

Pre-Implantation Genetic Testing for Aneuploidy (PGT-A)

What is PGT-A?

Embryos with an abnormal number of chromosomes (aneuploid embryos) are known to have a reduced chance of producing a successful pregnancy and may also result in a baby being born with a genetic condition. Until recently, the only way to determine whether or not an embryo was aneuploid was to wait until a pregnancy was established and then perform chronic villus sampling (CVS) or amniocentesis, to test the chromosomal material in the foetus. If an abnormal number of chromosomes are detected, parents face the difficult choice of whether to continue or terminate the pregnancy. PGT-A (formally known as PGS) is a new technology which helps to identify embryos that have an abnormal number of chromosomes at this very early stage, and as such, are not transferred.



How is PGT-A carried out?

The technique of PGT-A involves carrying out a biopsy on suitable embryos (those that have developed to the correct stage) and testing for any abnormalities in the number of chromosomes, in the hope to identify and transfer an embryo with the correct number of chromosomes. The biopsy is performed on suitable blastocysts (at either the day 5 or day 6 stage). The embryos are frozen whilst we obtain the genetic result and then transferred in a future cycle.

What is the evidence to support PGT-A?

There is some evidence to show that IVF success rates can be improved if embryos are tested for aneuploidy, with only those found to have the correct number of chromosomes transferred to the womb^{1,2}. It has also been reported that PGT-A significantly reduces the chance of having a pregnancy affected by certain genetic conditions¹. A recent multi-centred randomised clinical trial reported no significant improvement in pregnancy outcomes in all women after PGT-A treatment³. PGT-A is not yet considered to be a standard technique and consequently we strongly recommend that patients who become pregnant through PGT-A undergo prenatal testing using CVS or amniocentesis, to confirm whether or not the foetus has a correct number of chromosomes.

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Is PGT-A for me?

We may recommend PGT if:

- You are over 35 and have a higher risk of having a baby with a chromosome problem (such as Down's syndrome)
- You have a family history of chromosome problems
- You have a history of recurrent miscarriages
- You have had several unsuccessful treatments where embryos have been transferred
- Your sperm are known to be at high risk of having chromosome problems

Please Note: You are not obliged to undergo PGT-A, even if your doctor recommends it.

Are there any risks with PGT-A?

In some cases, it may be that embryos are too poor quality to undergo the biopsy and testing process. If embryos are suitable for biopsy, there is a risk of damage to the embryo through the biopsy procedure, however this risk is quoted to be less than 1%. Additionally, although current PGT-A techniques are mostly very accurate, the test may give the wrong result (i.e. it may miss an abnormality, or detect one that isn't there).

If both the procedure and testing are carried out successfully, research has shown that babies born after PGT-A show no increase in congenital abnormalities above the general rate for IVF children⁴.

What is the cost of PGT-A?

There will be an additional charge per treatment cycle for PGT-A so please discuss this with your doctor.

The Human Fertilisation and Embryology Authority (HFEA) and PGT-A:

PGT-A is regarded by the HFEA as a treatment 'add on'. For more information on treatment add-ons please refer to the HFEA traffic-light system on their website:

<https://www.hfea.gov.uk/treatments/treatment-add-ons/>

Please discuss the current HFEA traffic-light status for the use of PGT-A with your fertility specialist.

This leaflet can be made available in different formats on request. If you would like to make any suggestions or comments about the content of this leaflet, then please contact the Patient Experience Team on 0151 702 4353 or by email at pals@lwh.nhs.uk

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1 Sacchi L, Albani E, Cesana A, et al. Preimplantation genetic testing for aneuploidy improves clinical, gestational, and neo natal outcomes in advanced maternal age patients without compromising cumulative live-birth rate. *J Assist Reprod Genet* 2019;36:2493-2504.

2 Greco E, Litwicka K, Minasi MG, Cursio E, Greco PF, Barillari P. Preimplantation genetic testing: where we are today. *Int J Mol Sci* 2020;21:4381-4381

3 Munné S, Kaplan B, Frattarelli JL, et al. Preimplantation genetic testing for aneuploidy versus morphology as selection criteria for single frozen-thawed embryo transfer in good prognosis patients: a multicentre randomised clinical trial. *Fertil Steril* 2019; 112 (6), 1071-79

4 Kuiper, D., Bennema, A., la Bastide-van Gemert, S., Seggers, J., Schendelaar, P., Mastenbroek, S. et al. Developmental outcome of 9-year-old children born after PGS: follow-up of a randomized trial. *Hum Reprod*. 2018; 33: 147–155