopic pregnancy can occur. Please contact us if you develop:

- heavy bleeding;
- brown spotting (discharge);
- sharp abdominal pain, particularly if on one side;
- shoulder pain;
- faintness.

Future cycles

When can we have another go?

If your cycle fails, we generally like to see you to discuss how the cycle went and what we might do to try and improve your chances if you want another go. We recommend that you wait at least two months before you have another attempt so you can have a break from treatment and allow yourself time to recover from such a big disappointment. Your body also needs a chance to recover from the medication. If you have frozen embryos, we will discuss how we use these in the future.

How many attempts can we have?

We do not have a set limit for the number of attempts a couple may have although we know that if you do not become pregnant after two PGD attempts, your chances of succeeding are low. We will be honest with you and may advise you to stop treatment. Some Strategic Health Authorities and Primary Care Trusts will only pay for one cycle of treatment and you may have to pay for other attempts yourselves if you can.

What are the risks of PGD treatment?

All types of medical treatments and procedures have risks. The PGD team will speak with you about the risks involved. Possible risks associated with PGD include:

Multiple pregnancy

The major complication of PGD is multiple pregnancy. We recommend that if the woman is under 35 years of age and the embryos are of good quality that

only one embryo is transferred (please see details on page 21). This gives a better overall chance of having a healthy baby. However sometimes we may recommend two embryos for transfer. We cannot exclude the possibility of a multiple pregnancy. The risk of complications is higher in a twin pregnancy. If you decide to have a confirmatory test in a pregnancy to ensure the PGD result was correct then, although possible, it can be more difficult to do such tests on a twin pregnancy. Our centre has recently published the outcome of research showing that single embryo transfer does not decrease the pregnancy rate, but improves the chance of having a healthy singleton pregnancy. Overall our PGD twin pregnancy rate is around 19% (about 1 in 5 of all pregnancies).

Ovarian Hyperstimulation Syndrome (OHSS)

Some women respond very sensitively to the fertility drugs and produce many follicles. This causes the ovaries to enlarge and blood oestrogen levels to rise. This is more common in younger women and those with polycystic ovarian syndrome (PCOS).

Development of OHSS is not always predictable or avoidable. We will identify if you have an increased risk by monitoring ovarian stimulation with ultrasound scans. We will occasionally recommend that you do not take the hCG injection, which keeps the hormone levels high. We will not collect the eggs and ask you to continue taking the downregulation medication until your symptoms improve. If we do collect the eggs, we might recommend that any embryos created are frozen, for replacement in a future cycle. A frozen embryo replacement cycle will not cause OHSS as the ovaries are not stimulated. If we replace the embryos in a fresh cycle and you become pregnant, your blood oestrogen level will start to rise again and make your condition worse. Symptoms of OHSS are most common around the time of egg collection or about ten days after embryo transfer. You may find that things improve only to worsen again nearer to the time of your pregnancy test.

In OHSS, the ovaries can enlarge to up to three times their normal size. Your blood protein level drops which causes fluid to leak out into the abdominal cavity or around the lungs. This can result in problems producing urine, mineral imbalances in your blood and clotting problems. Symptoms include:

- abdominal (tummy) pain and swelling;
- passing small amounts of concentrated urine;
- thirst;
- nausea and vomiting;
- diarrhoea;
- dizziness;
- shortness of breath.

If you have any of these symptoms please contact us immediately so we can give you the necessary advice. Most cases of OHSS are mild and are resolved by drinking three litres of fluid and using mild pain-relieving medication, such as Paracetamol.

If we are concerned that you are at risk of developing moderate or severe OHSS we will keep you under regular review. If your symptoms worsen we might have to admit you to St Thomas' Hospital for monitoring and treatment. Please also contact us if you have been to another hospital for advice or treatment. Having OHSS will not jeopardise the chances of you becoming

pregnant. However for those of you caring for children, a hospital admission may create difficulties with child care and we advise you to consider this when planning a PGD cycle.

Pelvic infection

Pelvic infection can occasionally follow an egg collection and, rarely, an abscess might develop. We try to make sure this does not happen by performing the collection under sterile conditions and giving antibiotics to women who are at higher risk of infection. It is not always possible to prevent an infection, despite these measures. Symptoms of an infection include:

- pain;
- bright red vaginal bleeding;
- smelly vaginal discharge;
- diarrhoea;
- fever;
- generally feeling unwell.

We will admit you to St Thomas' Hospital for antibiotic treatment. In severe cases, an operation might be necessary.

Other complications

There is a very small risk that the needle used for egg collection can puncture the bowel or blood vessels. The needle used is very fine and it is unusual to have any complications. Most cases of vaginal bleeding can be stopped at the end of the procedure by applying pressure to the puncture site. If there is a concern that a tiny hole has been made in the bowel, you will be given antibiotics.

