Abstract ID: - 75

Abstract Topic: - Clinical Genetics

Abstract Title: - Recurrent Pregnancy Loss associated genetic anomalies – case series from eastern India.

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Aims: - Idiopathic or enigmatic Recurrent Pregnancy Loss (RPL) is cases where clinical diagnosis remains helpless to find out any specific clinically known causal factor(s). Chromosomal heteromorphies like altered heterochromatin and/or satellite content as well as chromosomal aberrations like translocation, deletion and inversions have remained associated with many diseases along with other nucleotide variations. The present study was done with the objective to study prevalence of chromosomal and genetic anomalies with idiopathic RPL.

Methods: - Patients with history of idiopathic RPL were subjects of this study. Ethical clearance from the IRB was taken. Written informed consents were also taken from the subjects before inclusion of their data in the study. No personal information is being shared upon. Subjects were clinically assessed first for any relevant diagnosis. Patients having idiopathic situations were subjected to karyotype analysis as well as NGS test.

Results: - Idiopathic RPL cases were found to carry 9qh+ as predominant heteromorphy followed by 21ps+, 15ps+, 14ps+ and others. Heteromorphies were significantly higher in females than males except for 14ps+. The chromosomal aberrations have been found in few cases and these include deletion, inversion and translocation (both Reciprocal and Robertsonian) with two Novel Translocations t(14;22)(q34;q13) and t(2;13)(p23;q14) has also been seen. Nucleotide variations were also found but with much lesser frequency.

Conclusions: - Prevalence of chromosomal anomalies was found to be more than 10% among RPL cases with yet unknown molecular mechanism of damage. It has also been observed that heteromorphies in chromosomes are always found to be very closely situated to the centromeric regions of the concerned chromosomes. Therefore, chromosomal heteromorphies may play significant roles in cell divisions.

Keywords: - Idiopathic RPL cases were found to carry 9qh+ as predominant heteromorphy followed by 21ps+, 15ps+, 14ps+ and others. Heteromorphies were significantly higher in females than males except for 14ps+. The chromosomal aberrations have been found in few cases and these include deletion, inversion and translocation (both Reciprocal and Robertsonian) with two Novel Translocations t(14;22)(q34;q13) and t(2;13)(p23;q14) has also been seen. Nucleotide variations were also found but with much lesser frequency.