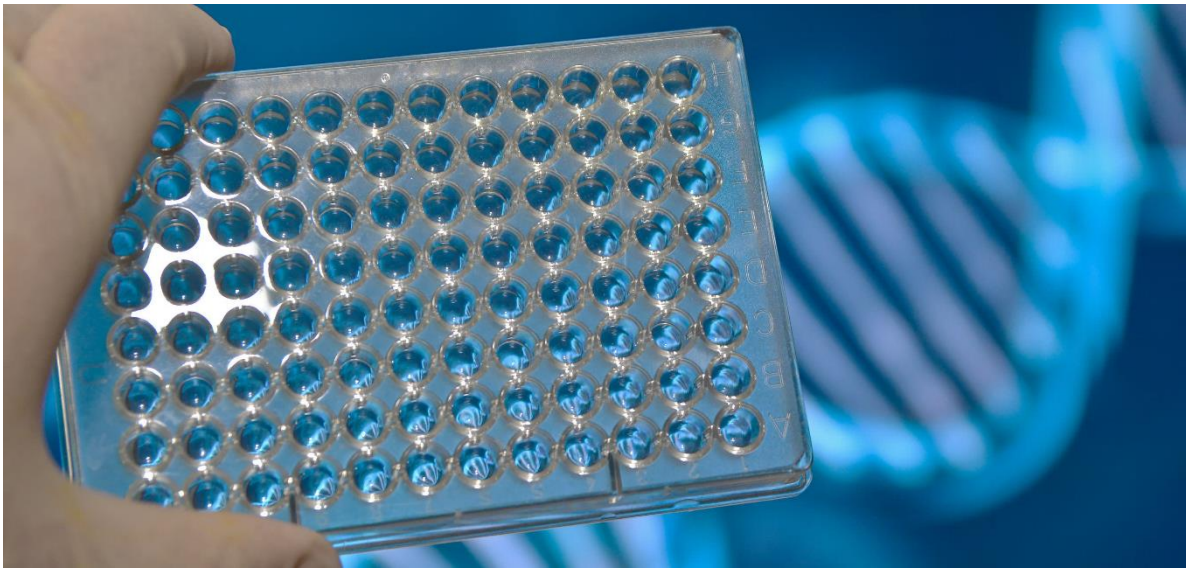
Preimplantation Genetic Testing for Monogenic/Single Gene Defects (PGT-M)

Many important factors come to mind when couples try to conceive. Most importantly, it is their desire to bear and care for a healthy child.

For men and women who have a genetic disorder, thankfully there are now technologies available to reduce the risk of passing it on to their children. Preimplantation genetic testing is an important breakthrough in fertility treatment.



What is PGT-M Testing?

Preimplantation genetic testing for monogenic/single gene defects (PGT-M) is a genetic test that screens embryos for the presence of a specific genetic mutation. PGT-M can be performed for many single gene disorders. PGT-M is done within an IVF treatment cycle. By avoiding transfer of affected embryos, patients can be sure that children resulting from IVF are free from the disease.

Who is PGT-M for?

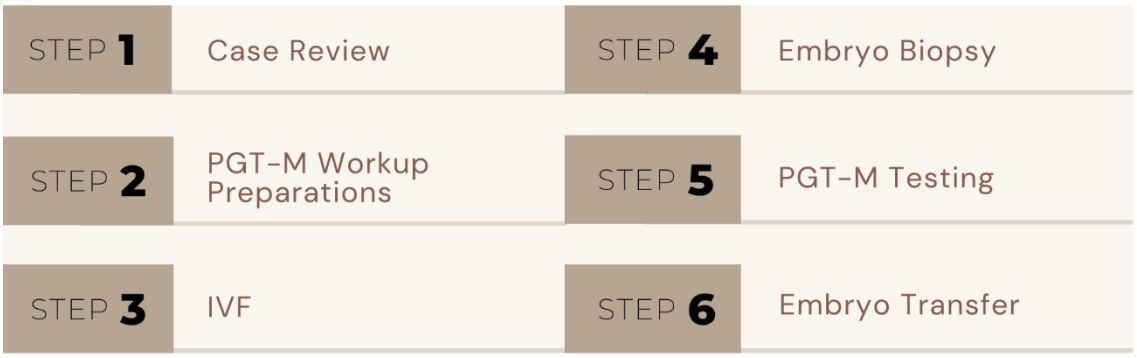
While PGT-M is not necessary for all couples, it is invaluable to those at higher risk of transmitting a genetic disease to their children [1]. You should consider if you and your partner are carriers of the same genetic disease or have had a child affected by a genetic disease.

