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Abstract Topic: - Prenatal, perinatal and developmental genetics

Abstract Title: - Laboratory experience of fetal RHD typing with NIPT in Indian population

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Aims: - The prevalence of RhD negative population can vary depending on the ethnicity. It is considered 1-3% for the Asian population and ~5% of the Indian population. Hemolytic diseases in fetus and new-born (HDFN) can be caused due to blood antigen (Rh antigen) incompatibility between a RhD negative pregnant mother and RhD positive fetus. However, not all Rh-negative pregnant woman will carry a Rh-positive child. An effective screening modality for detecting the status of RhD status in the fetus and implementing the anti-D only in RhD positive pregnancy will reduce auxiliary risks of anti-D administration and may aid in better pregnancy preparedness. There are limitations to existing tests of prenatal RhD testing due to its invasive nature or lesser sensitivity. Genetic tests can be more accurate in establishing the phenotypic status. Newer techniques such as cell-free fetal DNA (cff-DNA) testing can determine the genotype of the fetus from the circulating fetal DNA in the maternal blood.

Methods: - In this study we describe the laboratory experience of offering cff-DNA for fetal RhD testing in Indian population using real time PCR technique with sequence specific primers (SSP) to detect the presence of three exons – Exon 5,7, and 10 of the RHD gene. We describe the clinical outcome observed and its implication in genetic counselling for this test.

Results: - : Our laboratory experience over the past year shows fetal RhD gene was present in 87.70% of the samples, 9.84% showed absence of RHD gene and 2.46% were reported as inconclusive.

Conclusions: - With non-invasive prenatal testing, this screening can be implemented as early as the first trimester or early second trimester of pregnancy. It will reduce the redundancy in administration of anti-D immunoglobulin and reduce its adverse effects.

Keywords: - : Our laboratory experience over the past year shows fetal RhD gene was present in 87.70% of the samples, 9.84% showed absence of RHD gene and 2.46% were reported as inconclusive.