Understanding your PGT-A results:
Mosaicism

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.

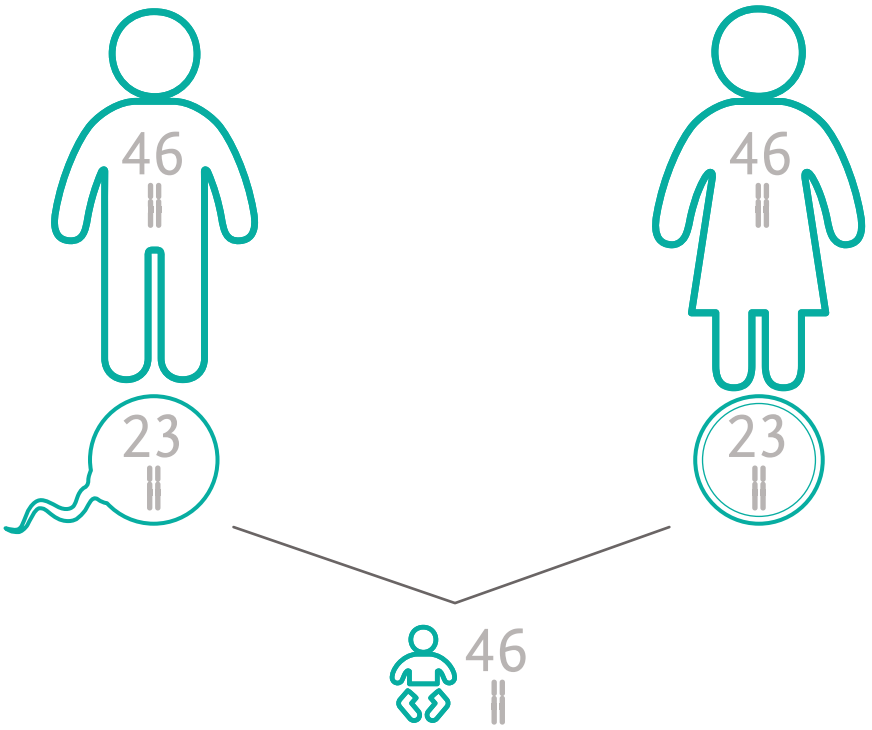
In either situation, the embryo may be considered unsuitable for implantation into the uterus, as most aneuploid embryos do not lead to a healthy live birth. Many cases of aneuploidy fail to implant in the uterus or lead to a spontaneous miscarriage. Even when implantation and pregnancy have been successful, 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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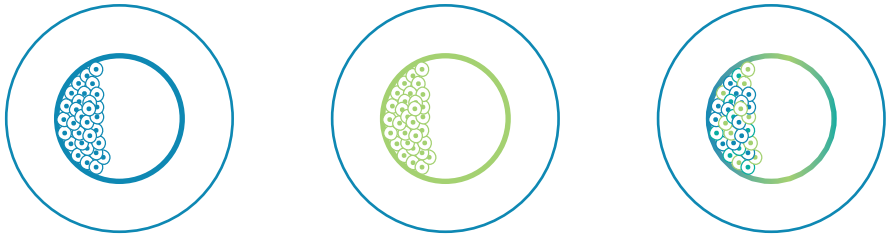
WHAT DOES MY PGT-A RESULT MEAN?

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.

Mosaic – Indicates that the sample has mosaicism, a condition in which some cells from the embryo sample have a normal number of chromosomes and other cells have an abnormal number of chromosomes (whole or segmental aneuploidy).



Normal euploid | Abnormal aneuploid | Mosaic

WHAT ARE THE POTENTIAL OUTCOMES OF TRANSFERRING A MOSAIC EMBRYO?

When an embryo is identified as a mosaic, it is unfortunately not possible to predict the chromosomal effects on pregnancy or the baby. Mosaicism has many potential outcomes, which depend on the amount of extra or missing chromosomal material, the chromosome(s) involved, the percentage of normal versus abnormal cells, and the cell types and/or organ systems ultimately impacted by the abnormality.

While it is possible to give birth to a healthy baby after implanting a mosaic embryo, other potential outcomes include failed implantation and miscarriage, or the birth of a baby with intellectual and/or physical birth defects.

The Preimplantation Genetic Diagnosis International Society (PGDIS) states that implanting mosaic embryos should only be considered when there is no alternative (e.g., no euploid embryos are available).

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

Reference:

Preimplantation Genetic Diagnosis International Society. PGDIS position statement on chromosome mosaicism and preimplantation aneuploidy testing at the blastocyst stage. *PGDIS Newsletter*, July 19, 2016.