GENOMICA
Next Generation Genetics

Advanced strategies for detection of genetic disorders and chromosomal abnormalities in embryos

Comprehensive PGT solutions designed to support more informed embryo selection and optimize IVF outcomes.

PGTADVANCE

PGTADVANCE
NEXT GENERATION PGT

PREIMPLANTATION GENETIC TESTING (PGT)

A transformative approach to embryo genetic testing

Couples who are carriers of an inherited genetic disorder or a chromosomal abnormality face an increased reproductive risk, as these conditions may be transmitted to their children. In such circumstances, prenatal diagnosis by **chorionic villus sampling** (**CVS**) or **amniocentesis** may be used to determine whether the fetus is affected by a genetic or chromosomal condition.

While these procedures are now well established in clinical practice, the diagnosis of an affected fetus may place couples in the difficult position of having to decide whether to continue the pregnancy or proceed with therapeutic termination.

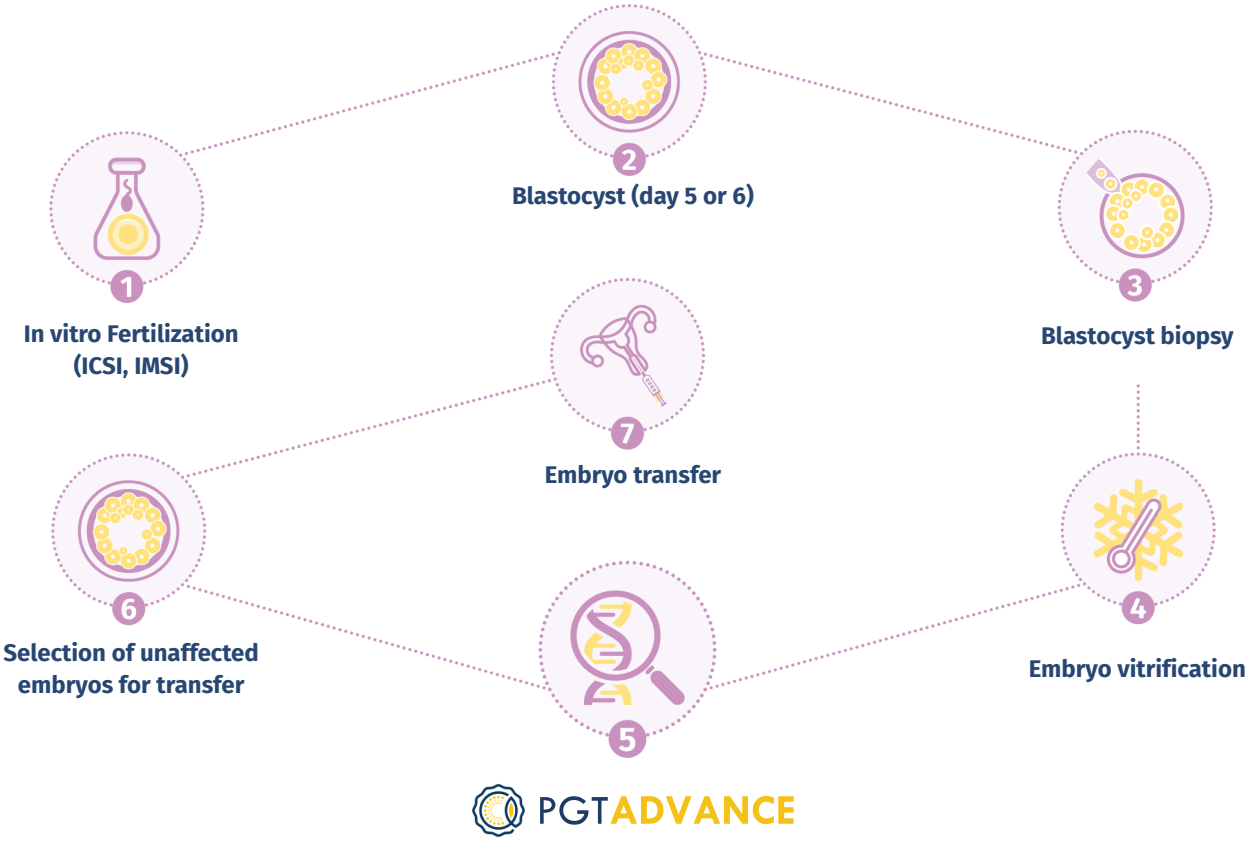
The evolution of **in vitro fertilization** (**IVF**) techniques, together with the possibility of obtaining embryonic cells suitable for genetic analysis, has significantly expanded the scope of reproductive genetics. It is now possible to move genetic diagnosis from the **post-implantation** stage to the **preimplantation** stage, allowing clinically relevant information to be obtained before pregnancy is established.

Within this context, **Preimplantation Genetic Testing** (**PGT**) represents a valuable procedure complementary to prenatal diagnosis. It enables the identification of genetic disorders and chromosomal abnormalities in embryos generated in vitro from couples at increased reproductive risk, at the earliest stages of development and prior to uterine transfer.

PGT therefore supports more informed reproductive choices and reduces the likelihood of facing therapeutic termination of pregnancy, an event that is often deeply burdensome from a psychological perspective and not always acceptable on ethical or moral grounds.

