

# PGT and IVF

Information about preimplantation genetic testing and IVF treatment

## What is PGT?

PGT stands for preimplantation genetic testing. It is meant for couples who have a significantly higher risk of having a baby with a severe genetic disorder or who have a high risk of a miscarriage due to a chromosome abnormality.

PGT requires in vitro fertilisation (IVF) treatment. During IVF, egg cells are fertilised by sperm in the IVF laboratory. Fertilized egg cells are called embryos. After fertilisation one or more cells are removed from each embryo. These cells are then screened in the Maastricht UMC+ PGT laboratory for the presence of the genetic disorder. Only embryos that do not carry the genetic disorder will be placed into the uterus.

## Preparing for the intake consultation

It is important that both partners read this leaflet carefully and watch the video on the website. If you have any questions, write them down and ask them during the consultation.

<https://www.pgtnederland.nl/pgt-intakegesprek>

The consultation helps us get to know you. You can explain your medical history and why you are considering PGT. We will briefly discuss your family: your father, mother and any children or siblings you may have. We will also explain the steps involved in the PGT procedure and how long they may take.

You may also indicate where you would like to undergo the procedure. In addition to Maastricht UMC+, you can choose to have the IVF procedure performed at UMC Utrecht, UMC Groningen or Amsterdam UMC.

## Costs

An informative or intake consultation for PGT is likely to cost you the amount of the medical insurance excess (also known as 'own risk' or 'eigen risico' in Dutch, i.e. the amount that you are required to pay for medical treatment yourself before your insurance starts to cover your costs).

The costs of the consultation and of the PGT treatment are always covered by the woman's insurance policy, even if it is the man who has the genetic disorder. Be sure to check in advance whether your health insurer covers PGT treatment under its basic insurance package. This is your responsibility as the patient. You will find more information on our website.

<https://www.pgtnederland.nl/kosten>

## Do you meet the PGT and IVF criteria?

### PGT criteria

- The Maastricht UMC+ PGT centre assesses the genetic aspects of the disorder for which you are considering PGT.
- Since its introduction in 1995, PGT has been used to screen for multiple disorders. If you request PGT for a disorder that has not been tested before, the national PGT indications committee must first approve the development of a new test.
- Maastricht UMC+'s Clinical Genetics Laboratory will assess whether the new test is technically feasible.

### IVF criteria

- A woman must be physically able to undergo IVF treatment and endure pregnancy. This is particularly important if she is the person who has the genetic disorder.
- If the woman is above a certain age, we will assess whether PGT is still appropriate.
- Due to the high risk of complications in IVF treatment and pregnancy, the woman's body mass index (BMI) may not exceed 35.
- The procedure involves for the woman an internal examination and an ultrasound scan of the ovaries.
- The procedure involves for the man a semen analysis. To ensure that this is successful, the most recent ejaculation should not have taken place more than 7 days ago and not less than 2 days before the examination.
- Blood tests of both partners will be performed to exclude certain infectious diseases.

After a few weeks, the gynaecologist will discuss the results with you and decide whether you can undergo IVF treatment.

## Preparatory genetic test

During the preparatory genetic test, blood samples will be taken from both partners and often from family members as well to analyse the genetic disorder. This blood test is required even if your blood has been previously tested for the disorder. Blood samples will be tested only for the genetic disorder for which you are undergoing the PGT procedure. You may have the blood samples taken at a hospital in your own region.

If the preliminary genetic examination is complete and the IVF test does not reveal any problematic factors, PGT treatment will be planned.

## The IVF treatment

During IVF, fertilisation occurs in the laboratory by combining egg cells and sperm cells. Before that can happen, egg cells must first be matured.

### Stimulation

Medication is used to stimulate egg cell maturation. Your doctor will discuss this with you. During the treatment, you will have several ultrasounds to monitor the growth of the follicles. Follicles are small sacs containing an egg cell. If there are enough follicles (usually after 10 to 14 days), the egg retrieval follows.

### Egg retrieval

The doctor inserts a needle in the ovaries and collects the fluid containing the egg cells. Prior to the procedure, you will be given pain medication intravenously. Gynaecologists call this 'harvesting the egg cells'. The follicular puncture takes about 15 to 30 minutes.

### Fertilisation

After the egg retrieval, the egg cells are fertilised in the IVF laboratory by combining them with the man's sperm cells. After the egg cells have been fertilised in the IVF laboratory, they begin to divide. We then refer to them as embryos. After 3 to 5 days, the embryo consists of an average of 8 to 60 cells.

