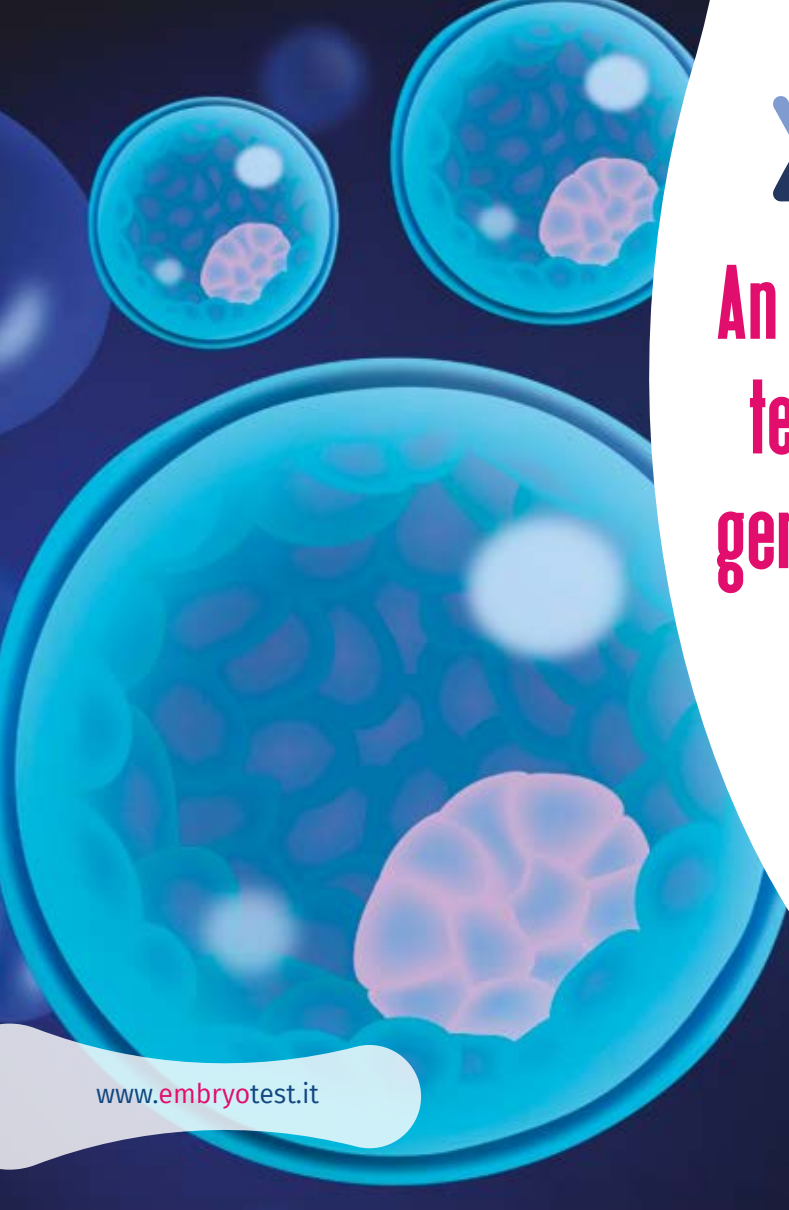


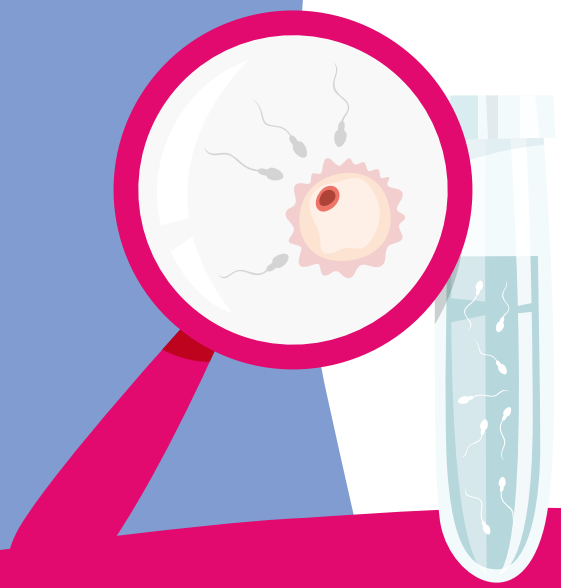
GENOMICA
Next Generation Genetics

An advanced preimplantation test that screens for severe genetic disorders in embryos

Inherited genetic disorders
De novo genetic disorders
Chromosomal abnormalities

EMBRYOTEST
SCREENING OF GENETIC DISORDERS IN EMBRYOS

www.embryotest.it



EMBRYOTEST

THE NEXT LEVEL IN PREIMPLANTATION GENETIC TESTING

EMBRYOTEST is the most advanced preimplantation genetic test that, using groundbreaking technologies, screens multiple genes to detect mutations causing severe inherited and *de novo* genetic disorders in embryos.

