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Abstract Topic:- Molecular and cytogenetic diagnostics

Abstract Title:- Testing for unbalanced translocation in embryo biopsy samples: Selective transfer of embryos with normal karyotype

Presenting author name :- Rashmi Rasalkar

Presenting author institute:- Medgenome Labs

Co-authors name:- Dr. Avinash Pradhan, Deepika Jena, Divya Jyothi Malla, Pranjali Thorat, Dr. Venkataswamy, Dr. Priya Kadam, Dr. Ramprasad VL, Dr. Sakthivel Murugan

Co-authors institute:-Medgenome labs

Aims:-The PGT-SR (Preimplantation Genetic Testing for Structural Rearrangements) is a specialized genetic test used as an adjunct to assisted reproductive technology (ART) to detect specific chromosomal rearrangements that may be inherited. This test is designed for individuals or couples who carry structural rearrangements in their chromosomes (unbalanced reciprocal translocation). These translocations are more prevalent chromosomal abnormalities in humans. Typically, carriers of this condition appear phenotypically normal, but they are at elevated risk of infertility, recurrent miscarriages, or having children with genetic disorders.

The purpose of PGT-SR is to examine embryos created through in vitro fertilization (IVF) for the selection of the embryos with normal karyotype. This testing helps increase the likelihood of a successful pregnancy by reducing the chances of miscarriage or the birth of a child with chromosomal abnormalities.

Methods:- A total of 36 families (113 PGT-SR (unbalanced translocation)) cases were studied in Medgenome lab from 2021- 2023. Each case was studied based on the karyotype report. Day 5 Trophectoderm biopsy samples (6-9 cells) were processed for whole genome amplification followed by library preparation (Kit based protocol) and next generation sequencing.

Results:- NGS data provided 0.5- 6 million reads and average quality score for all the samples was >0.03. Data was analysed using lab developed CNV calling software. Out of 113 Day 5 biopsy samples 46 samples were identified with normal karyotype and 67 samples had abnormal karyotype, the abnormal karyotype was in concordant with the translocation karyotype of each family (36 families).

Conclusions:- NGS based PGT-SR enables accurate results in identifying unbalanced reciprocal translocation. In addition, this test also provides information on aneuploidies and mosaic conditions (30%) for all the 23 pairs of chromosomes. In conclusion, this method allows patients to prioritize the transfer of euploid embryos.

Keywords:- Structural Rearrangement, Translocation, unbalanced translocation, copy number variation (CNV).