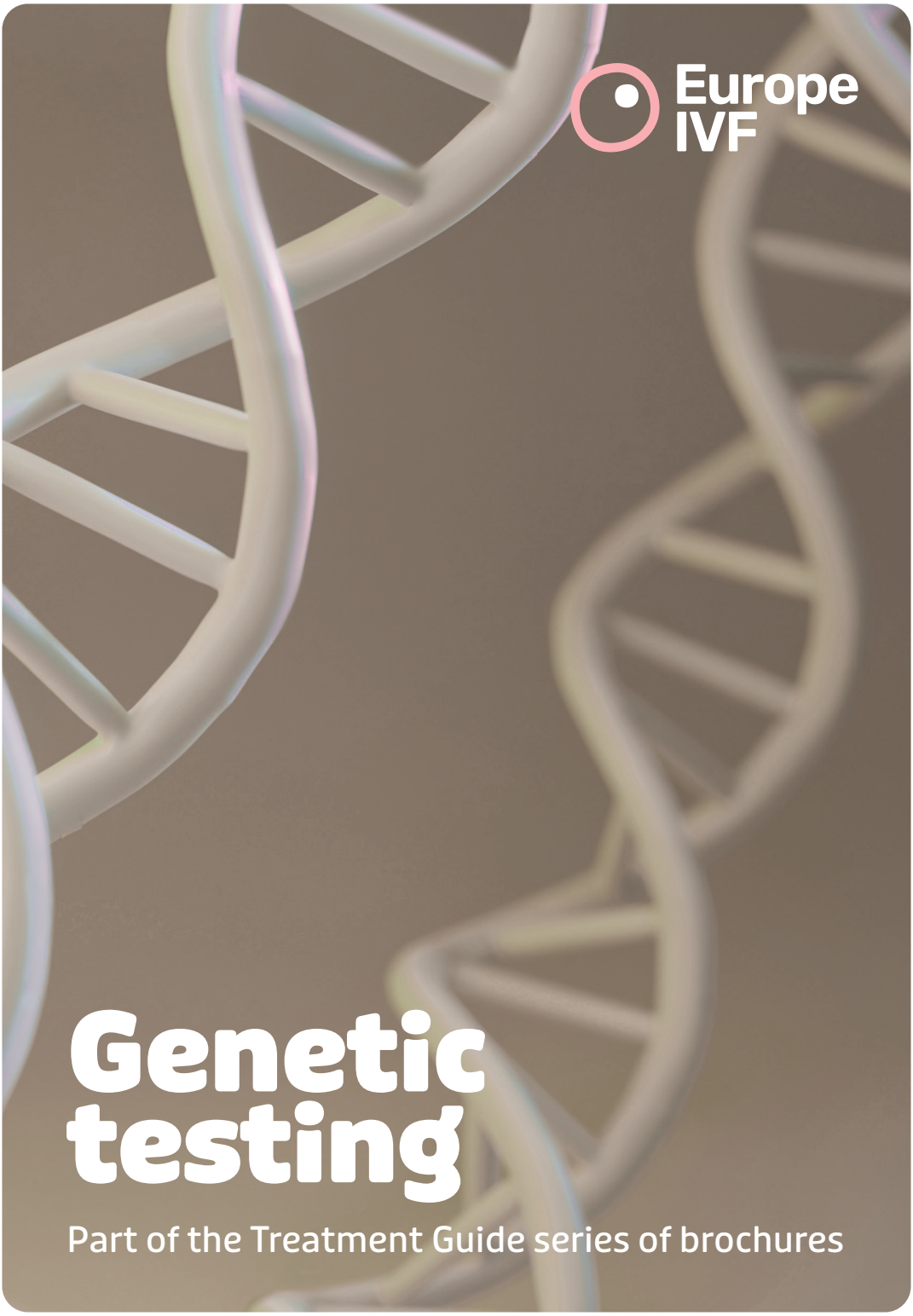

Europe
IVF
Genetic
testing
Part of the Treatment Guide series of brochures

Attention:

The information provided in this brochure should not be used as a substitute for information or advice provided by a doctor. Your doctor can help you choose the best option for you.

Introduction

Genetic testing in IVF (in vitro fertilization) is a key step for couples struggling with infertility or with an increased risk of genetic disorders. This brochure provides an overview of genetic tests available in IVF and their importance.

When is genetic testing recommended?

