PGD

Preimplantation Genetic Diagnosis for Single Gene Disorders

BOSTONIVF

IVF New England
A BOSTONIVF PARTNER

PGD is testing that can prevent the transmission of single gene disorders

Our collaborating laboratories have extensive experience and have analyzed more than 250 different single gene disorders.

What is PGD?

PGD is the early genetic diagnosis of an IVF embryo prior to transfer.

Embryos can be assessed for specific disorders which allows for selective transfer to the woman's uterus.

Indications for performing PGD

PGD is indicated for couples at higher risk of having a child with a specific single gene disorder. Examples include Cystic Fibrosis, Fragile-X syndrome, Spinal Muscular Atrophy, Huntington's disease and many others.

A panel of the more common diseases has sometimes already been developed for which a PGD test can be performed. PGD can now be performed for most known single gene disorders.

The option of performing PGD and PGS on the same embryo biopsy is also available and can improve healthy live birth rates.

5 Steps to perform PGD*

1 

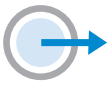
Consultation

Send in the genetic report and the consent form.
Answer typically within 3 business days from a genetic counselor.

2 

Pre-PGD Set-Up

Send us the requested DNA samples and Test Requisition Form.
Set-up complete typically within 3 weeks for common disorders & 6 weeks for less common disorders.

3 

IVF and Embryo Biopsy

Extended embryo culture. Day 5 embryo biopsy plus vitrification. Biopsy shipped to IVIGEN.

4 

PGD

Embryo results typically within 2 weeks.

5 

Embryo Transfer

* ICSI is required

Why PGD?

- Robust and reliable diagnosis
- Complimentary counseling from board certified genetic counselors
- >98% of embryos tested receive a result
- Allows the transfer of unaffected embryos

Transferring embryos without major chromosome abnormalities increases pregnancy success and promotes the birth of healthy babies.

*If you decide to add PGS to your treatment plan, please be sure to discuss with you Financial Coordinator, as there are often times additional out of pocket costs involved.

EXAMPLES OF SINGLE GENE DISORDERS

Autosomal dominant polycystic kidney disease
Becker's muscular dystrophy
Beta thalassemia
Cystic fibrosis
Charcot-Marie-Tooth disease type 1A
Duchenne muscular dystrophy
Familial amyloid polyneuropathy
Fragile X syndrome
Hemophilia A (F8)
Hemophilia B (F9)
Huntington's disease
Multiple endocrine neoplasia, type 2A
Myotonic dystrophy (Steinert's disease)
RhD incompatibility
Spinal muscular atrophy

PGS

Preimplantation Genetic Screening for Chromosomal Abnormalities

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Increase the chance of a successful pregnancy and a healthy baby

Preimplantation Genetic Screening (PGS) allows indentification of chromosomally normal embryos.

While aneuploidy is more common as you get older, it can occur at any age. That's why PGS can be an important part of IVF for women of all ages.

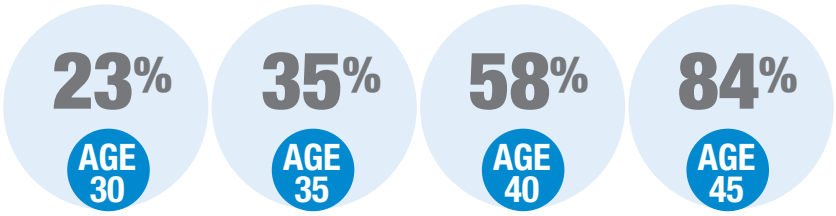
What is PGS?

PGS detects chromosomal abnormalities prior to embryo transfer to enable informed decisions.

Transferring embryos without major chromosome abnormalities increases pregnancy success and promotes the birth of healthy babies.

Maternal Age and Aneuploidy

Percentage of embryos with an abnormal number of chromosomes



Adapted from Franasiak JM et al. *Fertil Steril.* 2014;101(3):656-653.

Why PGS?

- Robust and reliable test performed by array technology (aCGH) or Next Generation Sequencing (NGS)
- 98% accuracy in detecting major chromosome abnormalities
- Can accommodate both fresh and frozen embryo transfers
- Results obtained on 98% of samples received
- Complimentary consultations with board certified genetic counselors - available by request

*If you decide to add PGS to your treatment plan, please be sure to discuss with you Financial Coordinator, as there are often times additional out of pocket costs involved.

The TRANSFER of a chromosomally normal embryo significantly improves your chance of pregnancy at any age

INDICATIONS FOR PGS

1. **Advanced maternal age** » The risk of chromosomal abnormalities, such as Down Syndrome, in embryos increases with the age of the mother.
2. **Recurrent miscarriage** » Approximately 50% of miscarriages occur due to chromosomal abnormalities. In the case of miscarriages, 68% of embryos are chromosomally abnormal. Embryos that are chromosomally normal are more likely to result in live birth.
3. **Chromosome abnormalities** » While a few chromosomal abnormalities are compatible with life, these babies may be born with birth defects and cognitive impairment. PGS screening detects major chromosome abnormalities allowing informed decisions about reproductive care.
4. **Implantation failure** » Some chromosomal abnormalities impair the embryo's ability to implant into the uterus. Embryos that are chromosomally normal have a better chance of successful implantation.
5. **Male factor infertility** » Male factor infertility (including abnormal quantity and quality of sperm) can increase the risk of chromosomal abnormalities in the embryo. PGS can aid in identifying chromosomally normal embryos.