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Neuberg DIAGNOSTICS | CENTER FOR GENOMIC MEDICINE

Phone : +91-6357244307, 079-61618111
Email : contact@ncgmglobal.com
Website : www.ncgmglobal.com

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PGT

PRE-IMPLANTATION GENETIC TESTING

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

PRE-IMPLANTATION GENETIC TESTING (PGT)

Preimplantation Genetic Testing (PGT) is an early form of genetic testing allowing transfer of genetically normal embryos and has become an integral part of Assisted Reproductive Technology (ART) procedures.

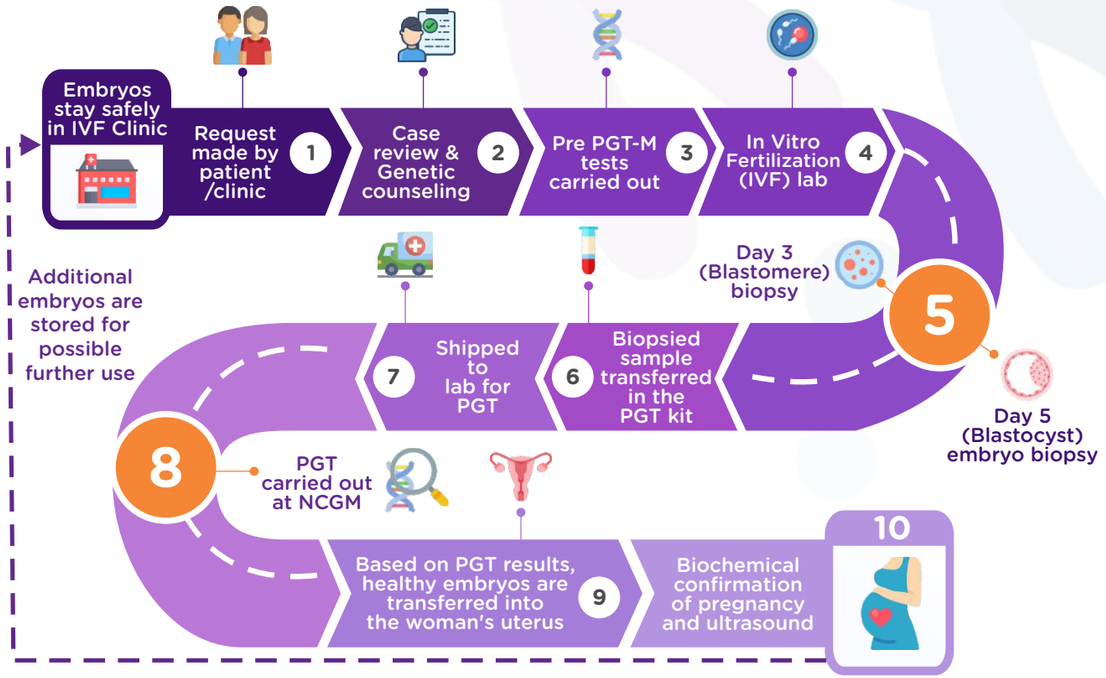
Identifying embryos with extra or missing chromosomes is important when selecting embryos for implantation for the couples who opt for in vitro fertilization (IVF). PGT helps screening embryos with aneuploidies and single gene disorders before implantation. The test helps identify genetic abnormalities that may lead to a genetic disorder during pregnancy or after the child's birth.

Types of PGT	PGT-A Pre-Implantation Genetic Testing for Aneuploidy	PGT-SR Pre-Implantation Genetic Testing for Structural Rearrangements	PGT-M Pre-Implantation Genetic Testing for Monogenic Disorders
Tests What?	Numerical chromosomal abnormalities across all 24 chromosomes (22 autosomes and 2 sex chromosomes	Specific imbalances arising from parental chromosomal rearrangements as well as other numerical or structural abnormalities across all 24 chromosomes	Specific monogenic disorders (autosomal recessive/ autosomal dominant/ X linked)
For Whom?	• Advanced maternal age (> 35 yrs) • Bad obstetric history • Implantation failure • Severe male factor infertility	• Chromosomal rearrangement like inversion, reciprocal translocation or Robertsonian translocation	• Previous child with a genetic disorder • Carrier for lethal autosomal recessive / autosomal dominant / X linked conditions

HOW IS IT DONE?

