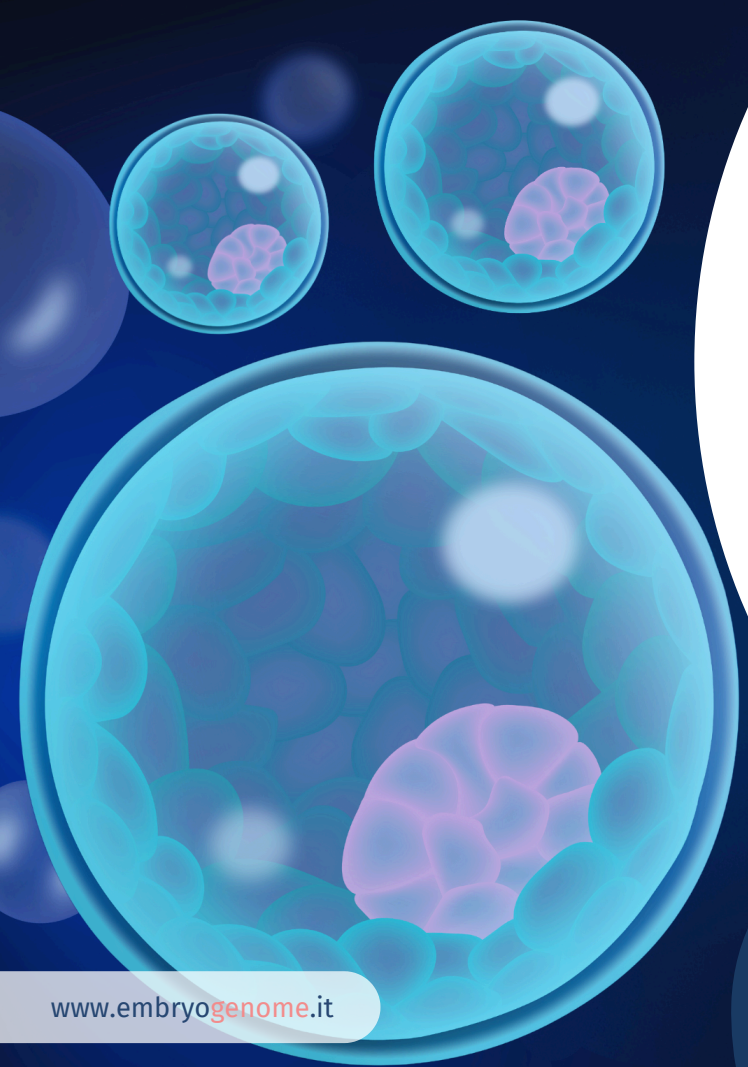

www.embryogenome.it

GENOMICA
Next Generation Genetics

The first preimplantation genetic test that screens the whole exome of IVF embryos

- Inherited and *de novo* genetic disorders
- Aneuploidies and structural chromosomal abnormalities
- Chromosomal mosaicism
- Microdeletion/microduplication syndromes
- Triploidy/Haploidy and uniparental disomy (UPD)
- Copy number variants (CNVs)

EMBRYOGENOME
GENOMIC PREIMPLANTATION GENETIC TESTING

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THE NEXT LEVEL IN PREIMPLANTATION GENETIC TESTING

EmbryoGenome is an advanced preimplantation genetic test that screens the **whole coding genome** (**exome**) of IVF embryos, enabling the simultaneous detection of:

- Mutations causing **~7,000** clinically recognized **severe genetic disorders**, including **inherited** and ***de novo*** conditions;
- Numerical chromosomal abnormalities (**Aneuploidy**);
- Chromosomal **mosaicism**
- **Triploidy/Haploidy** and **uniparental disomy** (**UPD**);
- Segmental chromosomal imbalances (**deletions/duplications**);
- **130+ Microdeletion/microduplication** syndromes;
- **Copy number variants** (**CNVs**).

Aneuploidies and structural chromosomal imbalances

+

~7.000 clinically known severe genetic diseases, both inherited and *de novo*

+

Deletions, duplications, and 130+ microdeletion/ microduplication syndromes

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REDEFINING EMBRYO SCREENING

EmbryoGenome represents a groundbreaking advancement in embryo screening, surpassing the limitations of traditional preimplantation genetic testing for aneuploidy (PGT-A), which focuses solely on chromosomal analysis. This state-of-the-art test integrates PGT-A, PGT-M, and PGT-SR into a single, comprehensive screening platform. By using whole-exome sequencing (WES), **EmbryoGenome** provides unparalleled insight into embryo genetics.

