SAMAA

FERTILIZATION CENTER

A guide to Preimplantation Genetic Testing (PGT)

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Preimplantation Genetic Testing (PGT)

Preimplantation genetic testing historically is divided into preimplantation genetic screening (PGS) or preimplantation genetic diagnosis (PGD). In 2017 the international glossary on infertility and fertility care revised the terminology. The new name for all tests is Preimplantation genetic testing (PGT). This includes:

i. PGT for aneuploidy (PGT-A) – previously PGS

PGT-A is offered to couples with two or more implantation failures following IVF embryo transfer, patients who have experienced recurrent miscarriages, and women aged 35 years and above. It is important to understand that ultimately it is the number and quality of the embryos that will determine whether you will benefit from performing PGS.

ii. PGT for single gene mutation (PGT-M) – previously PGD

PGT-M is a method of testing embryos produced during an IVF treatment cycle for a specific genetic disorder before they are transferred to the uterus and is now available for virtually all single gene disorders. The aim of PGT-M is to provide couples at high-risk of passing on an inherited disorder to their children with an improved chance of having an unaffected pregnancy.

iii. PGT for chromosomal structural rearrangements (PGT-SR) – previously PGS translocation.

PGT-SR is offered to couples who have been diagnosed as having an abnormal karyotype (the number and arrangement of the chromosomes within the cells). Either a Robertsonian or Reciprocal translocation may have been diagnosed or an inversion may have been detected. The presence of this re-arrangement will potentially make it difficult to conceive a healthy pregnancy naturally and may have already resulted in complete infertility or even recurrent miscarriage

Procedure:

The entire process of PGT consists of six (6) steps performed by scientists at different laboratories:

1) The first part is fertility treatment with intra cytoplasmic sperm injection (ICSI) by which the embryos are produced. This part occurs at the SFC.

2) The second part is the embryo biopsy. Embryo biopsy can be performed at either Day 3 of embryonic development (cleavage stage – blastomere biopsy) or Day 5 and/or Day 6 of development (blastocyst stage – trophectoderm biopsy*). This part also occurs at SFC by our own highly skilled embryo practitioners. **Currently only day 5/6 embryo biopsy is performed at most international clinics, as day 3 embryo biopsy has been shown to be deleterious to the further development of the embryo.*

3) After the cells are biopsied and placed in test tubes, the test tubes are transported by courier to an outside reference laboratory for genetic analysis.

4) The analysis of the cells is performed by an outside genetics' laboratory.

5) The results of the genetic analysis routinely take over 24 hours to be confirmed. Hence, all biopsied embryos on day 5/6 are vitrified (frozen) for future embryo transfer.

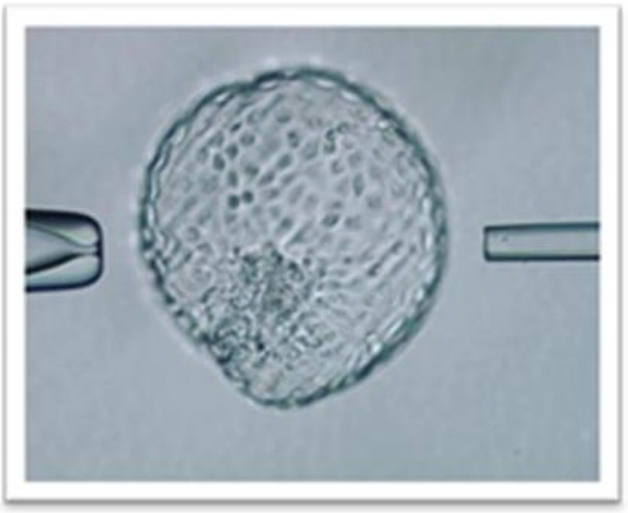
6) The final step involves the thawing and transfer of the embryos into the female patient which is performed by a physician at the SFC.

It is important to note that performing PGT could mean running a risk of no embryo transfer as it is possible that all embryos are classified as abnormal and therefore none will be found suitable for transfer. In the event of a diagnostic failure, you will be

advised by a member of the PGT team of the risks associated with the transfer of any undiagnosed embryos. The level of risk will vary from patient to patient. With respect to the embryo biopsy procedure there is a small risk that an embryo may become accidentally damaged during the procedure (less than 1%).

The phenomenon of mosaicism refers to the possibility of two chromosomally different cell lines co-existing within one embryo. Mosaic embryos can carry one or more cells with a different number of chromosomes to the other cells. This fact leads to a risk of misdiagnosis as we cannot analyze all the cells in the embryo. In the case of mosaicism, it is possible that a cell classified as normal by PGT analysis may belong to an aneuploid embryo and vice versa.

Regrettably it is possible for a miscarriage to occur even after a successful PGT cycle due to factors not related to the PGT procedure. Prenatal testing is always strongly advised following any type of PGT.



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