PGTADVANCE
NEXT GENERATION PGT

PGT PROCEDURE



PGT combines **in vitro fertilization** (**IVF**) techniques with the most advanced applications of molecular genetics, providing a sophisticated approach to embryo genetic assessment before transfer. The process begins with a medically assisted reproduction treatment cycle **(1)**, aimed at retrieving oocytes, which are then fertilized with paternal sperm to generate embryos in vitro.
Once the embryos reach the **blastocyst** stage **(2)**, they undergo **trophectoderm biopsy (3)**, a procedure that allows a small number of embryonic cells to be collected for genetic analysis. The embryos are subsequently **cryopreserved (4)** while awaiting the test results. The DNA from each embryo is then analyzed using the **PGT** approach **(5)**. Embryos found to be **unaffected** by the specific genetic condition under investigation are subsequently selected **(6)** for **uterine transfer (7)**, with the aim of achieving a pregnancy unaffected by the disorder being tested.

PGTADVANCE
NEXT GENERATION PGT

A COMPLETE PORTFOLIO OF ADVANCED PREIMPLANTATION TESTING SOLUTIONS

PGTADVANCE

Preimplantation genetic testing for chromosomal aneuploidy (PGT-A)
Advanced embryo screening for numerical chromosomal abnormalities.

Preimplantation genetic testing for structural chromosomal rearrangements (PGT-SR)
Advanced embryo assessment for carriers of balanced chromosomal rearrangements.

Preimplantation genetic testing for monogenic disorders (PGT-M)
Family-specific embryo testing for known inherited single-gene disorders.

EMBRYOGENOME

Genomic Preimplantation Genetic Testing
The first preimplantation genetic test that screens the whole exome of IVF embryos

EMBRYOADVANCE

Non-invasive PGT through analysis of cell-free embryonic DNA in spent culture medium
A non-invasive approach to embryo prioritization based on chromosomal copy-number assessment.

PGTADVANCE A

Advanced PGT-A for highly accurate embryo analysis

WHAT IS PGT-A?

Patients undergoing Assisted Reproductive Technology (ART) often present with a reproductive history characterized by repeated unsuccessful outcomes. These may include failure to achieve pregnancy, commonly associated with recurrent implantation failure after embryo transfer, as well as pregnancies that begin but end in miscarriage or, in some cases, are terminated following the prenatal diagnosis of a fetal chromosomal abnormality.

In a significant proportion of these cases, the reduced likelihood of achieving or carrying a pregnancy to term is associated with the presence of **numerical chromosomal abnormalities** in the embryos, known as **aneuploidies**.

Within this context, **Preimplantation Genetic Testing for Aneuploidy** (**PGT-A**) represents an advanced diagnostic approach aimed at identifying chromosomal abnormalities in embryos prior to uterine transfer, with the goal of improving the success rates of assisted reproduction treatments, particularly in patients with reduced reproductive prognosis.

THE PURPOSE OF PGT-A

With **PGT-A**, embryo selection is not based solely on morphological assessment, but also incorporates evaluation of chromosomal status, a key parameter closely linked to the embryo's potential to result in an ongoing pregnancy. By identifying **euploid embryos**, namely those without detectable aneuploidies, PGT-A enables more accurate embryo selection and contributes to improved outcomes in assisted reproduction.

PGTADVANCE A

Advanced PGT-A for highly accurate embryo analysis

ADVANCING THE STANDARD OF CARE IN EMBRYO TESTING

PGTADVANCE-A is GENOMICA's advanced PGT-A platform, developed to deliver highly accurate and clinically reliable embryo chromosomal assessment.

Unlike standard PGT-A methods that rely on DNA quantity alone, **PGTADVANCE-A** uses the proprietary **Dual-Seq** technology, which integrates two distinct analytical methods:

1. **Copy-number assessment by NGS**
   High-resolution sequencing of the embryonic genome using NGS technology, which enables **DNA** quantification across millions of genomic positions on each chromosome, allowing highly accurate determination of chromosomal copy number.

2. **SNP-based B-Allele Frequency (BAF) analysis**
   Analysis of thousands of **single nucleotide polymorphisms (SNPs)** through evaluation of **B-Allele Frequency (BAF)**, aimed at providing orthogonal confirmation of the copy-number results and enhancing the interpretive power of the test.

THE BENEFIT OF DUAL ANALYSIS

In addition to quantifying DNA copy number across the genome, **PGTADVANCE-A** examines thousands of genomic loci at which the DNA sequence may vary between individuals — variations known as **single nucleotide polymorphisms** (**SNPs**). These variations provide an additional, independent layer of information that strengthens result interpretation.

Supported by **advanced bioinformatic tools** and **machine-learning algorithms**, this dual strategy delivers embryo chromosomal assessment with superior accuracy and reliability compared with conventional approaches based solely on quantitative copy-number analysis, increasing confidence in result interpretation and supporting more precise embryo selection.