Advantages over traditional PGT tests

EmbryoGenome offers some advantages over traditional preimplantation genetic testing (PGT):

PGT-A

- **1000x higher resolution** than traditional PGT-A tests;
- Detect **triploidy** and **molar embryos** (complete uniparental isodisomy) that other labs might miss;
- Detect **microdeletions** + **microduplications** screened for during pregnancy but missed in traditional PGT;
- Plus all traditional PGT-A features

PGT-M

- No customized PGT workup is necessary, resulting in **faster turnaround time;**
- **Ideal for:**
- - challenging PGT-M cases that may have been denied by other companies;
  - patients with a previous pregnancy/child with a *de novo* variant;
- Can screen for hundreds of monogenic diseases simultaneously.

PGT-SR

- **Improved resolution** compared to traditional platforms used for PGT-SR;
- Increased chance of case acceptance;
- Patients can additionally **screen for hundreds of monogenic conditions simultaneously.**
- Includes all the features of the traditional PGT-SR

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- **Screen for hundreds of genetic conditions simultaneously**;
- **Detect *de novo* mutations**, causing conditions such as skeletal dysplasias, multiple congenital anomalies, autism, epilepsy, and/or intellectual disabilities.
- Identifies genetic diseases associated with a**dvanced paternal age**;
- Plus all traditional PGT-M, PGT-A, PGT-SR features

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UNPARALLELED INSIGHTS COMPARED TO STANDARD PGT

FEATURES	PGT-A	PGT-M	PGT-SR	Embryo Genome
Detects extra or missing whole chromosomes (Aneuploidies)	✓		✓	✓
Detects extra or missing chromosome segments (Deletions/Duplications**)**	✓		✓	✓
Mosaicism	✓		✓	✓
Triploidy/Haploidy				✓
Uniparental disomy (UPD)				✓
Microdeletion/microduplication syndromes				✓
Copy Number Variants (CNVs)				✓
Inherited chromosomal rearrangements			✓	✓
Specific inherited genetic disorders (known genetic risk, ie. parents are aware to be carrier of a specific monogenic disorder)		✓		✓
Inherited genetic disorders (unknown genetic risk, ie. parents are unaware carriers of mutations associated with recessive/X-linked disorders)				✓
***De novo* genetic disorders** (caused by mutations that onset randomly in the embryo and are not inherited)				✓

Standard PGT screening may miss unexpected genetic diseases. **EmbryoGenome** provides advanced and comprehensive analysis to detect conditions that current tests may overlook.

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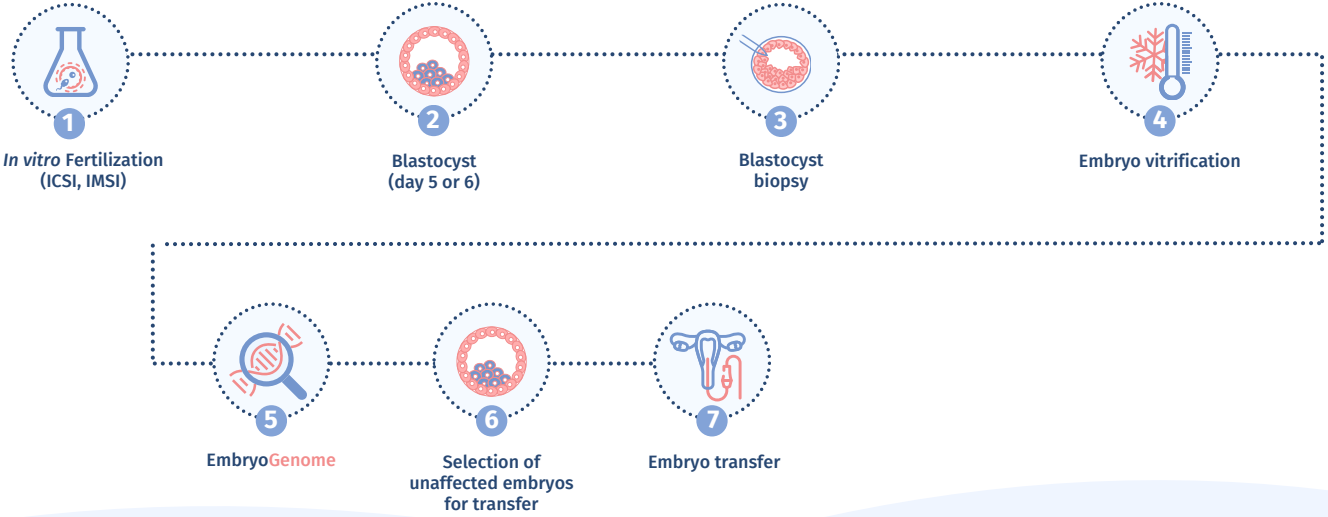
THE TESTING PROCESS



