

# Genetic analysis and PGS



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#### SCREENING OF HEREDITARY DISEASE

Nowadays, couples suffering from any hereditary disease and wishing to start a family can have healthy children thanks to In Vitro Fertilization and Pre-implantation Genetic Diagnosis.

For those patients with family history of genetic disease or suffering from pathologies transmittable to their descendants, it's possible to study whether there is a genetic cause before starting any assisted reproduction treatment.





#### **INFERTILITY STUDY**

There are genetic diseases causing infertility to those who suffer from them and that can also be transmitted to children. Some of the most known diseases such as X-fragile or gonadal dysgenesis can be studied and be eradicated in a family by selecting the embryos free from the disease through what is known as Pre-implantation Genetic Screening techniques.

In those cases where there is no concrete disease but previous clinical history recommends it, the analysis of the couple's chromosomes is performed to check whether they are normal as well as the analysis of sperm DNA alterations that can cause abnormalities, miscarriages or other chromosome diseases. Thanks to Pre-implantation Genetic Screening we can study the embryo's chromosomes and select the ones without alteration before transferring them into the uterus of the future mother.



#### RECURRENT MISCARRIAGES ASSESSMENT

Miscarriages can be due to hormone alterations, infectious agents, anatomic problems or immune system's rejections, but the majority happens for a genetic reason. Several times recurrent implantation failures are considered early miscarriages and are treated in the same way.

In order to know if there is a genetic cause, there are different tests to study the implantation process in the woman and to detect chromosome alterations in the embryo before its transfer into the uterus.

Thanks to all this it's possible to determine the reason of the miscarriages in a 60% of the cases. .



# WHAT IS PRE-IMPLANTATION GENETIC SCREENING?

Pre-implantation Genetic Screening (PGS) is a very early diagnostic technique to detect abnormalities in the genes or chromosomes of the embryos obtained by In Vitro fertilization, before they are transferred into the patient's uterus.

One or two cells of the embryos are analyzed to obtain the diagnosis. By performing the diagnosis before the embryo's implantation, the advantage is that we are minimizing the risk of having to interrupt the gestation once that the patient is already pregnant.

PGS is indicated not only in case of monogenic diseases, but also in cases of abnormalities in number and structure of chromosomes.

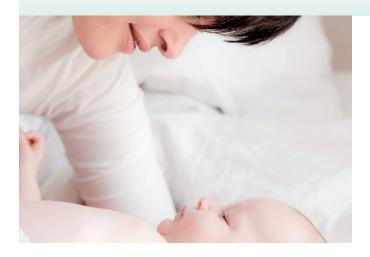


# SCREENING OF ALL THE EMBRYO'S CHROMOSOMES: CGH ARRAYS

Compared genomic hybridization (CGH) is a state-of-the-art technique allowing to study the 24 chromosomes of an embryo (22 + X,Y) with a microchip of DNA (array). Up until now other more conventional techniques were used, which analyzed at most 9 or 12 chromosomes of the 23 pairs.

Thanks to this new technology it's possible to detect up until 50% more abnormalities in the number of chromosomes and it's easier to achieve pregnancy in the clinically indicated cases.

Patients who have suffered from many miscarriages, couples in advanced age or with previous history that suggest it, might be favoured by the use of these techniques.



# SEX DETERMINATION

In couples with risk of transmitting X-linked diseases, we analyze the sex chromosomes to determine the sex of the embryos, being able to select the ones suitable to be transferred.

# HLA TYPING

For the past few years in Spain it has been possible for couples with children affected by a severe disease to request authorization to health authorities to select, by In Vitro fertilization and genetic selection's techniques, histocompatible embryos for future transplants. Nowadays we are provided with the greatest set of genetic markers of the market to find the maximum level of compatibility and to maximize chances of embryo transfer in each case.



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