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Abstract Title:- Preimplantation Genetic Testing for Monogenic Disorders (PGT-M): An approach by SNP Array Based Assay

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Aims:-Preimplantation Genetic Testing for Monogenic Disorders (PGT-M) is a cutting-edge genetic screening technique that holds immense promise in the field of assisted reproduction. PGT-M is designed to identify and select embryos free from specific monogenic disorders before implantation, ensuring a higher likelihood of healthy pregnancies and reducing the risk of transmitting these genetic conditions to future generations. Novel advances in molecular genetics and the development of powerful sequencing technologies, has allowed the successful application of PGT-M to several hundred disorders caused by single gene mutation. The PCR improvements together with the increasing use of next-generation sequencing (NGS), whole genome amplification (WGA), comparative genomic hybridization (CGH) arrays and single nucleotide polymorphism (SNP) arrays have increased the PGT-M disease diagnostic spectrum and application. Importantly, next generation sequencing has also facilitated the combination of PGT-M with testing for monogenic disorders. Awareness of the availability of PGT-M, and appreciation of the increasing efficacy of this important reproductive strategy, will help the medical community and society at large to avail themselves of these advanced techniques to decrease the burden of genetic disease.

Methods:- Before starting a clinical cycle, extensive genetic and reproductive counseling is provided to the prospective parent(s). For Pre-PGT-M workup it usually requires blood samples from the couple and reference sample (sibling or grandparents) along with carrier testing reports for considering the PGT-M request. After completion of successful Pre-PGT-M workup, test was performed on embryo biopsy samples which were subjected to whole genome amplification (WGA) followed by array based assay. In this study the retrospective review involved data from 2022 to 2023 from 33 PGT-M patients, from whom a total of 71 blastocysts were screened for different monogenic disorders.

Results:- The objective of array based assay is to identify informative SNPs, inherited from the mother or father or both and it is used to phase the SNPs in the embryo against the alleles of the reference. It creates a complete map of the chromosomes (haploblocks) inherited by the embryo for accurate assessment of the presence of severe single-gene disorders from a single embryonic cell. Of the 33 PGT-M patients total 31 embryo biopsy samples (43%) were recommended and 40 embryo biopsy samples (56%) were not recommended for implantation based on PGT-M results.

Conclusions:- Introduction of Array baed assay for screening of monogenic disorders making PGT-M a well-established, accurate, and safe clinical procedure to avoid the transfer of affected embryos with serious genetic disorder.

Keywords:- PGT-M, Single Nucleotide Polymorphism, WGA, NGS