IN-VITRO FERTILIZATION (IVF) AND PRE-IMPLANTATION GENETIC TESTING (PGT)

LANKA HOSPITALS
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Inherited Genetic Disorders and Genetic Mutations

Genetic disorders are caused by mutations that are inherited from the parents and are present in an individual at birth such as Down syndrome, Fragile-X syndrome, Thalassemia, X or Y linked disorders, breast cancer and intestinal cancer. At the present, Pre-implantation Genetic Testing - PGT) has led to overcome these conditions and to have a healthy child.

What is Pre-implantation Genetic Testing (PGT)?

Pre-implantation Genetic Testing is a widely-used; an advanced technique that accommodates Next Generation Sequencing (NGS) to analyse for certain genetic abnormalities of the embryos, which are developed from the In-Vitro Fertilization (IVF) process.

Further, this is the only technique that is currently available to detect genetic abnormalities in the embryos prior to the conception.

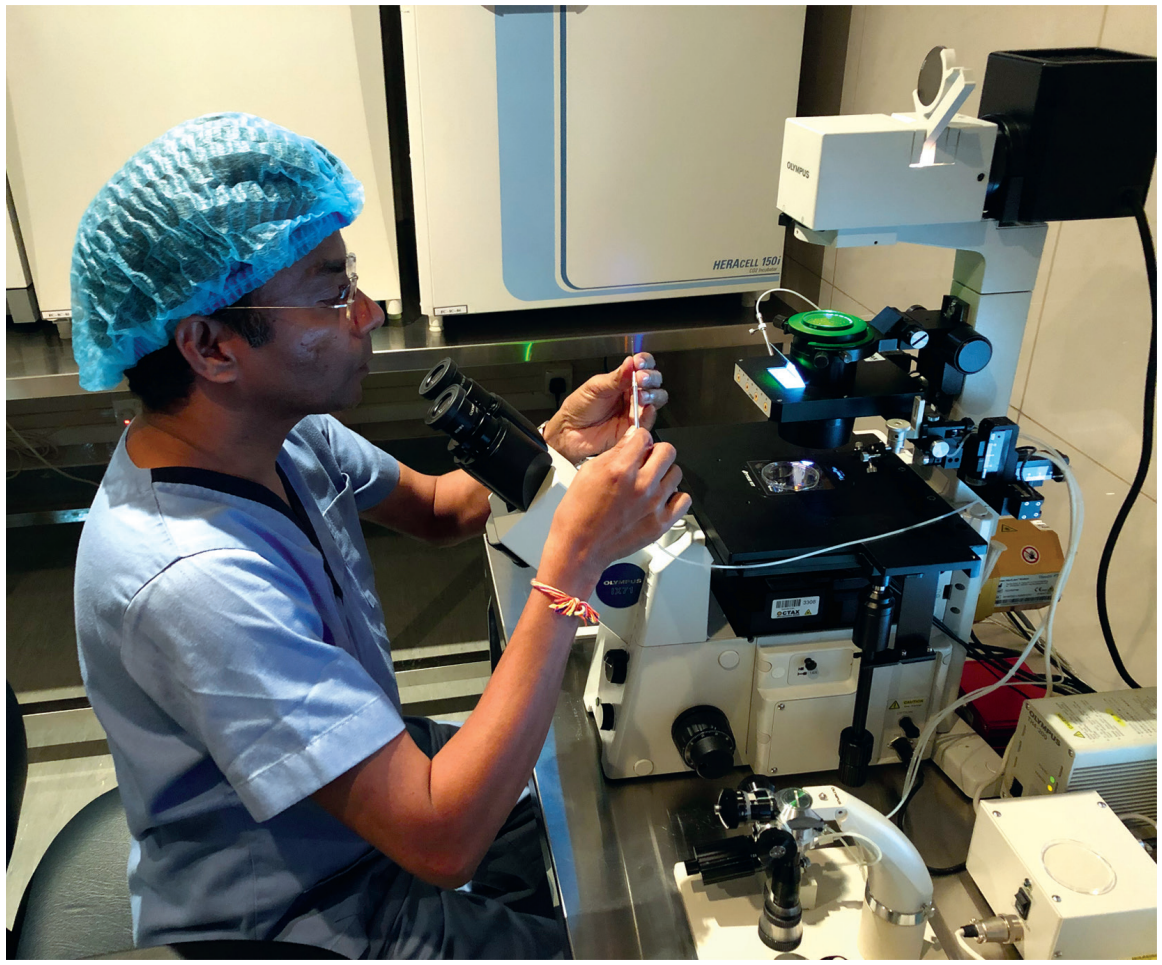
What are the different types of analysis of PGT?

There are three different types of analysis conducted in PGT based on the nature of the genetic mutations occur. They are PGT for Aneuploidies (PGT-A), PGT for monogenic/ single gene (PGT-M), and PGT for Chromosomal Structural Re-arrangement (PGT-SR).

