Understanding your PGT-A results

Preimplantation genetic testing for aneuploidy (PGT-A) can **improve your chances of having a successful pregnancy and a healthy baby** by identifying embryos with the correct amount of chromosomal material.

What is PGT-A?

PGT-A is a screening test used to guide embryo selection by checking an embryo for aneuploidy, a condition in which a cell contains an incorrect number of chromosomes.

Sometimes this happens when there are too many or too few **complete** chromosomes. Other times, in cases of segmental aneuploidy, there may be extra or missing **pieces** of chromosomes.

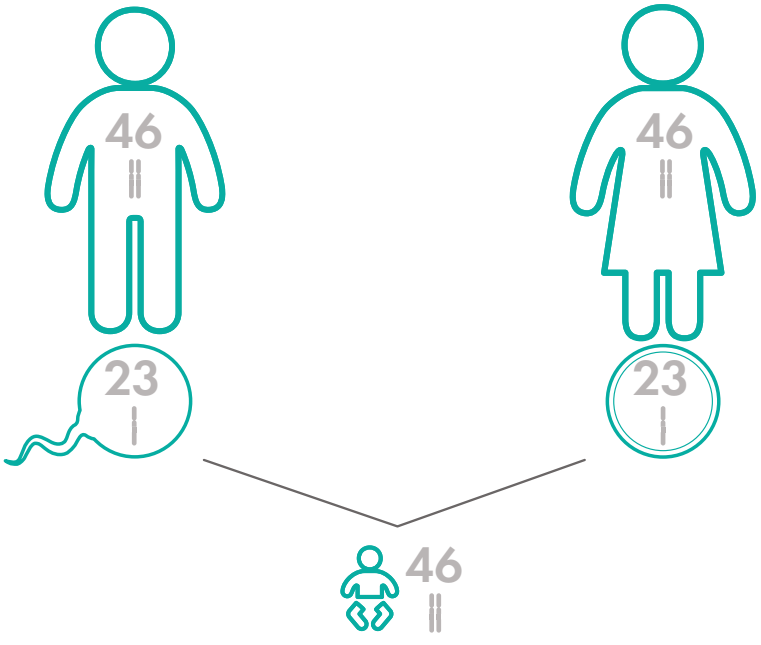
In either situation, the embryo may be considered unsuitable for implantation into the uterus, as most aneuploid embryos do not lead to a healthy live birth. Many cases of aneuploidy fail to implant in the uterus or lead to a spontaneous miscarriage. Even when implantation and pregnancy have been successful, some cases of aneuploidy have resulted in a baby with birth defects, intellectual disability, or other health problems.

What happens during a PGT-A?

Each embryo is made up of many cells. PGT-A involves the removal of a small sample of these cells. This is called a biopsy. The biopsied cells are then sent to a genetic testing laboratory to assess whether they have the correct number of chromosomes.

A healthy cell typically has 46 chromosomes (23 pairs). Half of these chromosomes are inherited from the egg and the other half from the sperm. The first 22 pairs of chromosomes, called autosomes, are numbered 1 through 22. The 23rd pair contains the sex chromosomes, X or Y.

For healthy growth and development, a person must inherit 23 chromosomes from each reproductive parent, receiving a combined total of 46.



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Understanding your PGT-A results *(continued)*

What does my PGT-A result mean?

Normal – The embryo sample is euploid, which means it has the correct number of whole chromosomes. The associated embryo is a **good candidate for IVF implantation**.

Abnormal – Indicates the presence of aneuploidy, which means the cells from the embryo sample contain an incorrect number of complete chromosomes, or that they contain extra or missing pieces of chromosomes. The associated embryo is likely **not a good candidate for IVF implantation**. A common example of an abnormal result is an extra copy of chromosome 21; this is called trisomy 21, or Down syndrome.

No results – The test did not yield results. This result is unlikely but may happen if the laboratory is unable to test chromosomes due to a lack of DNA or very poor quality DNA. In cases with no results, we recommend a re-biopsy to obtain a new sample from the embryo.

Indeterminate – The laboratory was able to test the chromosomes, but unfortunately, there was not enough data to provide a reliable result. If you receive an indeterminate result, we recommend a re-biopsy of the embryo and repeated testing.

Special considerations – The sample has the correct number of whole chromosomes. However, the interpretation was based on low-quality data. Sometimes, this interpretation will be treated as a normal result, but in certain cases, a re-biopsy is recommended to help confirm the test result.

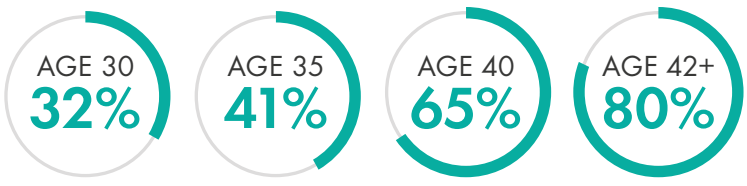
What do abnormal results mean for my embryo and me?

In general, abnormal embryos do not result in a healthy live birth: many lead to miscarriage, some fail to implant in the uterus, and some result in a baby with birth defects, intellectual disability, or other health problems. A common example of an abnormal result is an extra copy of chromosome 21; this is called trisomy 21, or Down syndrome.

Aneuploidy is usually an uncontrollable, sporadic event and is rarely inherited. While extra or missing chromosome(s) can come from either the egg or the sperm, most originate from the egg. Research shows that the chance of having an aneuploid embryo increases with maternal age. Below is a chart that summarizes the PGT-A aneuploidy rates by egg age, as found through Invitae testing.

Maternal age and aneuploidy: Percentage of embryos with an abnormal number of chromosomes

Adapted from internal data on over 90,000 embryos, 2018.



Questions regarding your test results, and decisions involving embryo transfer, should be discussed in detail with your healthcare provider. A genetic counselor can also help you understand your test results and options.

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