EMBRYOTEST is a complement to Genomica's market-leading PGTADVANCE test, which screens embryos for aneuploidies and segmental chromosome imbalances.

Inherited genetic disorders are caused by mutations carried by the couple. These mutations, detectable in the parents with pre-conception carrier screening tests, can be transmitted to the progeny at conception, and therefore become detectable in preimplantation embryos.

***De novo* genetic disorders** are caused by mutations, named *de novo* (a gene mutation that is not inherited), that arise randomly in the embryo. These genetic diseases often occur in the absence of a family history of the condition and cannot be detected by standard carrier screening, as *de novo* mutations are not detectable in the parents. *De novo* genetic disorders can cause serious conditions such as skeletal dysplasias, congenital heart defects, multiple congenital malformation syndromes, neurodevelopmental disorders, autism, epilepsy and/or intellectual disability.

EMBRYOTEST

A CUTTING-EDGE GENETIC TEST THAT OFFERS A FULL PANEL OF SCREENING OPTIONS

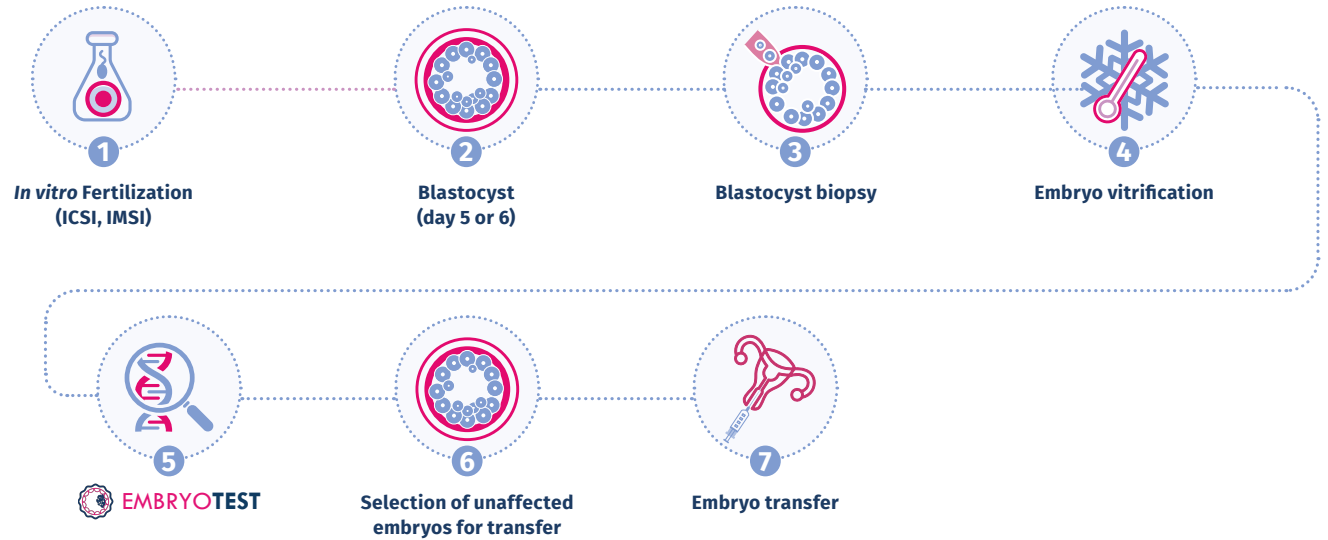
SCREENING LEVEL	GENES	DISEASES INVESTIGATED
EMBRYOTEST F Focus	**61**	It screens for **26** common **inherited genetic disorders** and for **53 severe genetic disorders** caused by ***de novo*** mutations.
EMBRYOTEST X Exome	**>4000**	**Clinical exome** sequencing, including **5000+** severe **inherited** and ***de novo* genetic disorders.**
EMBRYOTEST C Complete	**>4000** + Chromosomal Aneuploidies	It offers the most comprehensive level of screening, investigating **5000+** of the most common **inherited** and ***de novo* genetic diseases**, as well as **aneuploidies** and **structural chromosomal abnormalities.**

EMBRYOTEST also identifies genetic diseases associated with **advanced paternal age** (eg. Achondroplasia, Pfeiffer's syndrome, Apert's syndrome, Crouzon's syndrome, Osteogenesis Imperfecta, etc.), caused by DNA mutations that arise during the spermatogenesis process, providing couples with advanced age the opportunity to use a more comprehensive screening test.[1] **EMBRYOTEST** also allows identification in embryos of genetic conditions whose incidence is independent of maternal age. Genetic diseases detectable with this test have a cumulative incidence ~1/600, and ~1/300 for disorders determining developmental delay.[2]

