GENOMICA
Next Generation Genetics

An innovative genetic test to identify predisposition to producing embryos with aneuploidies

FERTILADVANCE

ANEUPLOIDY RISK

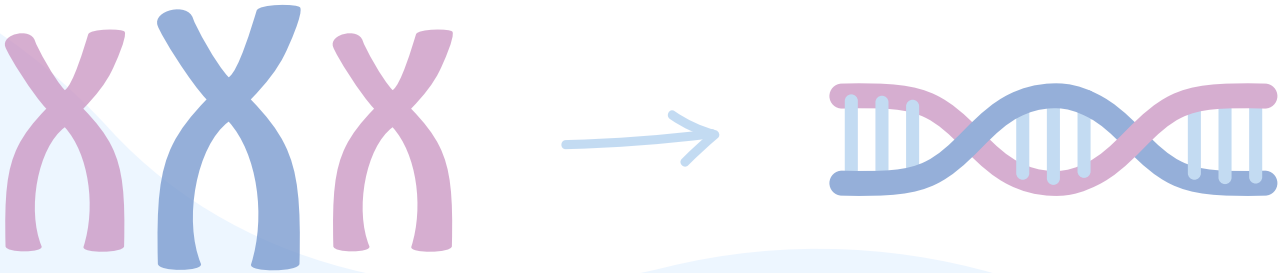
www.fertiladvance.it

FERTILADVANCE ANEUPLOIDY RISK

WHY TALK ABOUT ANEUPLOIDIES

Aneuploidies are numerical chromosome abnormalities: a cell may carry one extra chromosome (**trisomy**) or one missing chromosome (**monosomy**) compared with the normal chromosomal complement. In reproductive medicine, these abnormalities are among the most frequent causes of **implantation failure**, **early miscarriage**, and, more rarely, **ongoing pregnancies affected by a chromosomal abnormality** up to birth.

A key aspect—often relevant for clinical assessment and for interpreting laboratory findings (including **PGT-A**)—is that aneuploidy can have **different biological origins**, with different implications for pathophysiology and risk.



FERTILADVANCE ANEUPLOIDY RISK

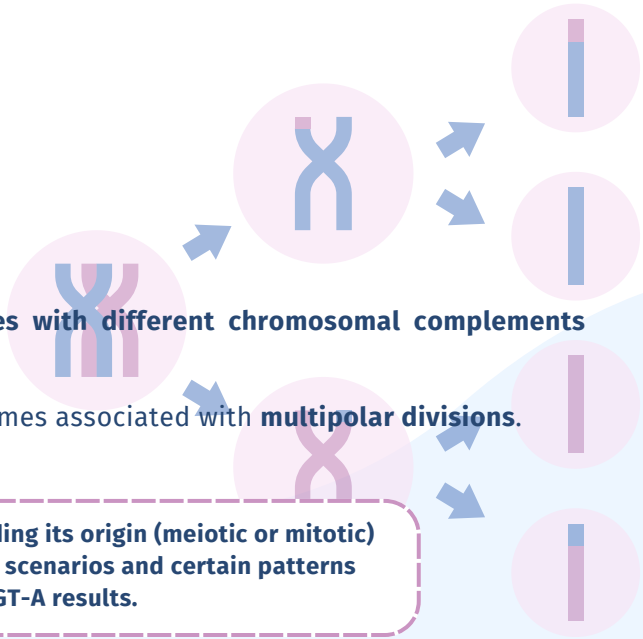
ANEUPLOIDIES: MEIOTIC ORIGIN VS MITOTIC ORIGIN

Aneuploidies of meiotic origin

These arise during **meiosis**, the process that leads to the formation of gametes (in particular, the **oocyte**). In humans, most meiotic aneuploidies are of maternal origin and show a well-known relationship with **age**; however, in addition to age, **individual genetic factors** may also contribute.

Aneuploidies of mitotic (post-zygotic) origin

These occur **after fertilization**, during the very first **mitotic divisions** of the embryo, due to chromosome segregation errors. Such errors may lead to:



- **embryonic mosaicism (the presence of cell lineages with different chromosomal complements within the same embryo);**
- **complex patterns** with multiple aneuploidies, sometimes associated with **multipolar divisions**.

Aneuploidy is not a "single" phenomenon. Understanding its origin (meiotic or mitotic) can help specialists better interpret specific clinical scenarios and certain patterns observed in ART pathways and PGT-A results.

FERTILADVANCE ANEUPLOIDY RISK

WHAT IS FERTILADVANCE ANEUPLOIDY RISK

FERTILADVANCE ANEUPLOIDY RISK is an advanced genetic test designed to assess predisposition to producing embryos with aneuploidies by analyzing a selected set of **13 genes** associated with an increased likelihood of **meiotic and/or mitotic** aneuploidies.

Main aims

The purpose of the test is not to "diagnose" an embryonic aneuploidy, but to provide specialists with an **individual risk-stratification tool** useful to:

- complement **reproductive genetic counseling**;
- enable a more informed **interpretation of complex clinical scenarios** (e.g., repeated implantation failures, recurrent pregnancy loss, a higher-than-expected proportion of aneuploid embryos);
- contextualize **results from previous investigations** (e.g., PGT-A patterns suggestive of meiotic or mitotic errors).

FERTILADVANCE ANEUPLOIDY RISK

ANALYZED GENES AND BIOLOGICAL RATIONALE

Genes have been identified in which **pathogenic** variants are associated with an increased likelihood of **meiotic** and/or **mitotic** aneuploidies.

