

# Preimplantation Genetic Testing–Aneuploidy [PGT-A]

Preimplantation genetic testing is performed on embryos created through in-vitro fertilization (IVF). An embryo is created after an egg is fertilized by a sperm. After fertilization, embryos develop from a single cell into a cluster of around 200 cells. In order to perform the genetic analysis, a few cells are biopsied from the embryo. The goal of PGT-A is to implant the embryo with the lowest chance to have a chromosomal condition in order to improve implantation rates and reduce miscarriage as well as the possibility of a pregnancy with a chromosomal condition.

## What does aneuploidy mean?

Our bodies rely on about 20,000 genes to guide our growth and development. These genes are packed into structures called chromosomes within our cells. Each chromosome carries around 500-1000 genes. Most people have a total of 46 chromosomes. However, sometimes there can be a missing or extra chromosome, which can cause extra or missing genes. This imbalance may lead to conditions like heart defects or intellectual disabilities. Aneuploidy is a term used to describe when there are too many or too few chromosomes.

## Why choose PGT-A?

A chromosomal condition can happen to anyone when they conceive, regardless of age or family history. However, some people have a higher chance of having embryos or pregnancies with a chromosomal condition. PGT-A helps to prevent implanting embryos with an abnormal number of chromosomes. If an embryo has too few or too many chromosomes, it is more likely to fail to implant, miscarry, or result in a pregnancy with a chromosomal condition.

Some individuals may choose not to have PGT-A because the test results could limit the embryos they can transfer, and the testing can be costly. Ongoing studies are evaluating the usefulness of PGT-A and determining who can benefit the most from the information. There is ongoing debate about whether PGT-A is helpful for women under 35 years old. It is important to discuss PGT-A with a healthcare provider to determine if it is an appropriate test for you.

## What are the limitations of PGT-A?

The limitations of PGT-A include the potential for false negative or false positive results since it is a screening test. Another limitation is that PGT-A analyzes cells from the embryo that would develop into the placenta and supporting structures, not directly assessing the cells of the future baby. While the results provide insights into the overall chromosome status of the embryo, there can be instances where the assessed cells do not accurately represent the chromosome number of the entire embryo. Therefore, testing for chromosomal conditions during pregnancy or after birth may still be necessary to obtain a comprehensive assessment.

### Resources:

Cheng, Xin et al. "Preimplantation Genetic Testing for Aneuploidy With Comprehensive Chromosome Screening in Patients Undergoing In Vitro Fertilization: A Systematic Review and Meta-analysis." *Obstetrics and gynecology* vol. 140,5 (2022): 769-777. doi:10.1097/AOG.0000000000004962

**For more information, please speak to your healthcare provider.**

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