Genetic testing is recommended for various reasons:

- **Family medical history:** If your family has a history of genetic disorders, genetic testing is recommended to determine the risk of passing these disorders to your children.
- **Personal risk factors:** Those with personal risk factors, such as advanced age or exposure to harmful substances, should consider genetic testing to identify possible genetic anomalies or risks associated with pregnancy.
- **Pregnancy planning:** Couples planning pregnancy can undergo genetic testing to determine the risk of passing on genetic disorders to their children and make informed decisions about their future children.
- **Infertility issues:** Genetic testing can be part of the diagnostic process for couples struggling with infertility to identify potential genetic causes of infertility and potential risks for their children.
- **Before IVF:** Before you undergo an IVF cycle, genetic testing may be recommended to identify genetic abnormalities or risks associated with pregnancy, which can lead to better IVF results and a reduced risk of miscarriage. Genetic testing is also recommended if you have experienced repeated unsuccessful IVF cycles.

Types of genetic tests in IVF

1 Preconception genetic testing

Preconception genetic testing is a process that helps couples understand genetic risks that may affect their future children. This test can identify genetic disorders and carriers of genetic mutations, which can help you plan a healthy pregnancy.

It is the first step in ensuring future parenthood.

Who is preconception genetic testing recommended for?

- ✓ **Preconception genetic testing is an important step for couples planning a pregnancy, especially if they have a family history of genetic disorders or are carriers of genetic mutations. This guide provides information about preconception genetic testing, its importance and the process.**
- ✓ **Couples with a family history of genetic disorders: If you have a family history of genetic disorders, it may be helpful to undergo preconception genetic testing to identify risk factors for your children.**
- ✓ **Carriers of genetic mutations: If one or both partners are carriers of genetic mutations that can be passed on to offspring, preconception genetic testing is recommended to determine the risk of hereditary diseases.**
- ✓ **Couples that already have a child with a genetic disorder, or couples experiencing repeated miscarriages or infertility.**

What to expect

There are generally several steps in preconception genetic testing:

- **Consultation with a geneticist:** The couple meets with a geneticist or a doctor specialising in reproductive medicine to go over their family medical history and personal risk factors. This interview may include questions about genetic disorders in the family, the history of previous pregnancies and other relevant information.
- **Sample collection:** The collected sample is used for genetic testing.
- **Genetic analysis:** The laboratory performs a genetic analysis of the sample, which may include testing for genetic mutations associated with inherited diseases or looking for chromosomal abnormalities.
- **Evaluation of the results:** After the genetic analysis, the doctor evaluates the results and discusses them with the couple. If any genetic risk factors are identified, the doctor can discuss its potential effects on pregnancy planning and recommend further steps, including potential treatment or genetic counselling.
- **Decision and planning:** Based on the results of genetic testing and a discussion with our reproductive medicine doctor, the couple can choose the next steps, including potential changes in pregnancy planning, complementary treatments, or further genetic counselling.

Types of tests

Preconception genetic testing includes several types of tests that can help identify genetic risks and inform future parents about possible genetic disorders. A list of the main types of tests is shown below:

- ✓ **Peripheral blood karyotyping: This test analyses the number and structure of chromosomes; it helps detect chromosomal abnormalities that can cause genetic disorders such as Down syndrome or Turner syndrome.**
- ✓ **Carrier tests (basic and advanced): Carrier screening focuses on finding hidden carriers of 'key' recessive gene mutations causing genetic disorders that can affect the children of healthy carriers. These include cystic fibrosis, spinal muscular atrophy and congenital metabolic disorders.**

Why should you undergo preconception testing?

- **Prevention of genetic disorders:** Identifying risk factors before conception can reduce the likelihood of having a child with a genetic disorder.
- **Informed decision-making:** It gives couples information that can help them make informed decisions about family planning.
- **Better pregnancy planning:** It helps doctors and parents prepare for potential health challenges.

Preconception genetic testing is a useful tool for couples planning a pregnancy. It gives the couple information about their genetic risk and allows them to make informed decisions about their future children. Consultation with a geneticist is crucial to choosing the right tests and understanding the results.

2 Preimplantation genetic testing (PGT)

Preimplantation genetic testing (PGT) is a specialised type of genetic test performed on embryos before they are transferred to the uterus during in vitro fertilisation (IVF). PGT aims to identify genetic abnormalities in embryos, helping to increase the chances of a successful pregnancy and the birth of a healthy baby.

Who is preimplantation genetic testing recommended for?