Please contact us if you feel any of the following symptoms:

- pain in your tummy;
- swelling/bloated feeling in your tummy;
- nausea and vomiting – especially fluids;
- you are passing a small amount of urine or if your urine seems concentrated;
- diarrhoea;
- shortness of breath;
- feeling feverish, shivery or generally unwell;
- heavy or irregular vaginal bleeding;
- dizziness.

Are there any risks to children born following PGD?

At present, no strong evidence exists to suggest that PGD babies are at any greater risk of abnormality than babies conceived through other forms of assisted reproduction (see information on outcome of IVF and ICSI babies, below). However long-term follow up data is limited and all couples undertaking PGD must be aware that it is still a relatively new technology with unknown long-term outcomes. At present most of the data collected about babies born following PGD relates to the outcome at birth and does not provide information on older children. There are plans to collect information about these children when they are a little older. In our centre we ask for information about the wellbeing of children at birth and all are offered an examination by a paediatrician (children's doctor) when they are one and two years old.

What are the risks associated with IVF and ICSI?

There are risks associated with any medical procedure and the PGD team will explain the risks to you before you sign the consent form. Please ask questions if you are uncertain. The first IVF baby was born in 1978 and the first following PGD in 1990. There is evidence that babies born following IVF or ICSI are more likely to be born early (prematurely) and to weigh less than naturally conceived babies born at the same age. In 2002, a large study from Australia suggested that IVF treatment might almost double the risk of abnormalities. It is important to remember that the risk of an abnormality in a natural conception is about 3%.

The Brussels Centre, which developed ICSI, has looked at the number of major malformations (those causing loss of function or requiring surgical correction) and neonatal (post birth) complications in 2889 children born after ICSI (1991-1999). It found no difference in the rates of complications when compared with the complications in 2995 children born after IVF (1983-1999).

More research is needed and is ongoing. Please feel free to talk to us about any concerns you might have about ICSI treatment.

Follow up of PGD babies

The number of babies born following PGD is quite small, so it is important to continue to monitor the progress of these babies. We collaborate with several other PGD centres across Europe, the USA and Australia to ensure that there is more data available to help answer the above question more thoroughly. We have a responsibility to follow up children born following treatment so that we gather information about the potential impact of PGD. All couples who have children following PGD are asked to bring their children for a paediatric check up at one, two and five years of age.

Research

We participate in research projects which are important for the continuing development of treatment.

Our research projects aim to:

- investigate how to improve the success rates of IVF
Immature eggs, or eggs which have failed to fertilise, and poor quality embryos, are examined for the presence of potential defects. It is hoped to develop sensitive tests which in the future will more easily be able to identify embryos which are most likely to implant.
- understand more about sperm and egg interaction and embryo development
- gain more information about preimplantation diagnosis for genetic disease

Immature or unfertilised eggs or poor quality embryos which would normally be discarded, are used to improve the specialised techniques used for PGD. This aims to increase the number of genetic disorders that can be diagnosed and to check the accuracy of the diagnostic tests and introduce techniques to improve them.

- understand how stem cells develop from early embryos
- try and develop stem cells for treatment and research

Stem cells are contained within a certain part of an embryo which has grown for five days. These stem cells have the potential to grow into the different types of tissues that make up the human body. To access these cells for research, it would be necessary to separate them from the other cells and this would mean that the embryo could no longer survive. However, by taking the stem cells and growing them in a dish in the laboratory, researchers can study what makes them grow into the different types of cells in the body, for example, skin cells, muscle cells, nerve cells etc. The process of separating and growing the stem cells in a laboratory is a difficult process and is not successful in many cases.

You are under no obligation to donate your spare eggs, sperm or embryos to research. Your decision will not affect your acceptance for treatment and it will not change in any way how your treatment is carried out.

This research is done in an anonymous manner. Any information that could identify you is removed and although the research might not be of direct benefit to you, it is aimed at improving our understanding of human development and genetic disease, ultimately improving the success of this type of treatment.

All research is strictly regulated by the HFEA and has to have the approval of the Hospital Ethics Committee. Research on early human embryos can be carried out only when they have been assessed as unsuitable for use in current or future (via freezing) treatment.

Before we can use your spare eggs, sperm and/or embryos for research, you must sign a consent form for each research project to confirm that you agree to take part. If you have any questions or concerns about any of the research projects, please see the enclosed leaflet or ask any of the medical, embryological or nursing staff for more information. You may decide that you wish to consent to some but not all of the research projects and this can be indicated on the consent forms.

Eggs, sperm and embryos donated to research are never used to treat other patients.

