PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

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What is PGD?

Preimplantation Genetic Diagnosis (PGD) is a procedure which aims to identify genetic defects in embryos in order to select the "healthy" ones.

With this procedure, embryos created during a IVF cycle undergo biopsy, which allows to analyze the DNA of each embryo, identify possible genetic defects and select those embryos which are genetically "healthy". Finally, the embryos free of genetic problems are transferred into the uterus of the future mother to achieve the desired pregnancy.

This procedure provides great security to the intended parents by minimizing the risk of genetic diseases.

PGD Tests

There are two types of genetic studies depending on the type of abnormality:

Preimplantation Genetic Diagnosis or Monogenic PGD allows for the early detection of serious monogenic mutations in parents, which can be inherited by their offspring. This procedure is indicated when both or one of the parents are carriers of genetic diseases such as:

- Huntington's disease
- Cystic fibrosis
- Spinal muscular atrophy
- Fragile-X syndrome

among many others, since there are hundreds of single gen disorders.

Preimplantation Genetic Screening (PGS), also known as Screening for Fetal Aneuploidy, PGD-A or chromosomal PGD allows to identify abnormalities in chromosome number and structure. This procedure is indicated in the following cases:

- Advanced maternal age. It has been proved that the older the mother, the higher the risk of chromosomal disorders. This risk increases in women age 35 and especially over 38.
- Recurrent miscarriages
- Implantation failures
- Karyotype defects present in any parent
- Previous aneuploid pregnancy
- Abnormal sperm FISH

How can PGD help me? In what cases should it be performed?

There are two cases in which PGD tests should be done:

- PGD for single gen diseases should be performed if any of the parents has been diagnosed with a genetic disease or is an asymptomatic carrier of a mutation responsible for a genetic disease in the family. With this procedure, the risk of transmitting an inherited disease to offspring is eliminated.

- Chromosomal PGD should be performed if any parent has an abnormal karyotype or sperm FISH. It is also recommended for women who have experienced recurrent pregnancy loss -caused by genetic disorders in 90% of cases.

  Chromosomal PGD can be performed in every IVF cycle so as to increase the chances of success and reduce miscarriage risk, implantation failures and disorders caused by chomosome diseases such as Down, Edwar and Patau syndroms. However, it is important to mention that even if this PGD test allows to identify healthy embryos with a good potential for implantation and development, it does not increase the chances of success of an IVF treatment. A successful pregnancy will mostly depend on the gamete quality and the absence of genetic abnormalities.