- ✓ **Couples with repeated unsuccessful IVF cycles or miscarriages: PGT can identify genetically normal embryos, which can increase the chances of a successful pregnancy.**
- ✓ **Couples at higher risk of genetic disorders: If you have a family history of genetic disorders or are a carrier of genetic mutations, PGT can help select genetically healthy embryos for transfer.**
- ✓ **Women over 35 years of age (at the time of childbirth)**

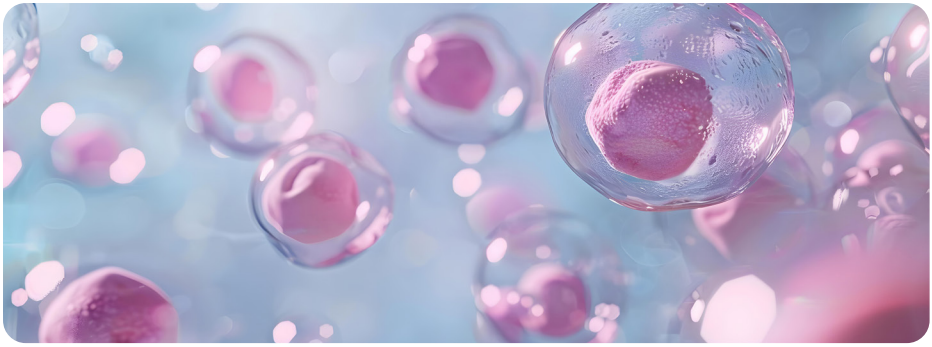
What to expect

Preimplantation genetic testing (PGT) is a process that is part of assisted reproduction treatment, especially in vitro fertilization (IVF). It involves several steps:

- **IVF and embryo creation:** The woman undergoes hormonal stimulation to produce more eggs, which are collected and fertilised with sperm in the laboratory. The resulting embryos are cultured for several days, usually to the blastocyst stage (5–6 days after fertilisation).

- **Embryo biopsy:** When the embryos reach the blastocyst stage, a few cells are taken from the outer layer (trophectoderm) of the embryo. This process is known as an embryo biopsy and is performed with minimal risk of damage to the embryo.
- **Genetic analysis:** The collected cells are genetically tested for chromosomal abnormalities (PGT-A), genetic mutations associated with specific hereditary diseases (PGT-M) and structural chromosome rearrangements (PGT-SR).
- **Embryo selection:** Embryos that do not have identified genetic abnormalities based on the results of the genetic analysis are selected. These embryos are considered suitable for transfer to the uterus.
- **Embryo transfer:** A selected healthy embryo is transferred to the woman's uterus with the goal of pregnancy. The transfer can be done within the same IVF cycle, or after freezing and thawing the embryo in a later cycle.

The entire preimplantation genetic testing process is carefully managed and performed by doctors and reproductive medicine and genetics specialists. Each step is important for achieving successful outcomes and minimise risks for the future parents.



Types of tests

- PGT-A (Preimplantation genetic testing for aneuploidy)

  Purpose: Detects chromosome number abnormalities (aneuploidy) such as trisomies (e.g. Down syndrome) or monosomies.

  Benefits: Helps select embryos with a normal number of chromosomes, increasing the likelihood of a successful pregnancy and reducing the risk of miscarriage.

- PGT-M (Preimplantation genetic testing for monogenic disorders)

  Purpose: Identifies specific genetic mutations associated with monogenic (single-gene) inherited disorders such as cystic fibrosis, Huntington's disease and Tay-Sachs disease.

  Benefits: Helps select embryos that do not carry a particular genetic mutation, which reduces the risk of having a child with the given genetic disorder.

- PGT-SR (Pre-implantation genetic testing for structural rearrangements)

  Purpose: Detects structural rearrangements of chromosomes such as translocations, inversions or deletions.

  Benefits: Helps select embryos without structural chromosomal abnormalities, increasing the chances of a successful pregnancy and a healthy baby.

Benefits of PGT

- **Higher success rates of IVF:** Choosing genetically healthy embryos increases the chances of a successful pregnancy.
- **Reduced risk of miscarriage:** There is a lower risk of miscarriage with embryos without chromosomal abnormalities.
- **Prevention of genetic disorders:** Makes sure the children won't have specific inherited disorders.
- **Informed decision-making:** Gives couples important information that assists them in family planning.

Preimplantation genetic testing (PGT) is a valuable tool for couples struggling with infertility or at risk of genetic disorders. By identifying genetically healthy embryos for transfer, it greatly increases the chances of a successful pregnancy and the birth of a healthy baby. Consultations with a doctor and embryologist are key to understanding the procedure and test results.