PGT for Aneuploidies (PGT-A) is, testing the embryos by counting the 46 chromosomes of the embryonic cells to look for extra (trisomy) or missing (monosomy) chromosomes. This technique is used to select the embryos that are most likely to implant and result in a successful pregnancy. It also minimizes the chance of having a child with extra or missing chromosome such as Down syndrome, Edward syndrome etc.

The second method of PGT is PGT for monogenic/ singe gene (PGT-M), which is used to detect the embryos with single gene mutations that lead to develop diseases such as Thalassemia, Cystic Fibrosis, Huntington disease etc. This helps individuals or couples reduce their risk to have a child with a known inherited disorders caused by mutations of a single gene.

The third method is PGT for Chromosomal Structural Re-arrangements (PGT-SR). Babies are born with hereditary abnormalities due to mother's or father's or both having balanced chromosomal structural re-arrangements such as translocation or inversions. Using PGT-SR minimizes the risk of having a pregnancy or child with an unbalanced structural abnormality, which involve extra or missing genetic material and typically results in a pregnancy loss.



What are the other benefits of PGT?

Initially, PGT was mainly employed to increase the chance of conception of In-Vitro Fertilization (IVF). However, studies indicate that the women who are over 35 years have a higher risk of conceiving children with genetic disorders such as Down syndrome. Thus, PGT is beneficial for such community to have a healthy child.

How could a couple undertake PGT?

A couple can be benefited by PGT only if they are undertaking IVF treatments. Because, the embryos can be analysed for a certain genetic abnormalities only if they are developed under the laboratory conditions (outside of the body). Removal of cells is performed from embryos created through the IVF process. The embryos that selected through PGT are most likely to implant in the uterus and leading to a healthy ongoing pregnancy.

What are the steps in PGT process?

A cell from a Day 3 embryo (6-8 cell embryo) or number of cells from the trophoblast of a Day 5 embryo (Blastocyst) is extracted (embryo biopsy), and the cell sample is subjected to genetic analysis. The analysis of the sample is carried out using the Next Generation Sequencing (NGS) in a Genetic Laboratory. Usually a result of the biopsy receives within 14 days time. Until such time, all the developed embryos are vitrified. The biopsy report will be mentioned that if there is any embryo carries certain genetic mutation. The embryos which are normal genetic materials (euploid) will be selected and transferred to the uterus following a Frozen Embryo Replacement (FER) procedure.

What is In-Vitro Fertilization (IVF)?

IVF is a similar process, which takes place in fallopian tubes during the natural conception but performed under laboratory-controlled conditions. The most advanced method of IVF is ICSI (Intra-Cytoplasmic Sperm Injection) technology. ICSI is injecting a sperm directly into a mature egg using a micromanipulator. Hormone injections are administered over a pre-determined period on a daily basis, which commence from 2nd day of the menstruation to induce eggs during IVF procedure. The injections are self-injectable pens and can be administered by own or from the family physician. Therefore, it minimizes the number of visits to the hospital and helps conveniently carry out other day to day work. Further, the extraction of eggs (Oocyte Recovery-OR) is a day-care procedure and also non-invasive surgical procedure. Following the extraction of eggs, ICSI is performed to fertilize the eggs with male partner's sperms. Furthermore, when there are significant male infertility factors such as poor morphology and high fragmentations of sperms, IMSI (Intra-cytoplasmic Morphologically selected Sperm Injection) is used. IMSI is a variation of ICSI, which uses higher magnification that allows the embryologist to look at the sperm head, neck and tail in details and to select a good-quality sperm. Therefore, IMSI helps achieving highest success rate. Then after, two of the developed embryos by Day 3 or Day 5 (Blastocyst) are transferred in to the uterus using a special catheter (ET catheter). The transferring of the embryos to the uterus can be viewed by an Ultra Sound Scan. All the excess, good-quality embryos are frozen (vitrification) and stored up to 5 years for later usage when the couple needed a second child.

Are there facilities available for PGT in Sri Lanka?

Lanka Hospitals Fertility Centre is committed to offer most advanced technologies, which enable its patients to undergo IVF treatment with most appropriate/informed embryo selection and have now equipped with state-of-the-art high technology laser-assisted system to perform embryo biopsy for PGT. Moreover, it has employed an internationally qualified and trained embryologist and other laboratory staff with the knowledge in fertility and genetics.

The Lanka Hospitals Fertility Centre has currently reported over 1300 successful IVF deliveries and over 100 ongoing pregnancies. Reaching another milestone, the Lanka Hospitals Fertility Centre intends to provide world-class heath care service by facilitating PGT for the couples seeking IVF treatments and giving the opportunity to get transferred embryos without genetic abnormalities.

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