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Abstract Topic:- Genetic counselling

Abstract Title:- Unraveling the Genetic Basis of Recurrent Hydatidiform Mole: Significance for Genetic Counseling, Diagnosis, and Treatment Approaches.

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Aims:- Recurrent hydatidiform mole (RHM) is a rare condition characterized by repetitive molar pregnancies, presenting significant challenges for both affected individuals and healthcare providers.

Methods:- We present a case report involving a 26-year-old patient with a history of recurrent molar pregnancies. Genetic Counseling was provided and Whole Exome Sequencing was advised for the couple. We identified homozygous pathogenic variant c.2471+1 G>A in the NLRP7 gene in female partner and heterozygous pathogenic variant c.2471+1 G>A in the NLRP7 gene in male partner causing Recurrent hydatidiform mole-1 (AR, OMIM#231090), providing valuable insights into the genetic basis of RHM.

Results:- Our findings unveil the possibility of categorizing RHM into two distinct groups, primarily based on familial history and genetic predisposition. Patients with a positive family history or RHM often experience biparental complete moles, while those without familial recurrence predominantly present with androgenetic complete hydatidiform moles. Genetic testing such as exome sequencing, have proven to be an indispensable tools in diagnosing and differentiating these subtypes, offering critical information for patient prognosis and family planning. This can help manage the condition, as women with androgenetic complete moles who undergo in-vitro fertilization (IVF) and pre-gestational diagnosis may have normal pregnancies, lowering the risk of further complete moles. Conversely, women with RHM might consider IVF using a donor egg to achieve a viable pregnancy.

Conclusions:- Recurrent hydatidiform mole is a challenging condition with profound emotional and medical consequences. This case unravels the need of exome sequencing and the pivotal role of Genetic Testing and Genetic Counseling. By doing so, we aim to contribute to a deeper understanding of RHM, ultimately guiding more informed decision-making and management strategies for individuals with this rare reproductive disorder.

Keywords:- Genetic Counseling; Reproductive Genetics, Recurrent Hydatidiform Mole, Exome Sequencing