EmbryoGenome usually requires couples to undergo *in vitro* fertilization (IVF) treatment. 1 This process begins with hormonal stimulation to enable the collection of multiple eggs from the mother. The eggs are then fertilized using the father's sperm, and the resulting embryos are cultured in an incubator. Once the embryos reach the blastocyst stage 2 a trophectoderm biopsy 3 is performed to retrieve embryonic cells.The embryos are then cryopreserved 4 while awaiting test results. **EmbryoGenome** analyzes the DNA of each embryo, 5 identifying those unaffected by genetic or chromosomal abnormalities. 6 These selected embryos can then be transferred to the womb 7 for implantation.

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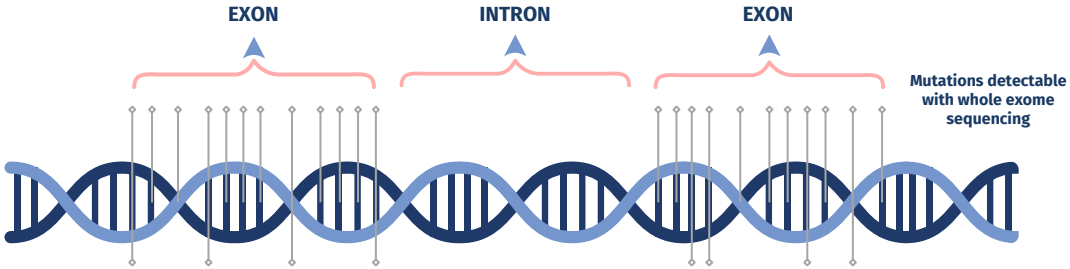
CUTTING-EDGE TECHNOLOGY BEHIND



The groundbreaking **deep Whole Exome Sequencing** technology, combined with advanced bioinformatic analysis powered by a proprietary algorithm, enables the precise detection of DNA mutations in embryos causing hundreds of severe inherited or *de novo* genetic diseases. In addition to genetic mutation analysis, **EmbryoGenome** provides a comprehensive evaluation of the embryonic karyotype. This includes detecting aneuploidies, segmental chromosomal imbalances, and microdeletion/microduplication syndromes—all in a single test. Leveraging state-of-the-art Next Generation Sequencing (NGS) technology, **EmbryoGenome** screens over 20.000 genes through whole exome sequencing. This cutting edge methodology delivers the most comprehensive insights currently available in PGT.

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WHOLE EXOME SEQUENCING: REVOLUTIONIZING PGT

- **Whole exome sequencing** (**WES**) has become a critical tool for diagnosing severe genetic conditions caused by single-gene mutations.
- This advancement is widely used to diagnose children and adults with congenital anomalies or developmental delay or intellectual disability.[1]
- WES is also recommended for pregnancies affected by fetal structural anomalies.[2]
- With an **incremental diagnostic rate** of **~30%**[3], WES offers transformative insights.
- Now, WES can be applied at the embryo stage. Trophectoderm biopsies are screened for **hundreds of monogenic conditions**, enabling the identification and prevention of genetic diseases before embryo transfer.
- This cutting-edge approach represents the most comprehensive preimplantation genetic testing available today.