The following is a list of the type of individuals who are possible candidates for PGT-M:

- Carriers of sex-linked genetic disorders
- Carriers of single-gene disorders
- Those with chromosomal disorder

Some examples of diseases which PGT-M can detect are thalassaemia, cystic fibrosis, Duchenne muscular dystrophy, Huntington's disease, haemophilia, fragile X syndrome and hereditary cancer syndromes such as BRCA 1 and 2 [2].

How is PGT-M performed?



Step 1: Case Review

The couple speaks to the fertility specialist on the need for PGT-M when trying for a child. A clinical diagnosis is made based on the couple's DNA analysis report. The couple then sees a genetic counsellor to discuss about their genetic condition, the risks involved and the implications of undergoing PGT-M as part of IVF treatment.

Step 2: PGT-M Workup Preparations

Once the PGT laboratory gives approval for the couple's case to proceed, they will then begin with workup preparations. Here, the couples' blood samples will be required. This step is crucial as PGT-M testing on embryos are done according to the workup preparations.

Step 3: IVF

The woman has her eggs retrieved and the eggs are fertilised with the sperm through ICSI. The embryos then develop over a course of 5-6 days in the laboratory.

Step 4: Biopsy

Good-quality embryos, now called 'blastocysts' are selected for biopsy. Biopsy is the step where a few cells of the blastocysts are gently removed. Once biopsy is complete, the blastocysts are immediately frozen.

Step 5: PGT-M Testing

Cells taken from the blastocysts are loaded in a tube and sent to a genetic laboratory for testing. Here, the laboratory uses advanced equipment to identify whether these cells contain the affected gene, which in turn informs about the blastocyst's status.

Step 6: Transfer

After the PGT-M results are obtained, normal unaffected blastocysts can be thawed and transferred into the uterus in a subsequent frozen cycle. On the other hand, embryos which carry the diseased genes are discarded upon the couple's consent.

Is embryo biopsy and PGT-M safe?

PGT-M is safe and effective in reducing likelihood of having children affected with these serious hereditary diseases. According to the PGT consortium, tens of thousands of PGT cycles have been reported from 1997-2013 resulting in the birth of many healthy babies [3]. The procedure does not appear to affect the embryo's ability to develop, the subsequent pregnancy and health of the child once born.

Where does PGT-M testing occur?

If you choose to undertake a PGT-M procedure, your embryos will be created and biopsied in our own laboratory before the cells are transferred to a PGT laboratory for testing.

Limitations of PGT-M testing

The following are several limitations associated with the use of PGT:

- PGT-M does not increase the overall chance of having a baby.
- PGT-M procedure is in addition to the costs of IVF.
- PGT-M could reveal that there are no normal embryos for transfer.
- PGT-M only screens for the selected genetic condition. There remains a possibility of other genetic diseases or disorders that goes undetected.

References

1. FAQ for AR Centres on PGT-M. Ministry of Heath, Singapore. https://www.moh.gov.sg/docs/librariesprovider5/licensing-terms-and-conditions/faqs-for-ar-centres-on-pgt-m-and-pgt-sr-rtcs.pdf

2. Approved Providers for PGT-M and PGT-SR. Ministry of Health, Singapore https://www.moh.gov.sg/resources-statistics/approved-providers-of-pre-implantation-genetic-testing-for-monogenic-single-gene-defects-and-chromosomal-structural-rearrangement-services

3. van Montfoort, A., Carvalho, F., Coonen, E., Kokkali, G., Moutou, C., Rubio, C., Goossens, V., & de Rycke, M. (2021). ESHRE PGT Consortium data collection XIX–XX: PGT analyses from 2016 to 2017. *Human Reproduction Open*, *2021*(3).