

# The pros and cons of pre-implantation genetic testing for aneuploidy (PGT-A)



**Pre-implantation genetic testing for aneuploidy (PGT-A)** is an “add on” used in IVF to help choose embryos with the right number of chromosomes. Depending on your circumstances, the technique will have different pros and cons.

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For example, if you’re a woman aged over 36, it can reduce the risk of miscarriage, but regardless of your age, it does not improve your overall chance of having a baby.

PGT-A is also known as **pre-implantation genetic screening (PGS)**. This brochure will help you understand what’s involved and whether it is worth paying for.

Here we describe the most common PGT-A technique. Some clinics are now offering a less invasive technique where, rather than removing cells from the embryo, they test the fluid that the embryo is grown in to determine if the embryo has the right number of chromosomes. Publications with information about how well this test performs are pending.

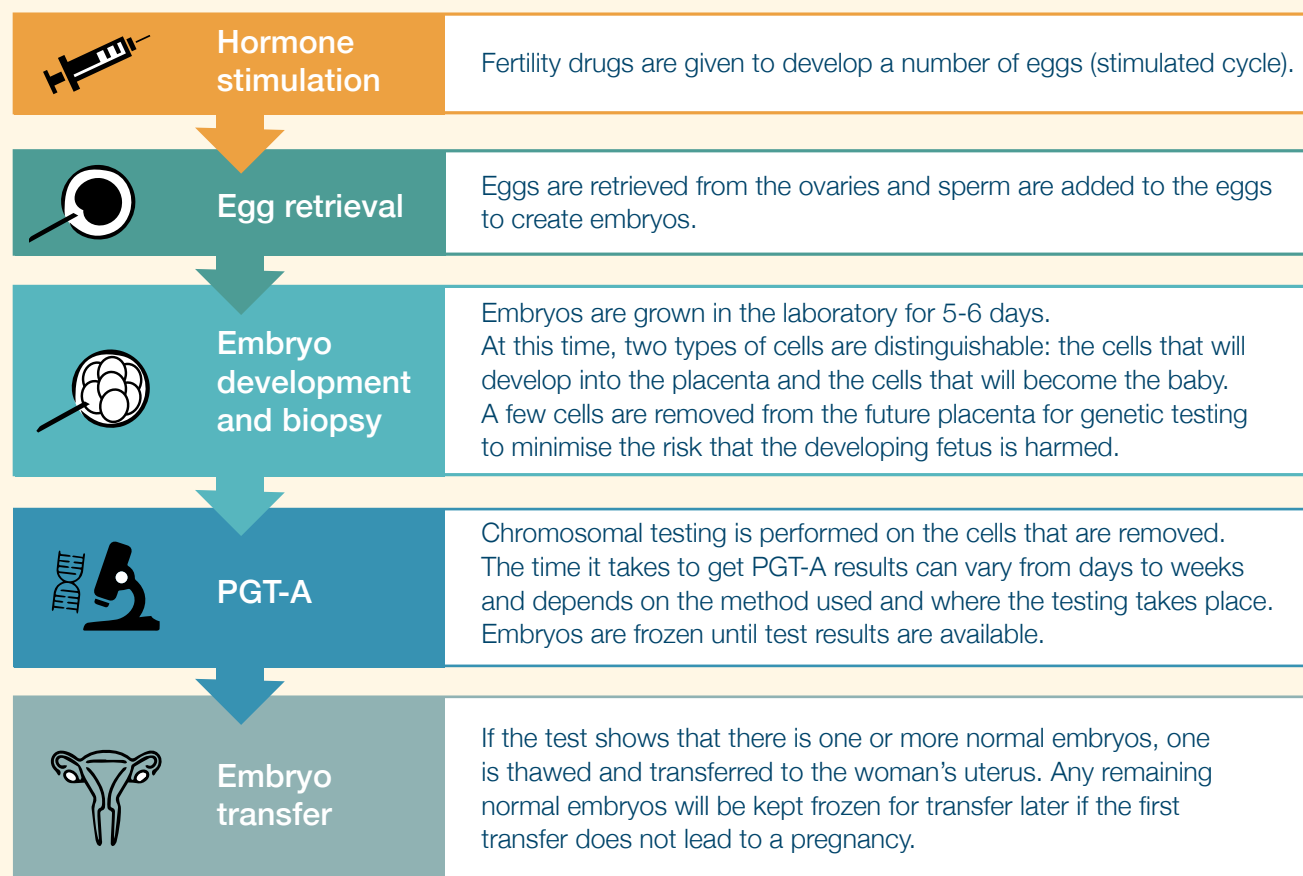
## Understanding the chromosomal makeup of embryos

Each cell in the human body contains packages of genes called **chromosomes**. Normal cells have 46 chromosomes arranged in 23 pairs (half from each parent). Females have two X chromosomes, and males have one X and one Y chromosome in all their cells.

Embryos are called **euploid** if they have the right number of chromosomes and **aneuploid** if they have extra or missing chromosomes. An aneuploid embryo is unlikely to implant and if it does implant, there is a high risk of miscarriage or serious health problems if a baby is born.