To perform PGT, few cells are biopsied from the day 5 embryo using a high-powered lasermicroscope by the embryologist. It is then sent to NCGM to screen for specific genetic disorder and/or numerical aberrations and/or structural rearrangements using Next Generation Sequencing (NGS).Based on the PGT report, embryos are selected for the transfer.

WORKFLOW



Advantages of PGS by NGS over other screening approaches

- Partial chromosomal gains and losses can be detected more precisely
- Aneuploidy and segmental imbalances can be screened at the same time
- Provides more accurate detection of mosaicism
- Offers reduced costs and enhanced precision. It allows parallel analysis for multiple embryos for a single patient.

PGT-A?

- Leads to higher implantation rates and improved IVF outcomes
- Reduces the number of IVF cycles required to achieve a successful pregnancy
- Increases success rates for single embryo transfer
- Reduces the likelihood of miscarriage due to Aneuploidies
- Increases reproductive success rates in women above 35 years
- More certainty in transferring a single “unaffected” embryo

PGT-SR?

- A structural rearrangement is an aberration in the normal chromosomal rearrangement and can manifest as an inversion, duplication, deletion or a translocation.
- 1 in 200 individuals is a carrier of a balanced chromosome rearrangement
- PGT-SR, or pre implantation genetic testing for chromosomal structural rearrangements, is a genetic test available for couples who are carriers of chromosome rearrangements.

  Though carriers of balanced chromosome rearrangements are typically healthy, they may pass down these rearrangements to their progeny, resulting in reduced viability of the embryos or potential for children with physical and/or intellectual disability.
- PGT-SR can help identify embryos with the correct amount of chromosomal material that are most likely to lead to a successful pregnancy and healthy live birth.

PGT-M?

This is carried out when one or both genetic parents have a known genetic abnormality or are carriers for a genetic disorder which increases the risk of transmitting the disease to their children. For every PGT-M request, prior case discussion with our Genetic Counsellors is mandatory.

PRE-REQUISITES FOR PGT

A Pre-PGT work-up is an essential requisite for PGT-M

- Female and male reproductive history, gynecological and fertility status
- Reports on health problems of female and male partners that may affect genetic diagnosis, or the outcome of IVF and pregnancy (when appropriate).
- For PGT-M and PGT-SR: a genetic counselling report together with full pedigree and family data and prior case-discussion is mandatory.
- Results of any genetic testing, karyotype or any other specific testing of the index patient, spouse or partner, children or other family members diagnosed with a genetic disorder has to be provided.

LIMITATIONS OF PGT

- Normal PGT results do not eliminate the risk for the embryo(s) to have a genetic condition
- Every pregnant woman has a background risk of approximately 3-5% to have a child with a genetic condition or birth defect. PGT can only "screen" for chromosomal abnormalities (by PGT-A or PGT-SR) or test for a specific known genetic condition (by PGT-M). There is no single genetic test that can eliminate this risk or identify all diseases or birth defects. Hence a normal PGT result does not guarantee a normal child
- The results of PGT are highly accurate; however, it is still considered as a screening test. This means that false positives and false negatives can occur. If you transfer an embryo that has been tested by PGT, it is still recommended that you consider confirming the normal results through diagnostic prenatal testing such as chorionic villus sampling (CVS) or amniocentesis

Your referring physician or a genetic counselor can review these options with you in more detail, as there are risks and limitations for each test and that should be considered carefully.

REFERENCES

- ESHRE PGT Consortium good practice recommendations for the organisation of PGT, Human Reproduction Open, Volume 2020, Issue 3, 2020, hoaa021, https://doi.org/10.1093/hropen/hoaa021

Your PARTNERS in HEALTH

DR. SHIVA MURARKA
Senior Scientist
(Molecular Genetics)
PhD Reproductive Sciences
shiva.murarka@supratechlabs.com

DR. UDHAYA KOTECHA
Clinical Geneticist
M.D. Pediatrics,
Fellowship in Medical Genetics
udhaya.kotecha@supratechlabs.com

DR. SANDIP SHAH
Consultant Pathologist, Lab Director
M.D (Pathology & Bacteriology)
drsandip@neubergdiagnostics.com