1. Kong A, et al.: Rate of *de novo* mutations and the importance of father's age to disease risk. Nature 2012, 488:471-475.
2. McRae J, et aL Prevalence and architecture of *de novo* mutations in developmental disorders. Nature 542. "33--438

EMBRYOTEST

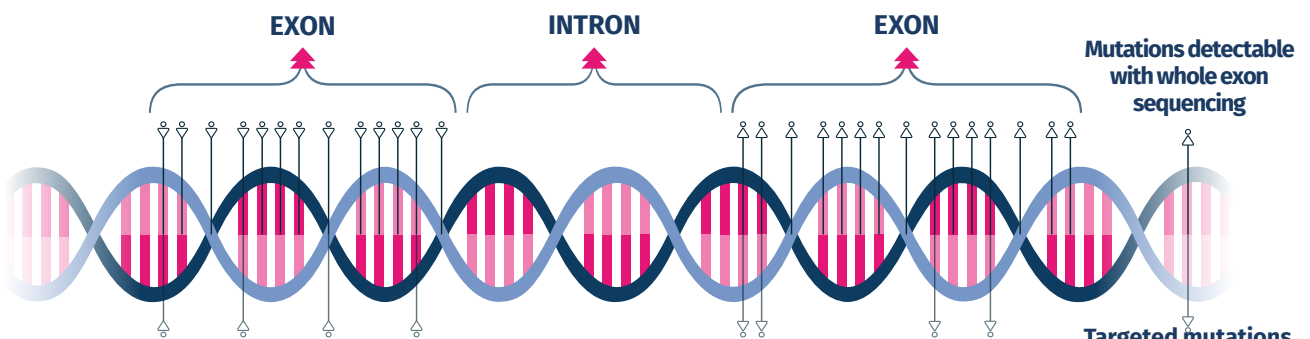
THE TESTING PROCESS



EMBRYOTEST usually requires that the couple undergoes to an *in vitro* fertilization (IVF) treatment. 1 This involves hormonal treatments that allow the collection of multiple eggs from the mother. The eggs are then fertilized using the father's seminal fluid and the resulting embryos are transferred to an incubator. Once they have reached the blastocyst stage 2 embryos will be subjected to a trophectoderm biopsy 3 for the recovery of the embryonic cells and subsequent cryopreservation 4 awaiting the test results. Then the DNA of each embryo will be analyzed with **EMBRYOTEST** 5 The unaffected embryos will be selected 6 to be transferred to the womb 7

EMBRYOTEST

A GROUNDBREAKING TECHNOLOGY ALLOWING FOR A SCREENING TEST THAT IS REVOLUTIONARY



A groundbreaking technology coupled with an advanced bioinformatic analysis allows for detection of DNA mutation in embryos causing serious genetic disorders, providing the most comprehensive information available from a preimplantation genetic test to date. **EMBRYOTEST** uses a state-of-the-art technological process, named **Next Generation Sequencing (NGS)**, carried out performing full-exon sequencing of all the genes included in the panel, which allows a more comprehensive assessment of each gene and related diseases

EMBRYOTEST

TEST RESULTS

POSITIVE: Pathogenic mutation(s) detected:
*this test result indicates that one or more disease causing mutations have been detected in the targeted genes screened. In such cases, embryo is considered to be at **high risk.***

NEGATIVE: No pathogenic mutation(s) detected:
*this test result indicates that no disease causing mutations have been detected in the targeted genes screened. In this case, the embryo is considered to be at low risk for the genetic diseases investigated. **A low risk** test result greatly reduces the chances that the embryo is affected by a genetic disorder but it cannot guarantee a healthy baby.*

Turnaround time

15 days

EMBRYOTEST

INDICATION FOR TESTING

- Couples with a family history of a genetic disease;
- Couple wishing to lower the risk for genetic disorders in embryos;
- Couples requiring gamete donation, in order to minimize the reproductive risk;
- Patients who are pursuing pregnancy with assisted reproductive technologies combined with PGT.

GENOMICA
Next Generation Genetics

Advanced molecular diagnostics solutions using state-of-the art technologies

GENOMICA is recognized as one of the most advanced molecular diagnostics laboratory in Europe, both for the state-of-the-art instruments and technologies, as well as for its high quality standards. With a comprehensive portfolio of over 10.000 genetic tests, GENOMICA is able to satisfy increasingly specialised requests in the field of molecular genetics, providing physicians and their patients with innovative and highly specialised diagnostic solutions for any clinical need.

- **Test performed in Italy** (Rome or Milan)
- **Professionals with 20+ years experience** in the field of genetics and molecular diagnostics
- Laboratories with **groundbreaking technologies** and high quality standards
- Over **100.000** genetic tests/year
- Fast TAT **15 days**
- **Worldwide genetic testing provider**
- **Dedicated R&D team**
- **International partnerships**
- **Personalized genetic counseling** with genetic counselors experts in discussing genetic test results and familial risks

LABORATORIES

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Embryotest M Eng rev.00