The test includes the analysis of the following genes →

SMC1B, RNF212, CCNB1IP1 (HEI10), C14orf39, CCDC66, CEP120, AURKB, AURKC, KIF18A, KIF20A, TLE6, TP73, PLK4

Overall, the analyzed genes fall into three major functional areas:

1. **meiotic cohesion and recombination** (key determinants of non-disjunction risk);
2. **segregation and checkpoint control** (relevant in both meiosis and mitosis);
3. **centrosome function and spindle dynamics** (relevant to early mitotic errors and complex/mosaic patterns).

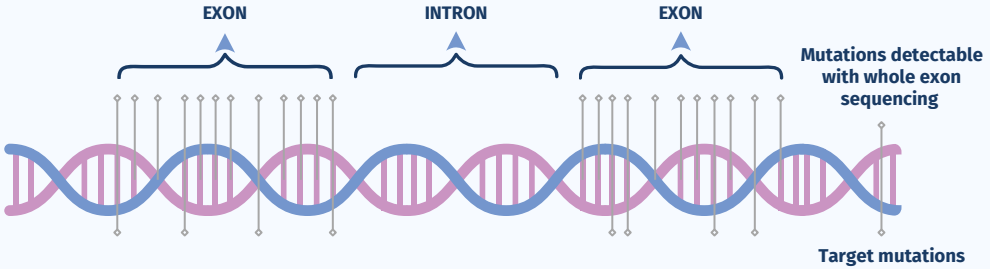
1.Carioscia et al. 2026 Common variation in meiosis genes shapes human recombination and aneuploidy Nature 2026 Jan 21. doi: 10.1038/s41586-025-09964-2.
2.Sun et al. 2023 Identifying risk variants for embryo aneuploidy using ultra-low coverage whole-genome sequencing from preimplantation genetic testing. Am. J. Hum Genet 2023; 110:2092-2102
3.Tyc et al, 2020 Exome sequencing links CEP120 mutation to maternally derived aneuploid conception risk. Hum Reprod. 2020 9;35:2134–2148.
4.Nguyen et al. 2017 Identification and characterization of Aurora kinase B and C variants associated with maternal aneuploidy. Mol Hum Reprod 2017 13;23:406–416.
5.Biswas, et al. 2024 Maternal genetic variants in kinesin motor domains prematurely increase egg aneuploidy, Proc. Natl. Acad. Sci. U.S.A.(2024) 121 (45) e2414963121.
6.Lledo et al. 2023 Identification of novel candidate genes associated with meiotic aneuploidy in human embryos by whole-exome sequencing. J Assist Reprod Genet . 2023;40:1755-1763.
7.McCoy et al. 2015 Common variants spanning PLK4 are associated with mitotic-origin aneuploidy in human embryos. Science. 2015 10;348:235–238.

FERTILADVANCE ANEUPLOIDY RISK

TECHNOLOGY: HIGH RESOLUTION AND ADVANCED BIOINFORMATICS

FERTILADVANCE ANEUPLOIDY RISK uses next-generation sequencing (**NGS**) **technology** with sequencing of the entire coding region (exons) of the investigated genes and an advanced bioinformatics pipeline.

Advantage over "target" tests: it does not search only for selected mutations, but enables identification of **any variant** within the sequenced regions.



FERTILADVANCE ANEUPLOIDY RISK

TEST RESULTS

Positive: pathogenic variant detected

The presence of rare variants with plausible functional impact in genes involved in meiosis/mitosis may indicate a **predisposition to producing embryos with aneuploidies**. Interpretation should always be integrated with clinical data and, when appropriate, with genetic counseling.

Negative: no pathogenic variant detected

No variants associated with an increased risk of producing embryos with chromosomal aneuploidies are identified. A negative result **does not exclude** aneuploidy risk, as the phenomenon is multifactorial and depends also on maternal age and other biological variables not investigated by the test.

FERTILADVANCE ANEUPLOIDY RISK

TEST INDICATIONS

The test may be considered when investigating a possible genetic contribution to an increased aneuploidy risk in scenarios such as:

- **repeated ART/IVF cycles** with a high proportion of aneuploid embryos, especially if unexpected for maternal age;
- **recurrent implantation failure** or **recurrent pregnancy loss**, particularly when there are data suggestive of a chromosomal origin;
- a previous conception/pregnancy affected by **chromosomal aneuploidy**;
- the need for a **preconception** or **pre-IVF** assessment to clarify possible genetic contributors;
- the presence of patterns suggestive of **mitotic errors** (e.g., “chaotic” patterns, suspected tripolarity) or **meiotic errors** (recurrent aneuploidies consistent with non-disjunction).

FERTILADVANCE ANEUPLOIDY RISK

A SIMPLE, STEP-BY-STEP WORKFLOW

Kit request

Completion of kit documentation

Sample collection

Sample shipment

Report delivery

Turnaround time **15 days**

GENOMICA
Next Generation Genetics

Leading diagnostic laboratory and research center of excellence specializing in genetics and molecular diagnostics.

GENOMICA is a highly innovative company with extensive technical and scientific expertise, active in both clinical applications and research. Supported by a team with over 20 years of experience in molecular diagnostics, GENOMICA combines cutting-edge technology with a strong commitment to innovation, delivering increasingly accurate and accessible diagnostic services.

Over **100.000 genetic tests/year**

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

Dedicated **R&D Team**

International **Partnerships**

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

20+ years experience in prenatal molecular diagnostics

FertilAdvance Aneuploidy Risk 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
VAT No.: 14554101007 - **REA:** RM - 1530210

Visit the website dedicated to the test