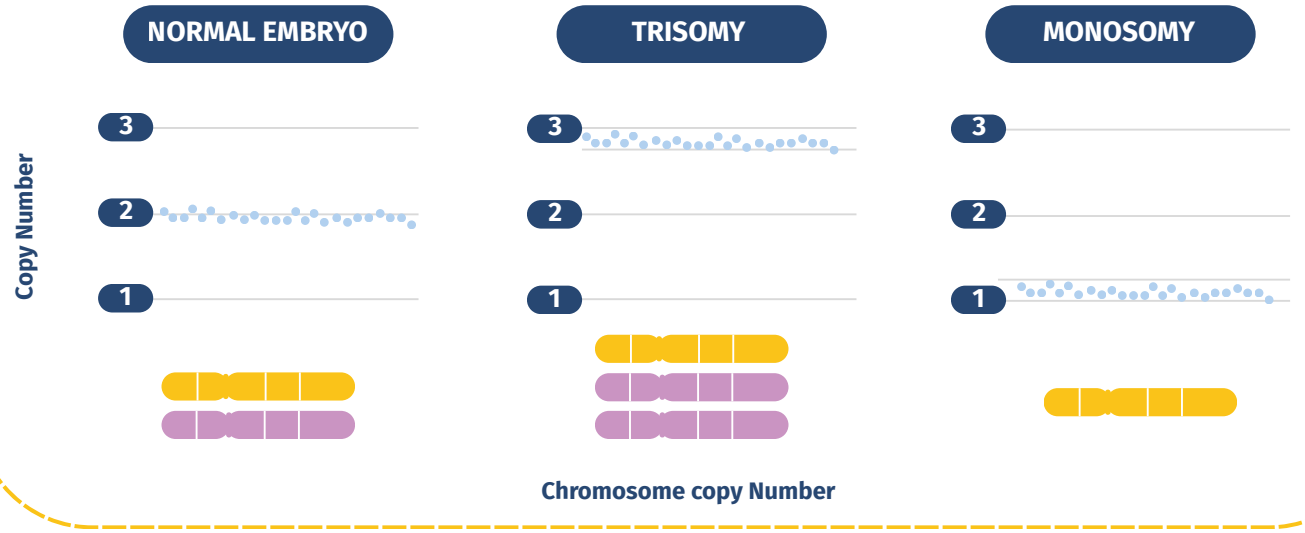
PGTADVANCE A

Advanced PGT-A for highly accurate embryo analysis

TWO TECHNOLOGIES. ONE HIGHLY CONFIDENT RESULT

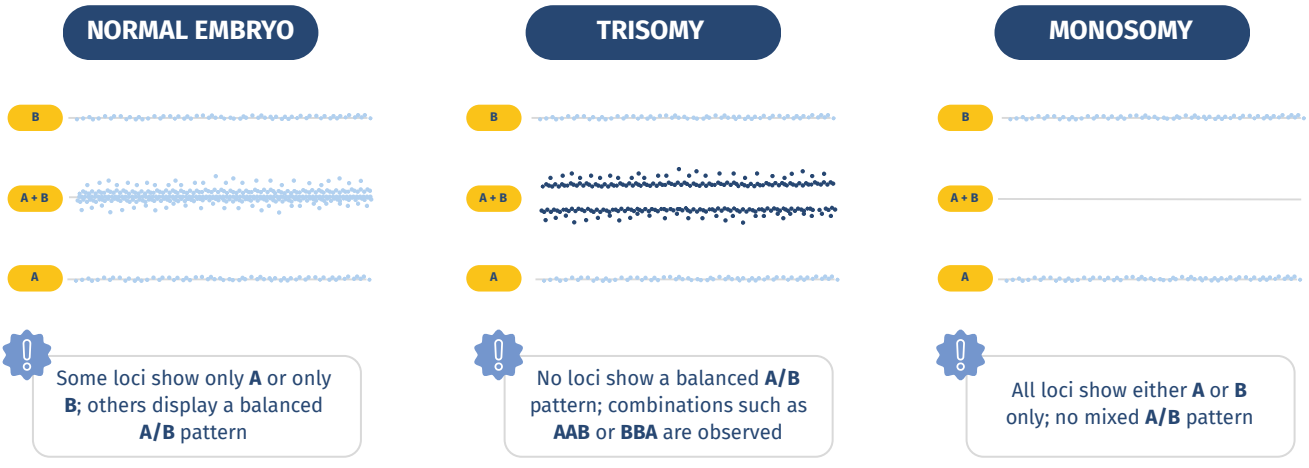
1 NGS-based copy-number assessment

Next-generation sequencing quantifies DNA across millions of genomic positions on each chromosome, enabling highly accurate estimation of chromosome copy number.



2 SNP-based BAF analysis

Thousands of SNP loci are analyzed in parallel to identify characteristic allele patterns that distinguish euploid, monosomic, trisomic, haploid, and triploid states — each displaying distinct SNP signatures.



At each locus, the sequence may be represented by one of two possible forms, conventionally referred to as **A** or **B**. The allelic distribution follows characteristic patterns in different chromosomal states, enabling more accurate differentiation between **euploid**, **trisomic**, and **monosomic** embryos.

PGTADVANCE

Advanced PGT-A for highly accurate embryo analysis

TWO TESTING OPTIONS TAILORED TO YOUR CLINICAL NEEDS

PGTADVANCE

GENOMICA's advanced in-house PGT-A solution for highly accurate embryo chromosomal screening.

PGTADVANCE *Plus*

GENOMICA's most comprehensive PGT-A workflow, expanding beyond standard aneuploidy testing to provide additional diagnostic and quality-control features to address more complex clinical scenarios.

PGTADVANCE A

Advanced PGT-A for highly accurate embryo analysis

WHAT PGTADVANCE-A DETECTS

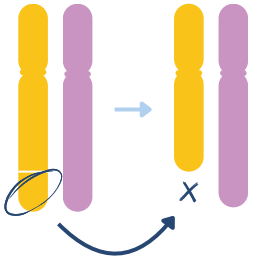
Whole-Chromosome Aneuploidy

Gain or loss of one or more entire chromosomes.

Segmental Aneuploidy

Gain or loss of a chromosomal segment.

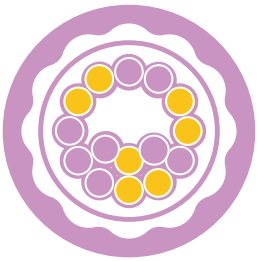
PGTADVANCE-A supports detection of segmental abnormalities typically **>6 Mb.**



Mosaicism

Presence of both chromosomally normal and abnormal cell lines within the same embryo biopsy sample.

