

Preimplantational genetic diagnosis

Is it possible to perform a genetic analysis of the embryos before they are transferred to the uterus?

Yes. It is possible to analyze genetically the embryos prior their transfer to the uterus. This technique is called Preimplantational Genetic Diagnosis or Embryo Genetic Analysis.

Does 'Embryo Genetic Analysis' increase pregnancy rate?

Yes, because it avoids the transfer of embryos that contain an abnormal number of chromosomes (aneuploid embryos). Using this procedure, only chromosomally normal embryos with a good morphology are transferred.

Does 'Embryo genetic analysis' decrease the rate of spontaneous miscarriage?

Yes. 'Embryo Genetic Analysis' can significantly reduce spontaneous miscarriage because most of them are due to genetic anomalies.

Does 'Embryo Genetic Analysis' prevent the birth of children with chromosomal abnormalities?

Yes. It can avoid the birth of children with chromosomal abnormalities such as trisomy 21 or Down's syndrome.

Can all the chromosomes be analyzed?

No. Human cells have 23 pairs of chromosomes, some of them are so important that their alteration impair embryo development. Chromosomes 13,16,18,21,22, X and Y are the most frequently affected. At Institut Marquès, 9 pairs of chromosomes are normally analyzed. This allows to study up to 85% of all chromosomal abnormalities.

For patients that are carriers of a genetic disease, embryo genetic analysis is directed to study the specific abnormal gene.

How is embryo genetic análisis performed?

In vitro fertilization-derived embryos are cultured until Day 3 of development at which stage embryos have 6 to 8 cells or blastomeres. Then, a hole is made in the zona pellucida (the layer that covers the embryo) and one cell is aspirated using a special micropipette (biopsy procedure). The aspirated cell is processed for genetic analysis while the embryo is maintained in culture. Genetically normal embryos are transferred to the uterus the next day.

Could this technique damage the embryo?

The biopsy procedure is highly sophisticated technique that when performed by experienced embryologists should not negatively affect embryo viability.

Is it possible to prevent a genetic disease using 'Preimplantational Genetic Diagnosis'?

Yes. The group of patients that benefit most from Preimplantational Genetic Diagnosis are couples with a history of a genetic disease. This is so because it allows to transfer normal embryos free of the genetic disease and, therefore, pregnancy termination is avoided. There are two types of monogenic disorders: **autosomal disorders** such as Cystic fibrosis, Myotonic Dystrophy, Huntington's disease or **sex-linked disorders** as Fragile X Syndrome, Duchenne's Muscular Dystrophy or Hemophilia.

Can patients with a previous spontaneous miscarriage benefit from 'Preimplantational Genetic Diagnosis'?

Yes. They can prevent further pregnancy loss by performing Preimplantational Genetic Diagnosis. It is of paramount importance in the management of infertility couples with an embryo factor.

Can any couple benefit from 'Preimplantational Genetic Diagnosis'?

Yes. Taking into account all the benefits mentioned above, more and more IVF patients choose to perform Preimplantational Genetic Diagnosis. Specially when the future mother is over 37 years of age or had a previous IVF failure.