Are you ready to take the first step?

If you are ready, book your first appointment at our clinic.
Our coordinators can offer you in-person, phone or video appointments.
We are available during normal business hours if you request, at a time that suits your preferences.

Book a consultation with one of our leading fertility specialists

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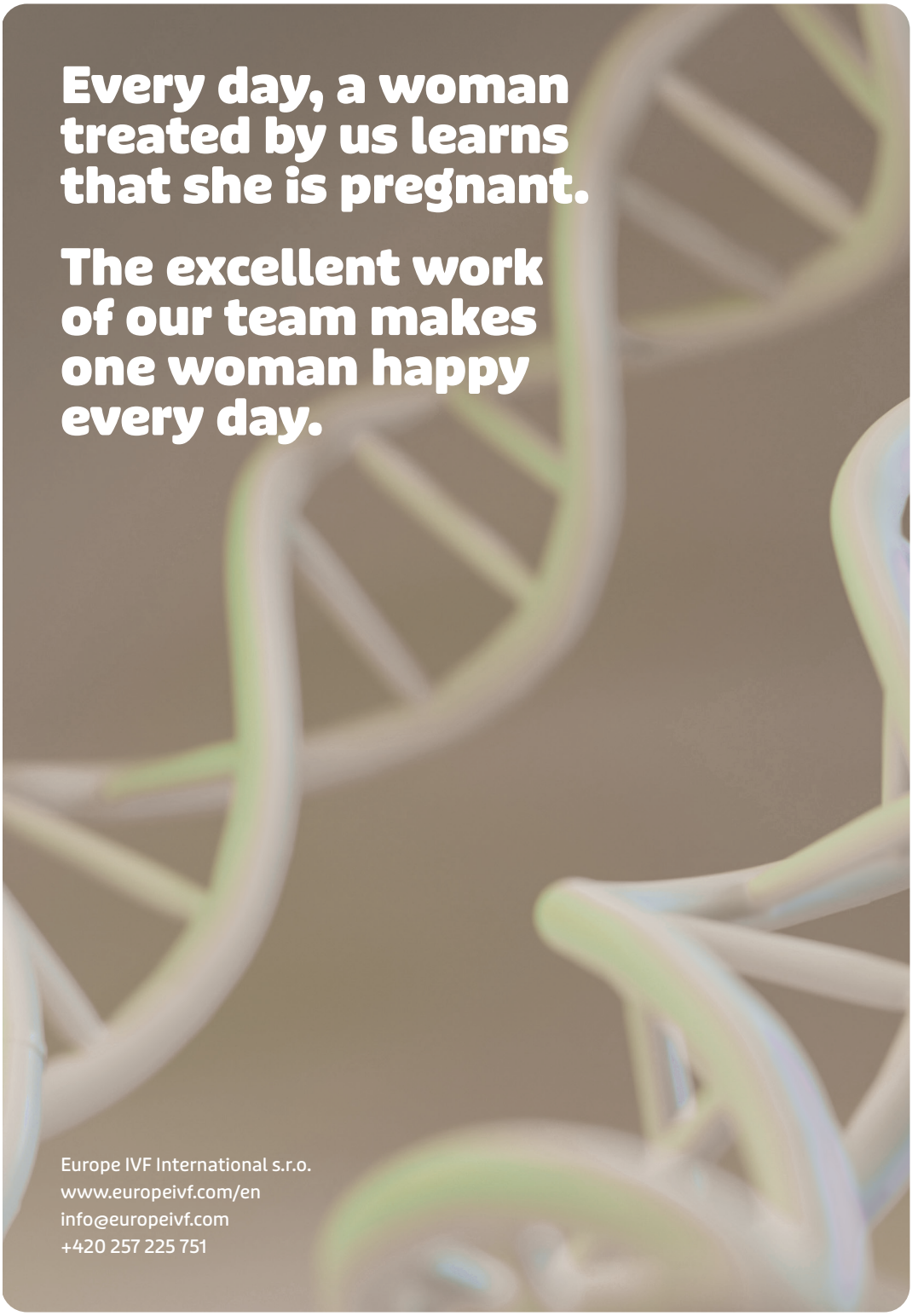
If you have any questions, ask us.

Europe IVF

You dream. We care.


Every day, a woman treated by us learns that she is pregnant.
The excellent work of our team makes one woman happy every day.
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