- **High-degree mosaicism**: approximately **50–80%** abnormal cells
- **Low-degree mosaicism**: approximately **30–50%** abnormal cells



Mosaic findings are associated with reduced implantation potential and increased miscarriage risk, although healthy live births from mosaic embryo transfer have been reported.

PGTADVANCE A Plus

Advanced PGT-A for highly accurate embryo analysis

BEYOND STANDARD ANEUPLOIDY SCREENING

GENOMICA's most comprehensive PGT-A workflow, expanding standard aneuploidy testing with additional diagnostic and quality-control features for more complex clinical scenarios.

A parallel **SNP-based NGS strategy** complements conventional chromosome copy-number analysis and enables the detection of abnormalities not reliably identified by standard workflows alone. SNPs provide characteristic inheritance patterns that can be leveraged to detect **ploidy abnormalities**, **contamination**, and **genetic relatedness between samples**.

Key advanced features of PGTADVANCE-A Plus

- **Ploidy assessment** (haploidy and triploidy);
- **Microdeletion / microduplication syndromes detection;**
- **DNA contamination assessment** (maternal and external);
- **Genetic PN Check:** molecular confirmation of fertilization status;
- **Origin of aneuploidy analysis**
- **Cohort check (embryo sibling QC)**
- **DNA fingerprinting** (when parental samples are provided)

PGTADVANCE SR

Advanced detection of chromosomal rearrangements in embyos

WHAT IS PGT-SR?

In individuals carrying a **balanced structural chromosomal rearrangement**, such as a chromosomal translocation, a substantial proportion of the gametes produced may be **chromosomally unbalanced**, and the same may consequently apply to the embryos derived from them. In these situations, reproductive risk is increased both in terms of **reduced fertility**, associated with a high incidence of chromosomally unbalanced embryos, and the possibility of conceiving a child affected by a chromosomal abnormality.

Within this context, **Preimplantation Genetic Testing for Structural Rearrangements** (**PGT-SR**) enables the identification and selection of embryos free from chromosomal imbalance, thereby contributing to improve reproductive outcomes and reducing the risk of **miscarriage**, as well as pregnancies or live births affected by severe chromosomal abnormalities.

PGT-SR can be applied across a broad range of structural chromosomal rearrangements, including:

- **reciprocal translocations**
- **Robertsonian translocations**
- **inversions**
- **deletions**
- **duplications**

Example of reciprocal translocations

PGTADVANCE SR

Advanced detection of chromosomal rearrangements in embyos

ADVANCED DETECTION OF CHROMOSOMAL REARRANGEMENTS IN EMBRYOS

PGTADVANCE-SR is GENOMICA's advanced **PGT-SR platform**, developed for couples in which one or both partners carry a structural chromosome rearrangement.
The assay analyzes embryos generated through IVF to distinguish embryos that are **normal or balanced** for the parental rearrangement from those with **unbalanced chromosomal content**, thereby reducing the risk of miscarriage and chromosomally abnormal pregnancy.

High-confidence detection of chromosomal imbalance

PGTADVANCE-SR is performed using **next-generation sequencing** (**NGS**) combined with **SNP-based analysis**, enabling a dual-assessment strategy that provides greater robustness than copy-number analysis alone and supports more informed embryo transfer decisions.

In addition to detecting imbalances associated with the parental rearrangement, **PGTADVANCE-SR** also includes assessment of **aneuploidies**, thereby **incorporating the analytical scope of PGT-A into the PGT-SR workflow**.

PGTADVANCE SR →

>99%
accuracy for detecting chromosomal imbalances and aneuploidy.

PGTADVANCE SR

Advanced detection of chromosomal rearrangements in embyos

TWO TESTING OPTIONS TAILORED TO YOUR CLINICAL NEEDS

PGTADVANCE SR

GENOMICA's advanced in-house PGT-SR solution for highly accurate detection of chromosomal structural imbalances in IVF-derived embryos from carriers of balanced rearrangements.

PGTADVANCE SR *Plus*

GENOMICA's most comprehensive PGT-SR workflow, expanding beyond standard PGT-SR to provide additional diagnostic and quality-control features to address more complex clinical scenarios.

PGTADVANCE-SR Plus is particularly suitable for detection of **very small chromosomal imbalances (>1 Mb)** resulting from a parental structural rearrangement.

PGTADVANCE SR

Advanced detection of chromosomal rearrangements in embyos

WHAT PGTADVANCE-SR DETECTS

Chromosomal structural imbalance

Gain or loss of chromosomal segments resulting from a parental structural rearrangement.
PGTADVANCE-SR typically supports detection of structural imbalance **>6 Mb**.

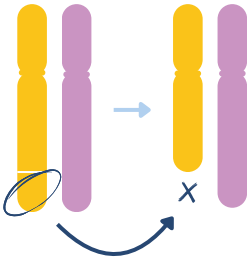
Whole-Chromosome Aneuploidy

Gain or loss of one or more entire chromosomes.

Segmental Aneuploidy

Gain or loss of a chromosomal segment.

PGTADVANCE-SR supports detection of segmental abnormalities typically **>6 Mb.**



Mosaicism

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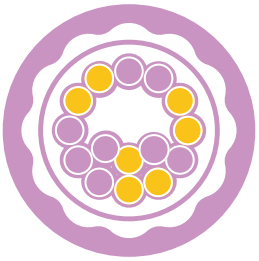


Mosaic findings are associated with reduced implantation potential and increased miscarriage risk, although healthy live births from mosaic embryo transfer have been reported.