The Human Fertilisation and Embryology Authority (HFEA)

What does the HFEA do?

The HFEA was set up to license, monitor and regulate clinics performing IVF and treatments using human sperm, eggs and embryos, in accordance with the Human Fertilisation and Embryology Act (1990).

The HFEA:

- inspects and licenses clinics offering treatment to infertile couples;
- publishes a *Code of Practice* for centres carrying out licensed treatment;
- gives advice and information to patients, clinics and doctors;
- publishes patient information such as *Your Guide to Infertility*, *Patients' Guide to IVF clinics*, *Patients' Guide to DI* and other booklets available in our Unit.

The guidelines in the *Code of Practice* consider:

- the respect which is due to human life at all stages in its development;
- the right of people who are or may be infertile to proper consideration of their request for treatment;
- a recognition of the benefits, both to individuals and to society from the responsible pursuit of medical and scientific knowledge.

We must have an HFEA licence to provide PGD treatment for each genetic condition that we offer.

Welfare of the child

Under the terms of the Human Fertilisation and Embryology Act (1990), we must consider the welfare of any existing or future child(ren) born as a result of treatment. As part of this initial assessment, we will ask you to complete a questionnaire. If any information given raises concerns, these will be discussed with you and you will be asked to meet with a counsellor. Very occasionally, it may be necessary to delay or cancel your treatment.

The HFEA register

Although most couples requesting PGD will not require egg or sperm donation, some will and it is important that you are aware of the law in relation to this. The HFEA keeps a confidential register about patients, donors and treatments. This register was set up on August 1, 1991 and has information about children conceived from licensed treatment since that date.

If you are aged 16 or over and considering getting married or aged 18 or over, you can ask the HFEA whether they were born as a result of licensed assisted conception treatment, and if so, whether you are related to the person you want to marry.

Although most couples having PGD do not use egg or sperm donors, it is important to know that the HFEA collects information from donors on their physical appearance, interests and occupation. From April 1, 2005, the HFEA also holds identifying information about the donor, including the name and date of birth. Children conceived as a result of sperm, egg or embryo donation will also have access to this information when they reach 18 years of age.

If you would like more information about the HFEA, you can contact it directly at:

The Human Fertilisation and Embryology Authority
21 Bloomsbury Street
London WC1B 3HF
Tel: 020 7291 8200

Email: admin@hfea.gov.uk
www.hfea.gov.uk

Further information

What if I am unhappy with the service?

Please talk to us if you are unhappy with any aspect of your treatment in the ACU. We will do everything possible to put things right. If you do not want to do this

or are not satisfied with the response you receive, then please contact **the Patient Advice and Liaison Service (PALS)**. PALS offers patients and visitors information, support and advice about the services at Guy's and St Thomas' NHS Foundation Trust. Tel: 020 7188 8801 at St Thomas' or 020 7188 8803 at Guy's or email pals@gstt.nhs.uk

How can I find out more information?

If you have any questions or concerns about assisted conception and the services we provide, please speak to a member of the PGD team. Phone us on:

PGD Coordinator: 020 7188 0504

Genetic Counsellors: 020 7188 1364

Information about our services is also available on our website at

www.pgd.org.uk

You might also find the following information helpful:

Knowledge & Information Centre (KIC)

If you want more information about health conditions, support groups and local services, or want to search the internet and send emails, please visit the KIC on the Ground Floor, North Wing, St Thomas' Hospital. Tel: 020 7188 3416 or email kic@gstt.nhs.uk or visit their website at www.kic.gstt.nhs.uk.

NHS Direct can also provide health information. Tel: 0845 4647 or visit their website at www.nhsdirect.nhs.uk.

How to contact us

The PGD team work in two departments. The first consultation will be in the Genetics Department. PGD treatment is based within the Assisted Conception Unit (ACU).

ACU,
11th Floor, Tower Wing,
Guy's Hospital
Great Maze Pond,
London SE1 9RT
Tel: 020 7188 0504
Email: pgd@kcl.ac.uk

Clinical Genetics,
7th Floor, Borough Wing,
Guy's Hospital,
Great Maze Pond,
London, SE1 9RT
Tel: 020 7188 1364

We are open Monday to Friday, 8.30am to 4.30pm and the Unit is easily accessed directly from London Bridge Station via the 2nd floor entrance to the hospital. Follow the purple signs to Tower Wing

If you have any minor problems outside of office hours, there is an answering machine where you can leave a message. We will contact you as soon as possible, after the Unit re-opens. There is always a doctor from the unit on call for emergencies. If you have an urgent problem at night or during the weekend, please call 020 7188 2300. The answerphone message will also give you a mobile telephone number to call. This number should only be used when you are in need of urgent medical assistance, not for appointments and scheduling. The doctor on call is unlikely to be in the Unit and will not have access to your notes or the appointment books. If you are diverted to voicemail, please leave a message with your name and contact number so the doctor can return your call.