## How is PGT-A done?

**PGT-A adds one extra step to the IVF process. Here are the steps:**



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### Advantages and disadvantages

It is important to know that **PGT-A does not guarantee the birth of a healthy baby**. Women who become pregnant are advised to undergo prenatal testing (e.g. DNA testing or chorionic villus sampling) to confirm PGT-A results.

Here are some of the advantages and disadvantages of PGT-A. For more detailed information and to find out if PGT-A is for you, please speak to your doctor or genetic counsellor.

**Please note:** You cannot choose the sex of your baby through PGT-A. Sex selection is only allowed when an embryo is tested to avoid passing on a genetic condition which only affects boys or girls.

Advantages	Disadvantages
<ul style="list-style-type: none"> <li>PGT-A reduces the risk of having a child with a chromosomal abnormality.</li> <li>For women who have had unexplained miscarriages, PGT-A can reduce the risk of future miscarriages.</li> <li>PGT-A can reduce the risk of having to make difficult decisions about whether to terminate or continue a pregnancy where the fetus has a chromosomal abnormality.<sup>1</sup></li> <li>For women over the age of 36 PGT-A can reduce the risk of miscarriage and the number of embryo transfers to achieve a pregnancy.<sup>2,3</sup></li> </ul>	<ul style="list-style-type: none"> <li>PGT-A does not increase the overall chance of having a baby.<sup>4</sup></li> <li>PGT-A is expensive and is in addition to the costs of IVF. The cost of PGT-A is not covered by Medicare.</li> <li>Embryos may not survive the biopsy procedure.<sup>5</sup></li> <li>Due to technical challenges, there is a small chance that the test results may not reflect the true health of the embryo.</li> <li>Some embryos have a mixture of normal and abnormal cells. This is called <b>mosaicism</b>. This can cause a false positive or false negative PGT-A result.<sup>6</sup> <ul style="list-style-type: none"> <li><b>A false positive result</b> means that the few cells that are tested show abnormalities, while the remaining cells are chromosomally normal. Based on the test result, an embryo that may have led to the birth of a healthy baby may be discarded.</li> <li><b>A false negative result</b> means that the few cells that are tested are normal while the remaining cells are chromosomally abnormal. Based on the test result, a chromosomally abnormal embryo may be transferred.</li> </ul> </li> <li>Sometimes no embryo is suitable for transfer. Your doctor will discuss your results and options with you.</li> <li>Embryos may not survive the thawing process.</li> </ul>

#### References

- <sup>1</sup> Lamb, B., Johnson, E., Francis, L., Fagan, M., Riches, N., Wilson, A., Johnstone, E. (2018). *Pre-implantation genetic testing: decisional factors to accept or decline among in vitro fertilization patients*. Journal of Assisted Reproduction and Genetics, 35(9), 1605-1612.
- <sup>2</sup> Neal, S. A., Morin, S. J., Franasiak, J. M., Goodman, L. R., Juneau, C. R., Forman, E. J., Scott Jr, R. T. et al. (2018). *Preimplantation genetic testing for aneuploidy is cost effective, shortens treatment time, and reduces the risk of failed embryo transfer and clinical miscarriage*. Fertility and Sterility, 110(5), 896-904.
- <sup>3</sup> Lee, E., Chambers, G. M., Hale, L., Illingworth, P., & Wilton, L. (2018). *Assisted reproductive technology (ART) cumulative live birth rates following preimplantation genetic diagnosis for aneuploidy (PGD-A) or morphological assessment of embryos: A cohort analysis*. Australian & New Zealand Journal of Obstetrics & Gynaecology, 58(5), 525-532.
- <sup>4</sup> Homer HA. *Preimplantation genetic testing for aneuploidy (PGT-A): The biology, the technology and the clinical outcomes*. Aust N Z J Obstet Gynaecol. 2019;59(2):317-24.
- <sup>5</sup> Cimadomo, D., Capalbo, A., Ubaldi, F. M., Scarica, C., Palagiano, A., Canipari, R., & Rienzi, L. (2016). *The impact of biopsy on human embryo developmental potential during preimplantation genetic diagnosis*. BioMed research international, 2016.
- <sup>6</sup> Victor, A.R., Tyndall, J.C., Brake, A.J., Lepkowsky, L.T., et al. (2019). *One hundred mosaic embryos transferred prospectively in a single clinic: exploring when and why they result in healthy pregnancies*. Fertility and Sterility, 111(2):280-93.

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### Where can I go for more information?

Your fertility specialist and genetic counsellor can answer any questions you may have.

#### For more information regarding PGT-A

##### **VARTA booklet on PGT-A**

[www.varta.org.au/sites/varta/files/public/Images/Understanding%20the%20genetic%20health%20of%20embryos%20-%20preimplantation%20genetic%20testing%20for%20aneuploidy.pdf](http://www.varta.org.au/sites/varta/files/public/Images/Understanding%20the%20genetic%20health%20of%20embryos%20-%20preimplantation%20genetic%20testing%20for%20aneuploidy.pdf)

##### **What is PGT?**

<https://learn.genetics.utah.edu/content/disorders/whatispgt/>