



## PATIENT INFORMATION

### PGT-A mosaic result

#### What is a mosaic result?

Mosaic embryo results are common in preimplantation genetic testing for aneuploidy (PGT-A) (please see *Patient Information: PGT-A* for further information). The term *mosaic* or *mosaicism* refers to a mixture of two or more genetically different types of cell lines present in the same individual or embryo. In the context of screening embryos for chromosomes using PGT-A, mosaic results typically indicate some cells in an embryo biopsy sample have a 'normal' quantity of chromosomes, and some cells have an 'abnormal' quantity of chromosomes. Some mosaic results can indicate more than one chromosome error is present in an embryo.

Many mosaic embryos have potential for transfer and are thus retained in the N°1 Laboratory for consideration of future transfer. Couples who are considering transfer of a mosaic embryo are required to have a genetic counselling consultation with a N°1 Genetic Counsellor to discuss what the mosaic result means, the potential risks associated with mosaic embryo transfer and potential pregnancy.

#### How common are mosaic embryo results?

At N°1 Fertility, our genetic testing lab CooperGenomics reports that approximately 17% of PGT-A results are reported mosaic which is consistent with other IVF services globally.

#### What is the difference between 'low level' or 'high level' mosaic embryos?

Mosaic PGT-A results are reported as 'low level' or 'high level' for a given chromosome error. A low-level result indicates approximately 20-40% of the DNA tested from the embryo biopsy sample contains a chromosome abnormality. A high-level result indicates approximately 40-80% of the DNA tested contain a chromosome abnormality.

It is important to remember that PGT-A is a screening test, and an embryo biopsy takes a very small number of cells (approximately 3-5 cells). The levels of mosaicism reported are based on a very small sample size and may not be representative of the actual proportion of mosaicism in the whole embryo. This means the embryo may have a higher or lower proportion of abnormal cells than is reported.

#### Can I transfer my mosaic embryo?

Due to the limited evidence and research regarding mosaic embryos, the N°1 Fertility policy allows you to retain your mosaic embryos for consideration of future transfer, either at N°1 Fertility (subject to approval) or for export to another clinic.

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If you wish to consider transfer of a mosaic embryo, a consultation with the N°1 Genetics Department and/or Clinical Geneticist is required. Following this, the request may be reviewed by the Clinical Review Committee. Requests to transfer a mosaic embryo will either be approved or declined. This process must be completed prior to commencing a frozen embryo transfer (FET) cycle.

#### **What will happen if I transfer my mosaic embryo?**

The outcomes of mosaic embryo transfers remain largely unpredictable. There are case reports of successful pregnancies established following the transfer of a mosaic embryo, however it is important to know that a mosaic embryo has a higher chance of resulting in implantation failure, miscarriage, or pregnancies or children diagnosed with developmental problems or birth abnormalities. We encourage you to speak with the N°1 Genetics Department to arrange a consultation to discuss this in more detail.

#### **What testing is recommended after mosaic embryo transfer?**

If a pregnancy is achieved with a mosaic embryo, we recommend genetic testing in pregnancy. Specifically, we recommend considering a chorionic villus sample (CVS) and/or amniocentesis to determine if the chromosomally abnormal cells identified by PGT-A have persisted in the pregnancy.

*Chorionic villus sampling (CVS)* is an invasive diagnostic test performed at approximately 12 weeks gestation. This specialised test involves taking a small sample of placental tissue via ultrasound-guided needle. DNA analysis of a CVS sample can provide diagnostic information about the chromosome complement of the placenta. CVS procedures are performed by experienced specialist doctors, however there is approximately 1 in 500 risk of procedure-associated miscarriage. CVS may be limited in usefulness when a pregnancy is established following mosaic embryo transfer, as it samples cells from the placenta which may not be representative of the fetal cells.

*Amniocentesis* is an invasive diagnostic test performed at approximately 16 weeks gestation. This specialised test involves taking a small sample of amniotic fluid surrounding the baby via ultrasound-guided needle. DNA analysis of an amniocentesis sample can provide diagnostic information about the chromosome complement of the fetus. Amniocentesis procedures are performed by experienced specialist doctors, however there is approximately 1 in 1000 risk of procedure-associated miscarriage. Amniocentesis cells are the most reflective of the fetal chromosome complement and may be preferred rather than a CVS following mosaic embryo transfer.

These tests can be discussed further with the N°1 Genetics Department.

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### Why is my embryo mosaic?

Mosaic PGT-A results are common, and we are not aware of any factors such as diet or age, that influence the development of a mosaic embryo. Receiving a mosaic PGT-A result is not caused by something you have or have not done throughout your cycle or in your past. Current research suggests that these embryos likely begin as 'normal' (euploid, balanced), and through random error create a normal and an abnormal cell line that both divide and proliferate as the embryo grows. As mosaic embryos are believed to result from random errors, couples who have had a mosaic embryo PGT-A result reported in the past are *not* more likely to have this result in the future.

### What will happen to my mosaic embryo?

Victorian regulations require IVF clinics only retain embryos that may be intended for future use. If you do not have any intention of using your mosaic embryo(s) in the future and wish to discard this embryo(s), please inform the N°1 Genetics Department. Additional consent forms will be required to arrange this.

If you do not request to discard your mosaic embryos(s), it/they will remain in storage with N°1 Fertility. You will be responsible for paying the cost of embryo storage.

### What do people do with their mosaic embryos?

Consideration and transfer of a mosaic embryo is a personal decision. Some couples choose to transfer mosaic embryos and others choose not to. It depends on what feels right and comfortable for you to include or not include in your fertility journey. To support you as you consider transferring a mosaic embryo, you will have a consultation with a N°1 Genetic Counsellor to discuss your mosaic embryo(s) and outcomes in detail. Following the consultation, couples may choose to retain their mosaic embryo(s) for potential transfer in future, request to transfer their mosaic embryo, or discard their mosaic embryo.

### Questions?

If you have read this information and wish to discuss further, please contact the N°1 Genetics Department on 9132 9600 or email [genetics@number1fertility.com](mailto:genetics@number1fertility.com) to organise a time to speak to one of our Genetic Counsellors.

*The information provided above is intended for educational purposes only and should not be used as a substitute or replacement for medical advice received from a medical professional. It is important to discuss your individual circumstances and situation with your treating doctor.*

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