PGTADVANCE SR Plus

Advanced detection of chromosomal rearrangements in embyos

BEYOND STANDARD DETECTION OF CHROMOSOMAL REARRANGEMENTS

GENOMICA's most comprehensive PGT-SR workflow, extending standard PGT-SR with additional diagnostic and quality-control features for more complex clinical scenarios.

A **SNP-based NGS assay** complements chromosome copy-number analysis and enables the detection of abnormalities not reliably identified by standard workflows, including **ploidy abnormalities**, **contamination**, and **genetic relatedness between samples**.

PGTADVANCE-SR Plus is particularly suitable for detection of **very small chromosomal imbalances (>1 Mb)** resulting from a parental structural rearrangement.

Key advanced features of PGTADVANCE-SR Plus

- **Detection** of **chromosomal structural imbalance >1 Mb;**
- **Ploidy assessment** (haploidy and triploidy);
- **Microdeletion / microduplication syndromes detection;**
- **DNA contamination assessment** (maternal and external)
- **Genetic PN Check:** molecular confirmation of fertilization status;
- **Origin of aneuploidy analysis**
- **Cohort check (embryo sibling QC)**
- **DNA fingerprinting** (when parental samples are provided)

PGTADVANCE M

Advanced detection of inherited disorders in embryos

WHAT IS PGT-M?

Preimplantation Genetic Testing for Monogenic Disorders (PGT-M) enables the identification of embryos that are unaffected by a specific inherited disease in couples known to be carriers of a familial genetic condition.
This strategy has provided thousands of couples at increased genetic risk with the opportunity to pursue reproduction more confidently, supporting the possibility of having healthy children without having to renounce pregnancy in advance or later face the difficult decision of therapeutic termination.

PGT-M can be applied to a wide range of inherited disorders, including:

- **Cystic fibrosis**
- **Fragile X syndrome**
- **Muscular dystrophy**
- **Huntington disease**
- **Beta-Thalassemia**
- **Numerous additional monogenic disorders**

For couples who already have a child affected by a genetic condition whose treatment requires **stem cell transplantation** from an **HLA-compatible donor**—for example, in cases such as **beta-thalassemia**, **sickle cell disease**, **Fanconi anemia**, and related disorders—**PGT-M**, when combined with **HLA typing** (**Preimplantation HLA Matching**), enables the identification of embryos that are both **unaffected by the specific disease** and **HLA-compatible** with the affected child.
Such embryos may then be selected for uterine transfer. At birth, stem cells collected from the newborn's umbilical cord blood may, where clinically appropriate, be used therapeutically for the affected sibling, offering a concrete treatment perspective.

When clinically indicated, PGT-M may also be combined with **PGT-A** (**Preimplantation Genetic Testing for Aneuploidy**), allowing simultaneous assessment of both the **monogenic condition and the embryo's chromosomal status** from a single **trophectoderm biopsy**.

PGTADVANCE M

Advanced detection of inherited disorders in embryos

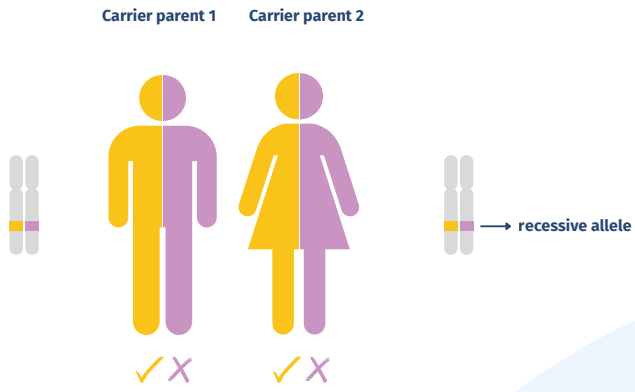
Supporting couples at increased genetic risk in achieving a healthy pregnancy

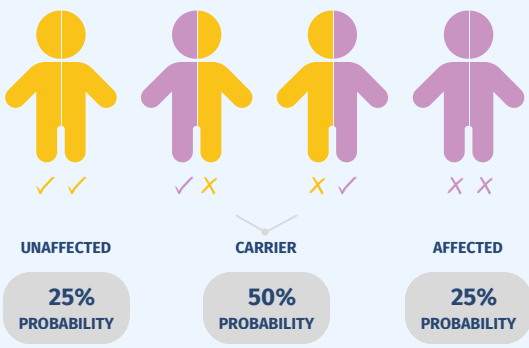
PGTADVANCE-M is GENOMICA's advanced PGT-M service, developed for couples at increased genetic risk who are undergoing **IVF**.

Through molecular analysis of embryonic DNA, **PGTADVANCE-M** enables the identification of embryos unaffected by a known familial monogenic disorder, significantly reducing the risk of a pregnancy affected by the specific genetic condition. **PGTADVANCE-M** can be applied to any genetic disorder for which the causative gene has been identified. Supported by a dedicated **Research and Development Department**, GENOMICA is able to design and optimize **PGT-M** diagnostic strategies across a wide spectrum of inherited conditions.

This development capability also extends to particularly complex cases and to **rare genetic disorders** for which no established diagnostic workflow is yet available. For each couple undergoing a **PGT-M** pathway, a fully personalized PGT strategy is designed and validated, specifically tailored to the genetic variants identified within the family. This customized protocol is then applied clinically to embryonic cells. This preliminary phase of assay design and optimization, known as the **preclinical work-up**, is a critical step in ensuring the highest standards of analytical accuracy and diagnostic reliability throughout the diagnostic process.