Glossary

ACU Assisted Conception Unit.

Anovulation When an egg is not released from the ovary, each cycle.

ART Assisted Reproductive Technology. Conception which involves the use of medical technology, e.g. IVF and PGD.

Azoospermia The complete absence of sperm in the male ejaculate.

Blastocyst An embryo that has developed for five to six days after fertilisation.

BMI Body Mass Index – your weight (in kg) divided by your height (in square metres).

Cervix The narrow passage at the lower end of a woman's uterus, connecting to her vagina.

Chromosome A threadlike structure of DNA containing genes, which carries genetic information.

Downregulation Part of infertility treatment, using medication, which make a woman's ovaries temporarily inactive to prevent the release of eggs before they are collected.

DI Donor insemination. The introduction of donor sperm into the uterus.

DNA Di-ribonucleic acid basic formation of genetic code.

Ectopic pregnancy A pregnancy, which implants outside the uterus.

Egg Gamete produced by a woman during her monthly cycle.

Embryo A fertilised egg.

Endometrium The lining of the uterus, which grows and sheds during a normal menstrual cycle and which supports a pregnancy.

Endometriosis A female condition in which endometrial cells, which normally line the uterus, implant around the outside of the uterus and/or ovaries, causing pain, internal bleeding and reduced fertility.

Epididymis The area above a man's testicle where sperm are stored.

Fallopian tubes The tubes on either side of the uterus where the sperm should fertilise the egg and the embryo should travel through to the uterus.

Fertilisation The penetration of an egg by a sperm resulting in the formation of an embryo.

Fibroids Benign (non-cancerous) overgrowth of the muscle layer of the womb. Can be linked with fertility problems if the fibroids grow into the lining of the womb or are very large.

Follicle A collection of fluid within the ovary, which usually contains an egg if it grows to a certain size.

FSH Follicle Stimulating Hormone. The hormone produced by the brain to stimulate the ovary to make one or more follicles grow. Is available in synthetic form to use in fertility treatment.

Gametes Eggs or sperm.

hCG Human Chorionic Gonadotrophin. An injection given to a woman having IVF to mature the eggs. Also indicates pregnancy.

HFEA Human Fertilisation and Embryology Authority. A Government body which regulates clinics performing IVF and other treatments using sperm, eggs and embryos.

ICSI Intracytoplasmic Sperm Injection. When a single sperm is injected into a single egg.

IVF In Vitro Fertilisation. Eggs and sperm are mixed together in a laboratory dish to achieve fertilisation. Any fertilised eggs are then placed inside the uterus.

In vivo In the body.

In vitro In a laboratory setting.

Multiple pregnancy Two or more babies.

Oestrogen Female sex hormone produced by the ovary. Levels change throughout the cycle.

Oocyte/Ovum The female gamete (egg).

Ovary The female reproductive organ, which produces egg(s) from hormone stimulated follicles.

OHSS Ovarian Hyper Stimulation Syndrome. Can occur if there are a large number of follicles/eggs, causing abdominal bloating, pain, nausea and breathlessness.

PCOS Polycystic Ovarian Syndrome. A condition where multiple, small follicles arrange themselves around the outside of the ovary. Can be an isolated ultrasound finding but also associated with hormone imbalances and irregular ovulation. Made worse by excess weight.

PESA Percutaneous Epididymal Sperm Aspiration. A fine needle is passed into the epididymis, under local anaesthetic and sedation to find sperm in men where there is a blockage between production in the testicle and appearance in the ejaculate.

PGD Preimplantation Genetic Diagnosis. The removal of one or two cells from an embryo to test for genetic diseases. Only unaffected embryos are transferred.

Semen Ejaculated sperm.

Sperm Male gametes.

Stem cells Cells which have the potential to grow into different types of cells in the body.

TESA Testicular Sperm Aspiration. A sperm extraction technique, which involves inserting a needle into a man's testicle to remove testicular tissue.

TESE Testicular Sperm Extraction. A sperm extraction technique, which involves making a small cut in a man's scrotum and the removal of a small amount of testicular tissue.

Uterus A female organ, also known as the womb.

Zona Pellucida The outer coating of the egg. Embryos/fertilised eggs have to 'hatch' out of this before they can implant.

Factual information presented within this communication is based on accurate contemporaneous peer reviewed literature. Evidence of sources can be provided on request.

Further Information

For further information, please contact us:

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Information about our services can also be found on our website at

www.pgd.org.uk

Guy's Hospital is in the heart of London

The Trust's website provides further information about our services

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