1. Manickam et al. Genetics in Medicine (2021) 23:2029–2037; 2. Van den Veyver et al. Prenat Diagn (2022);42(6):796–803 3. Mellis et al. Prenat Diagn. 2022;42(6):662–685
3. Mellis et al. Prenat Diagn. 2022;42(6):662–685;

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CLINICALLY VALIDATED APPROACH

GENOMICA's researchers conducted an extensive validation for whole exome sequencing-based PGT (PGT-WES). The validation included a prospective, double-blind study comparing PGT-WES with conventional PGT methods. **EmbryoGenome** has demonstrated high accuracy in detecting inherited and *de novo* pathogenic variants, aneuploidy, and microdeletion/microduplication syndromes on standard trophectoderm biopsies of preimplantation embryos, providing unparalleled insights into embryo genetics.

Inherited genetic disorders

The validation results showed a **high level of concordance** with standard PGT methods. Outcomes for PGT-M were **fully consistent** across all embryos tested, reliably detecting target mutations and uncovering additional genetic variations associated with monogenic disorders—even in embryos from unrecognized carrier parents.

De novo genetic disorders

EmbryoGenome accurately detected *de novo* pathogenic mutations linked to severe early-onset genetic conditions.

Chromosomal abnormalities

The CNV detection capability of **EmbryoGenome** matched the performance of standalone PGT-A assays, accurately identifying both whole-chromosome and segmental aneuploidies. Ploidy status was **100% concordant** across all embryos screened for PGT-A. Pathogenic CNVs **>200 kb** associated with parental microdeletion/microduplication syndromes were reliably detected, owing to the **enhanced resolution** of PGT-WES.

EmbryoGenome sets a new standard in embryo screening, combining accuracy, comprehensiveness, and innovation to deliver the most advanced insights into embryo genetics. By integrating cutting-edge technology with rigorous validation, it offers families the highest confidence in their reproductive choices.

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KEY ADVANTAGES

Unmatched Accuracy

With **over 99% accuracy**, **EmbryoGenome** significantly outperforms conventional PGT methods, providing the highest ***detection rates*** for genetic and chromosomal abnormalities.

Comprehensive Screening

EmbryoGenome combines PGT-A, PGT-M, and PGT-SR with the ability to detect **hundreds of severe monogenic disorders**, providing an unprecedented level of detail on the embryo's genetic profile.

Advanced Technology

EmbryoGenome uses cutting-edge technologies and sophisticated bioinformatics analyses.

Advanced Detection

Unlike standard PGT, which may overlook unexpected genetic diseases, **EmbryoGenome** identifies conditions that other tests often miss.

EmbryoGenome helps families maximize their chance for a successful pregnancy and healthy baby by screening embryos using whole exome sequencing.

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INDICATION FOR TESTING

While **EmbryoGenome** is beneficial for all patients, certain groups may find it particularly advantageous:

- **Couples with a family history of genetic disorder or chromosomal abnormality;**
- **Couples seeking to minimize reproductive risks;**
- **Patients using donor gametes or embryos;**
- **Patients Using Assisted Reproductive Technologies (ART) combined with preimplantation genetic testing (PGT)**
- **Patients concerned about *de novo* variants:** This includes individuals of advanced paternal age or those who have had a previous child affected by a *de novo* genetic variant.

EMBRYOGENOME

GENOMIC PREIMPLANTATION GENETIC TESTING

Turnaround time **15 days**

Collaborating with us

The collaboration process with GENOMICA is designed to be efficient and straightforward. After an initial orientation, clinicians can easily order **EmbryoGenome**, with all shipping logistics expertly managed. Using the same 6-8 cell trophectoderm biopsy, **EmbryoGenome** delivers comprehensive results, accompanied by individualized reporting and consultation to support clinical decision-making.

Quick and easy ordering

Order tests efficiently, without any added work. GENOMICA handles shipping logistics for all samples.

1. Request the shipping kit
2. Fill in the test requisition form
3. Collect samples
4. Ship the samples to Genomica
5. Receive results

Samples required

Blood samples
Couple + family members (if required)

+

Embryo biopsies

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**

Laboratories with **groundbreaking technologies** and **high quality standards**

Over 100.000 genetic tests/year

Dedicated **R&D team**

Personalized genetic counseling with genetic counselors experts in discussing genetic test results and familial risks

Professionals with 20+ years experience in the field of genetics and molecular diagnostics

EmbryoGenome M ENG rev.01

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Scan the QR Code and visit EmbryoGenome website