Example of autosomal recessive inheritance





PGTADVANCE M

Advanced detection of inherited disorders in embryos

A TEST TAILORED TO EACH FAMILY

Family-specific design for high analytical precision

Each **PGTADVANCE-M** assay is developed on a family-specific basis and typically includes:

1. **Direct Analysis**
   Detection of the causative **pathogenic variant**(s) responsible for the familial condition.

2. **Indirect Analysis**
   Evaluation of flanking chromosomal regions through **linked genetic marker analysis** (STRs/SNPs).

3. **Haplotyping**
   Family-specific **linkage analysis** to strengthen embryo classification and ensure result robustness.

4. **Classification**
   Accurate embryo classification as **affected**, **unaffected**, or **carrier**, per inheritance pattern.

DNA **samples** are generally required from **both reproductive partners** and, where available, from additional family members, in order to define the genetic signature associated with the disease-causing allele.

PGTADVANCE M

Advanced detection of inherited disorders in embryos

STATE-OF-THE-ART TECHNOLOGY

Advanced diagnostic strategies for reliable embryo assessment

PGTADVANCE-M employs advanced molecular and bioinformatic methods to assess whether a preimplantation embryo is likely to be affected by a specific inherited disorder. The approach combines **direct mutation testing** with **indirect marker-based haplotyping** to maximize diagnostic robustness and analytical confidence.

This **dual strategy approach** provides an independent confirmation of embryo classification results, substantially improving the overall analytical accuracy.

Key capabilities:

- Unlike other PGT-M strategies, the **availability of samples from the couple's relatives is not mandatory**;
- **faster assay development** and **validation** timelines;
- **improved analytical accuracy** and **diagnostic reliability**;
- simultaneous assessment of **multiple conditions**, when indicated;
- **HLA matching** capability, in selected cases;
- enhanced robustness through **case-specific assay design**.
- testing across a **broad spectrum of inherited disorders**.

GENOMICA offers PGT-M for the majority of monogenic disorders for which the causative variant has been identified.

EMBRYOGENOME

GENOMIC PREIMPLANTATION GENETIC TESTING

THE NEXT LEVEL IN PGT

EMBRYOGENOME is an innovative preimplantation genetic testing platform designed to analyze the **whole coding genome** (**exome**) **of IVF embryos**, enabling the simultaneous detection of:

- **~7,000** clinically recognized **severe genetic disorders**, including both **inherited** and **de novo** conditions;
- chromosomal **aneuploidies**;
- haploidy, triploidy, and uniparental disomy (**UPD**);
- chromosomal **mosaicism**;
- **structural chromosomal imbalances**, including segmental deletions and duplications;
- **130 + microdeletion** and **microduplication** syndromes;
- small copy number variants (**CNVs**) (**>1 Mb**);
- the **gametic origin** of chromosomal abnormalities;
- genetic relatedness among embryos (**Cohort Check**);
- genetic concordance between embryo biopsies and parental DNA (**DNA Fingerprinting**);
- DNA **contamination**.

Through this exceptional analytical breadth, **EMBRYOGENOME** enables the identification of genetic and chromosomal abnormalities that conventional PGT approaches may not detect, delivering an **unprecedented level of genetic insight** in embryo screening.

EMBRYOGENOME

GENOMIC PREIMPLANTATION GENETIC TESTING

REDEFINING EMBRYO SCREENING

EMBRYOGENOME represents a major technological advancement in embryo screening, overcoming the limitations of conventional preimplantation genetic testing and, in particular, those of PGT-A, which has traditionally focused primarily on chromosomal analysis.

This next-generation platform uniquely integrates the capabilities of PGT-A, PGT-M, and PGT-SR into a single, comprehensive system, offering unprecedented levels of accuracy, completeness, analytical resolution, and clinical value.

Advantages over conventional PGT approaches

Test	Advantages
PGT-A	• Up to **1,000-fold higher resolution** than conventional **PGT-A** methods; • detection of **triploidy** and **molar embryos**, conditions that may be missed by standard technologies; • identification of **microdeletions** and **microduplications** not detectable by traditional **PGT-A** workflows; • inclusion of all the core capabilities of conventional **PGT-A**.
PGT-SR	• **Greater analytical precision**, enabled by a higher resolution than currently available **PGT-SR** platforms; • increased likelihood of **case acceptance** in the PGT-SR setting for structurally complex rearrangements; • **expanded screening scope**, with additional analysis for hundreds of genetic disorders; • inclusion of all the functionalities of conventional **PGT-SR**.

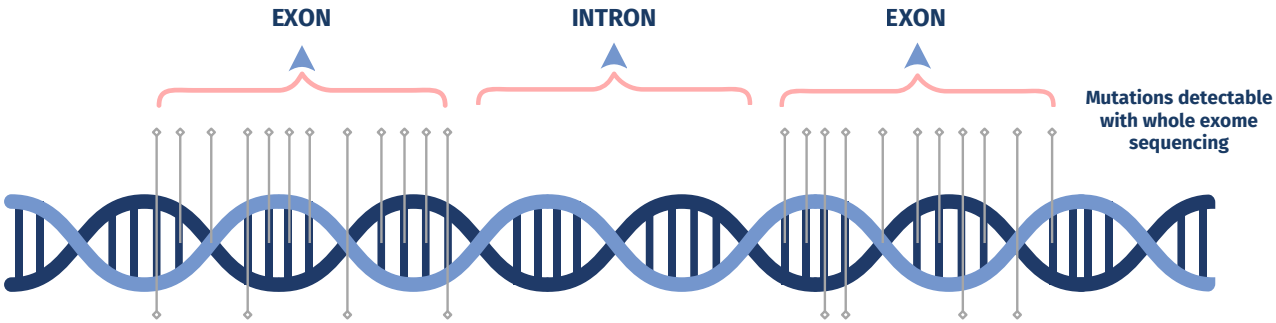
PGT-M	• A **streamlined workflow**, with no requirement for a preliminary custom assay set-up, resulting in shorter turnaround times;
	• particular value in **complex clinical scenarios**, including:
	  ◦ cases not accepted by other PGT laboratories;
	  ◦ patients with previous pregnancies or children affected by **de novo variants**;
	• **expanded analytical scope**, with the ability to assess hundreds of monogenic disorders simultaneously.

