

Abstract ID:- 177

Abstract Topic:- Genetic counselling

Abstract Title:- Donor carrier screening as a crucial step in ART conception with donor gamete: A case of Spinal Muscular Atrophy (SMA)

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Aims:-To emphasize the importance of carrier screening (for atleast common disorders) in gamete donor(s), thereby reducing the risk of genetic condition in the offspring(s). Here, we present a case that depicts the importance of donor carrier screening.

Methods:- A non-consanguineous, endogamous couple were referred for Genetic Counselling as their first-born (conceived through ART conception with donor oocyte) twin babies (male and female) succumbed to Spinal Muscular Atrophy (SMA).

Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication analysis by MLPA , was performed for the female baby followed by the male partner.

Results:- Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication analysis by MLPA in female baby was indicative of pathogenic homozygous deletion of exons 7 and 8 in SMN1 gene, causative of SMA. Further testing in the male partner revealed heterozygous deletion of exons 7 and 8 in SMN1 gene, establishing the carrier status.

Conclusions:- Carrier screening for gamete donors will increase preconception and prenatal care. Guidelines for screening gamete donors have been published by the American Society for Reproductive Medicine (ASRM). According to ASRM, screening for cystic fibrosis, spinal muscular atrophy, and thalassemia/hemoglobinopathy carrier status should be performed on all oocyte and sperm donors. Additionally, screening for fragile X can be performed on the oocyte donors. Screening of gamete donors is capable of reducing the incidence of genetic conditions in the offspring. In all of the scenarios considered, genetic counseling (pre-test and post-test) will be of central importance and enables the couple to take informed decisions. In present times, genetic counseling can be considered one of the most effective means to bring awareness on the possibilities of preventing common genetic disorders by educating/making the patient aware of their genetic risks.

Keywords:- Spinal Muscular Atrophy, gamete donor, carrier screening, genetic counseling, genetic risk