

PATIENT INFORMATION

Mosaic PGT-A Results

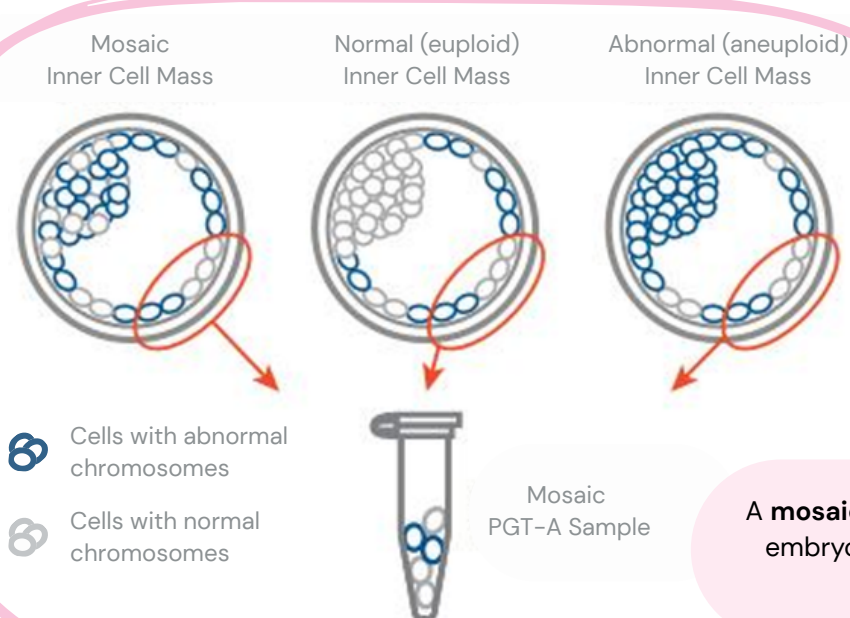
This fact sheet is designed to help you understand what a mosaic result means, why it occurs and how it may be useful in your decision making. If you are considering transferring a mosaic embryo, a consultation will be arranged for you with a genetic counsellor to discuss your specific result.

What is PGT-A?

PGT-A is a genetic test performed on embryos created through IVF. A small number of cells are taken from the outer layer of the embryo (the trophoctoderm), which later forms the placenta. These cells are tested to estimate whether the embryo has the usual number of chromosomes. The cells that go on to form the baby (known as the inner cell mass or ICM) are not tested directly.

PGT-A results are commonly reported as one of the following:

- **Euploid:** Biopsy sample contains a typical number of chromosomes
- **Aneuploid:** Biopsy sample contains an abnormal number of chromosomes (too many or too few). Aneuploid embryos usually aren't able to develop into a healthy pregnancy.
- **Mosaic:** Biopsy sample contains a mixture of cells with different chromosome make-ups. This result usually indicates some of the cells have a normal number of chromosomes (euploid) and some of the cells have an abnormal number of chromosomes (aneuploid).



PGT-A tests a small number of cells from the embryo, so a **mosaic result may not reflect all cells within the embryo**

Aneuploid and euploid results are typically an accurate representation of the rest of the embryo

A **mosaic** result means that the rest of the embryo could be normal, abnormal or a mixture of both

What causes Mosaic PGT-A results?

Mosaic embryos are common and occur entirely by chance. They are not caused by medication, lifestyle factors or the IVF process. Having a mosaic result does not increase the chance of having another mosaic result in future.

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What is the chance of a successful transfer with a mosaic embryo?

Compared with embryos with normal chromosomes, mosaic embryos have a lower chance of implantation and a higher chance of miscarriage. Despite this, many mosaic embryo transfers result in ongoing pregnancies and healthy babies. Reported cases of babies born with chromosome conditions after mosaic embryo transfer are uncommon, but ongoing data collection continues.

What will be discussed in my Mosaic Consult?

During your appointment, your genetic counsellor will review

- The degree of mosaicism found in your embryo (the proportion of cells affected by a chromosome change)
- The type of chromosome change seen in your embryo
- How this information fits with your personal values and reproductive goals

Low Level Mosaic
20-40% of tested cells
were abnormal

High Level Mosaic
40-80% of tested cells
were abnormal

Can mosaic embryos result in healthy pregnancies?

Yes. Many mosaic embryos have resulted in successful implantation, ongoing pregnancies and healthy babies. However, mosaic embryos have lower implantation rates and higher miscarriage rates compared to embryos with normal chromosomes. The chance of success depends on the specific mosaic result and other individual factors.

What tests are recommended in an ongoing pregnancy?

Most babies born following mosaic embryo transfers are healthy. However, there is a small chance of an ongoing pregnancy resulting in a baby born with a chromosome condition. For this reason, diagnostic testing by amniocentesis is usually recommended in an ongoing pregnancy. Amniocentesis directly tests cells from the amniotic fluid and can detect most chromosome conditions in the baby.

You may also choose to have standard screening tests, such as NIPT. While screening can provide additional information, it cannot reliably detect or exclude mosaicism in the baby

Your genetic counsellor will discuss these testing options with you in detail, both before transfer and again if pregnancy occurs, so you can make an informed decision about what feels right for you.

What are the next steps?

You may choose to keep your mosaic embryo in storage, proceed with transfer, or sensitively discard the embryo. Storage fees apply if the embryo remains in storage.

The decision whether or not to transfer a mosaic embryo is a personal one, and there is no right or wrong choice. A genetic counselling appointment can help you consider how your specific result fits with your goals and circumstances.

Mosaic embryo transfers require review by the No. 1 Genetics Department prior to transfer.

If you would like to meet with a genetic counsellor for more information or to discuss transferring your mosaic embryo, please contact the No. 1 Genetics Department to arrange a consultation on 03 9132 9609 or genetics@number1fertility.com.au.