EMBRYOGENOME

GENOMIC PREIMPLANTATION GENETIC TESTING

NEXT-GENERATION ADVANCED TECHNOLOGY

High-resolution **Whole Exome Sequencing** of embryonic DNA, combined with **sophisticated bioinformatic analysis** powered by proprietary algorithms, enables highly accurate and comprehensive embryo screening.



Leveraging state-of-the-art Next Generation Sequencing (NGS) technology, this advanced approach allows the identification of severe genetic disorders, including both **inherited** and **de novo** conditions, while simultaneously assessing the embryonic chromosomal profile for the detection of **aneuploidies**, **segmental deletions** and **duplications**, **microdeletion/microduplication syndromes**, and **chromosomal mosaicims**.

All of this is achieved through a **single integrated analysis**, delivering the highest level of diagnostic insight currently available in preimplantation genetic testing.

DNA extraction from TE cells

Whole Exome Sequencing by Next Generation Sequencing (NGS)

Embryonic DNA screening for mutations and chromosomal abnormalities

Test Report

EMBRYOGENOME

GENOMIC PREIMPLANTATION GENETIC TESTING

KEY ADVANTAGES

Comprehensive diagnostic insight

Integrates the core functionalities of conventional preimplantation genetic testing within a single platform, combining the strengths of PGT-A, PGT-M, and PGT-SR.

Advanced genetic screening

Extends beyond the limits of conventional technologies, enabling the detection of genetic and chromosomal conditions that may go undetected with traditional preimplantation testing.

Assessment of conditions associated with advanced paternal age

Supports the identification of genetic disorders associated with increasing paternal age.

Detection of de novo mutations

Enables the detection of de novo pathogenic variants associated with severe genetic conditions.

High precision

With an accuracy exceeding 99%, it provides a substantially higher level of precision than conventional PGT approaches, ensuring a high detection rate for both genetic and chromosomal abnormalities.

EMBRYOADVANCE

cell-free embryonic DNA-based PGT

A NEW FRONTIER IN NON-INVASIVE PGT

Cell-free embryonic DNA in culture medium

The recent identification of **cell-free embryonic DNA** in spent culture medium has opened new perspectives in the field of assisted reproduction, particularly in the area of **non-invasive preimplantation testing for chromosomal aneuploidies**.
During **in vitro** development, embryos generated through **IVF** naturally release DNA fragments into the culture medium. These fragments, referred to as **cell-free embryonic DNA**, tend to increase in concentration as embryonic development progresses.
This discovery has enabled the development of innovative testing strategies that, through the use of state-of-the-art technologies, allow **non-invasive assessment of embryonic chromosomal status**, without the need for **trophectoderm biopsy**.

EMBRYOADVANCE

EMBRYOADVANCE is GENOMICA's innovative non-invasive preimplantation test, developed to identify embryos with a higher probability of **euploidy** and, consequently, greater implantation potential.
Based on the analysis of **cell-free embryonic DNA** released into the culture medium during embryo development, **EMBRYOADVANCE** offers an advanced approach to **embryo prioritization**, without requiring trophectoderm biopsy.

Who it is for

EMBRYOADVANCE is intended for patients wishing to enhance their chances of pregnancy within an **IVF** pathway, while avoiding invasive procedures such as embryo biopsy.

EMBRYOADVANCE

cell-free embryonic DNA-based PGT

The procedure

EMBRYOADVANCE analyzes cell-free embryonic DNA present in the culture medium using **Next-Generation Sequencing** (**NGS**) technology and advanced bioinformatic tools. The test is performed on a sample of embryonic culture medium collected by embryologists at the IVF center on **day 6** or **day 7** of embryo development. The chromosomal regions represented within the cell-free embryonic DNA are then sequenced using **NGS technology**.

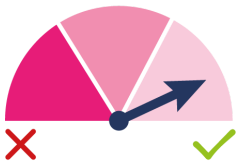
The resulting sequences are subsequently quantified and interpreted through advanced bioinformatic analysis. A proprietary algorithm ultimately assigns each embryo a **quality score** and, accordingly, a level of priority for uterine transfer (**High**, **Medium**, or **Low** Priority).

The results

EMBRYOADVANCE assigns each embryo a transfer priority level based on information regarding its chromosomal profile, obtained through the analysis of cell-free embryonic DNA.

HIGH SCORE

High Priority: indicates that the test has identified embryos with a high probability of **euploidy** and, therefore, a higher implantation potential

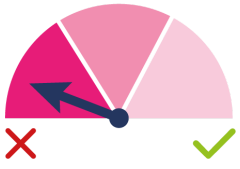


MEDIUM SCORE

Intermediate Priority: indicates that the test has identified embryos with an intermediate probability of **aneuploidy** and, therefore, reduced implantation potential or a higher risk of miscarriage.

