

Abstract ID:- 136

Abstract Topic:- Genetic counselling

Abstract Title:- Preventive Genetics in Reproductive Decision-Making: A Case Study of Preimplantation Genetic Testing for Hypomyelinating Leukodystrophy-18.

Presenting author name :- Naitika Bhavsar

Presenting author institute:- Anderson Diagnostic Services Private Limited

Aims:-This case study presents the genetic counseling and reproductive journey of a non-consanguineous endogamous couple who sought guidance after their first-born female child was diagnosed with a rare neurological disorder known as Hypomyelinating leukodystrophy-18. The index child exhibited symptoms including nystagmus, motor difficulties, sensory issues, spasticity, and global developmental delay. Trio Whole Exome Sequencing revealed a homozygous likely pathogenic variant c.565A>G in the DEGS1 gene in the index and both partners were identified as heterozygous carriers for the same DEGS1 gene variant.

Methods:- Given the autosomal recessive nature of the disorder, the couple was concerned about the recurrence risk and their desire to have a healthy child. Genetic counselling provided a comprehensive understanding of the 25% probability that their future offspring could inherit two abnormal genes and be affected by the condition. In light of this information, the couple opted for Preimplantation Genetic Testing for Monogenic disorder in embryo's derived through Assisted Reproductive Technology to identify and prevent Hypomyelinating Leukodystrophy-18 in their next child.

Results:- Preimplantation Genetic Testing for Aneuploidies along with Preimplantation Genetic Testing for c.565A>G variant in DEGS1 gene resulted in two euploid embryos, with one as homozygous wild type and the other being a heterozygous carrier for the c.565A>G variant in DEGS1 gene. Hereafter, both embryos were transferred, resulting in the birth of healthy twins.

Conclusions:- This case underscores the importance of genetic counselling, advanced reproductive technologies, and preimplantation genetic testing in enabling couples at risk of transmitting autosomal recessive disorders to have healthy offspring while minimizing the recurrence risk of the disease.

Keywords:- Genetics, Genetic counselling, Preimplantation genetic testing, rare diseases, preventive genetics.