## The biopsy

To perform the PGT analysis, we must first perform a biopsy in which we extract one or more cells from the embryo. There are two types of biopsies: a blastomere biopsy and trophoctoderm (TE) biopsy. During the procedure, a laser beam is used to create a tiny opening in the shell surrounding the embryo. A thin needle is then inserted into the opening and used to extract one or more cells. These cells will be analysed in the laboratory to determine whether the embryo is affected with the familiar disorder.

A **blastomere biopsy** is performed on the 3rd day after fertilisation. One cell is removed (in some cases two). Once we have completed the PGT analysis, we transfer one of the embryos found to be unaffected by the genetic disorder to the uterus.

A **trophoctoderm or TE biopsy** takes place on the 5th or 6th day after fertilisation. Several cells are extracted. Once we have completed the PGT analysis, we transfer one of the embryos found to be unaffected by the genetic disorder to the uterus.

## The preimplantation genetic test

During PGT, the cells obtained from the biopsy are tested for the genetic disorder for which you requested screening. This test always takes place in Maastricht and can take between 24 hours and 8 weeks, depending on the indication. Once the test results are known, we decide which embryos are eligible to be transferred to the uterus.

### Embryo freezing

Depending on the duration of the PGT analysis, in case of the blastomere biopsy, an embryo may be transferred to the uterus without being frozen beforehand. The best-quality embryo that does not have the genetic disorder will then be transferred to the uterus.

The other unaffected embryos will be frozen, as back-up in case the first procedure is unsuccessful or in case the couple would like to have a second child in the future.

If the PGT analysis takes longer, all embryos are frozen after the biopsy.

After a TE biopsy, all embryos are frozen. Following the genetic test, one of the unaffected embryos will be thawed and then transferred. In this case too, the other unaffected embryos will be stored.

## Pregnant and PGT diagnosis check

If you become pregnant after the PGT procedure, we can, at your request, check whether the PGT diagnosis was correct.

- You can have a chorionic villus sampling test performed in the 11th or 12th week of pregnancy.
- Or you can opt for an amniocentesis during the 16th week of pregnancy.
- Every pregnant woman in the Netherlands can choose to do non-invasive prenatal test (NIPT). More information is available on [meerovernipt.nl](https://www.meerovernipt.nl).
- In the 13th and 18th-20th week of pregnancy, you can choose to undergo an extensive ultrasound scan.

## Risks for the woman and the child

PGT carries some risk for the woman because it involves IVF treatment. There is a very small risk of complications arising from IVF. One of the more frequent ones (in 1-2% of all cases) is hyperstimulation, which means that too many follicles mature in the ovaries. The woman may experience abdominal pain and fluid retention. Sometimes this means that the procedure has to be discontinued, while in other cases hospital admission may be required. Other potential complications are haemorrhage or an infection.

As far as research can determine, the removal of one or more cells in a 3- to 5-day-old embryo (the biopsy) will not affect the embryo's chances of development. The latest international scientific literature also indicates that PGT does not increase the chance of a baby with a congenital disorder.

## Reliability and chances of success

PGT has reliability of 95-98%. In other words, after a PGT procedure, there is only a small risk (2-5%) of giving birth to a child affected by the disorder for which screening was requested.

The success rate of PGT is approximately 20% for each procedure. In the Netherlands, a couple may undergo 3 PGT procedures, provided that there are no special circumstances. On average, the PGT treatment leads in 40-50% of the couples to ongoing pregnancy (a viable pregnancy of 12 weeks or more) within 3 procedures.

If you do not become pregnant after the PGT procedure, than you can together with the IVF department decide whether you wish to continue the treatment. If you do, we will then schedule the next treatment cycle.

### Contact

In case you have questions or if you are not able to come to the hospital, please let us know.

Maastricht UMC+

Klinische Genetica

T: (+31)(0)43.387.58.55

E: [polikliniek.klinischegenetica@mumc.nl](mailto:polikliniek.klinischegenetica@mumc.nl)

### Website

- [pgtnederland.nl](http://pgtnederland.nl)
- [klinischegenetica.mumc.nl](http://klinischegenetica.mumc.nl)
- [mumc.nl](http://mumc.nl)

**PGT Nederland** is a partnership between the Maastricht UMC+ PGT-centre and UMC Utrecht, UMC Groningen and Amsterdam UMC.