LOW SCORE

Low Priority: indicates that the test has identified embryos with a high probability of **aneuploidy** and, therefore, very low implantation potential or a high risk of miscarriage.



PGTADVANCE
NEXT GENERATION PGT

CLINICAL INDICATIONS AND KEY BENEFITS

Test	Clinical Indications	Clinical Purpose	Key Benefits
PGTADVANCE-M	Couples who are carriers of inherited genetic disorders	To identify embryos unaffected by the specific familial genetic disorder under investigation	Reduces the risk of transferring embryos affected by inherited genetic disease
PGTADVANCE-SR	Couples who are carriers of balanced chromosomal rearrangements	To identify embryos free from chromosomal imbalances arising from the parental rearrangement	Reduces the risk of transferring embryos with unbalanced structural chromosomal abnormalities
PGTADVANCE-A	• Advanced maternal age • Recurrent miscarriage • Recurrent implantation failure • Severe male infertility	To identify embryos free from numerical chromosomal abnormalities (aneuploidies)	• Enhances embryo selection • Reduces the risk of transferring aneuploid embryos • Supports improved clinical IVF outcomes • Shortens time to pregnancy and helps reduce miscarriage risk
EMBRYOGENOME	• Personal and/or family history of genetic disease or chromosomal abnormalities • Couples wishing to minimize reproductive risk • Couples undergoing donor-assisted reproduction • Couples pursuing IVF with integrated PGT • Advanced paternal age	To identify genetic and chromosomal abnormalities that may not be detected by conventional PGT approaches, providing an unprecedented level of diagnostic insight in embryo screening	• Comprehensive diagnostic coverage • Advanced genetic screening • Assessment of conditions associated with advanced paternal age • Detection of de novo variants • High analytical accuracy
EMBRYOADVANCE	Patients seeking to improve their chances of pregnancy within an IVF pathway without resorting to invasive procedures such as embryo biopsy	To identify, in a non-invasive manner, embryos with a higher probability of euploidy and, therefore, greater implantation potential	• Improves efficiency within IVF pathways • Helps reduce miscarriage risk • Safe and non-invasive approach • No embryo biopsy required

PGTADVANCE
NEXT GENERATION PGT

CLINICAL PATHWAY

From referral to embryo transfer

1

Referral and specialist case review

Clinical and genetic documentation is reviewed for feasibility and case planning (**PGT-M and PGT-SR only**).

2

Genetic counseling consultation

Where appropriate, patients are offered specialist counseling regarding indication, workflow, and testing strategy.

3

Case acceptance and testing plan

A formal testing plan is issued to the referring clinic. For **PGT-M**, this includes any assay development requirements.

4

Custom assay development

For **PGT-M**, a family-specific assay is designed and validated based on the identified pathogenic variant(s). A formal work-up report is then issued to the clinic.

5

IVF cycle and embryo biopsy

Embryos are generated through IVF and trophectoderm biopsy is performed at the blastocyst stage.

6

Shipment of biopsy samples

Embryo biopsy samples are transported to **GENOMICA** under validated shipping conditions.

7

PGTADVANCE testing and result reporting

Embryonic DNA is analyzed using the relevant PGTADVANCE workflow and a formal genetic report is issued to the IVF team.

8

Transfer planning

Embryos classified as **unaffected**, **normal** or **balanced**, or **euploid**, as applicable, may be considered for transfer according to the clinical treatment plan.

PGTADVANCE
NEXT GENERATION PGT

WHY GENOMICA

A Proven Legacy in Reproductive Genetics

GENOMICA builds upon the collective expertise of specialists with more than **25 years of experience in preimplantation genetic testing**, combining established scientific knowledge with advanced molecular technologies to support accurate and clinically meaningful embryo assessment.

This longstanding commitment reflects a continuous dedication to innovation in reproductive genetics and to the ongoing refinement of PGT methodologies for clinical practice.

High Accuracy. Greater Confidence.

Improved genome coverage, optimized analytical workflows, and case-specific assay design contribute to highly reliable PGT results and increased confidence in embryo classification.

Through the integration of advanced laboratory methodologies, specialist clinical interpretation, and individualized case management, GENOMICA delivers a high standard of service for both clinicians and patients.

Genomica's commitment includes:

- **specialist genetic counseling support** at all stages of the testing process;
- **advanced nucleic acid amplification** and **analytical technologies**;
- **high quality laboratory** and **clinical service standards**;
- strong focus on analytical **accuracy**, result **reliability**, and individualized **patient care**.

GENOMICA
Next Generation Genetics

Excellence in Reproductive Genetics

GENOMICA is a highly specialized diagnostic laboratory and a recognized center of excellence in **reproductive genetics**, active in both **clinical diagnostics** and **scientific research**. Supported by a team with more than **25 years of experience in molecular diagnostics**, GENOMICA combines scientific expertise, advanced technology, and a strong commitment to continuous innovation to provide high-quality reproductive genetic services to IVF clinics worldwide.

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

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

ISO 9001 certified laboratory

Rapid Turnaround Time

PGTAdvance M_ENG rev.00

LABORATORIES

Rome: Via Arduino 38 - 00162
Tel.: 06.21115020
E-mail: info@genomicalab.it
www.genomicalab.it

REGISTERED OFFICE

Rome: Via Arduino 38 - 00162
PEC: info@pec.genomicalab.it
P. IVA e C.F.: 14554101007
REA: RM - 1530210

Visit the website dedicated to the test