Abstract ID: - 38

Abstract Topic: - Molecular and cytogenetic diagnostics

Abstract Title: - High Prevalence of Acro ps+/- of D/G group of chromosomes 13ps+/-, 14ps+/-, 15ps+/-, 21ps+/-, 22ps+/- Chromosome Polymorphisms (CPM), An Evaluation of Genetic Factor in 1400 Recurrent Pregnancy Losses (RPL) patients and reassessing CPM in 21st Century as

Presenting author name: - Ashish Fauzdar

Presenting author institute: - Redcliffe Labs

Co-authors name: - , , , , , , , ,

Co-authors institute: - , , , , , , , ,

Aims: - It's has been estimated that 70% of all all-human conceptions fails to complete full term of which 15-20% of clinically recognized pregnancies ends up as pregnancy loss or are spontaneously aborted before 20 weeks of gestation age. Furthermore 5% of these women experiences two consecutive miscarriages and 1-2% of them report with two or more failed clinical pregnancies with diagnosis of recurrent pregnancy losses (RPL) reported worldwide. As per official communication the current burden for recurrent miscarriages in India is quite high is around 7.4%. The purpose of the study was to investigate the chromosome abnormalities and possible association of chromosome polymorphism in recurrent pregnancy loss patients for further clinical management of patients.

Methods: - A single centre case-control retrospective study including 1400 individuals with 700 couples with an established clinical diagnosis of recurrent pregnancy to rule out chromosome abnormalities by doing chromosome preparation done from peripheral blood lymphocyte stimulated culture for both the partners through conventional cytogenetics culture techniques using the standard protocols.

Results: - In total 21/700 (3%) couples showed chromosome abnormalities and chromosome polymorphisms in 471/1400 (33.7%) individuals. After analysis of different types of polymorphic variations in the study and the control group, the prevalence of acro ps+/- polymorphisms involving D/G groups of chromosomes was observed to be significantly higher in the study group with 13ps+/-, 14ps+/-, 15ps+/-, 21ps+/-, 22ps+/- observed in 23.5 % (330/1400) in the study group and 15.8% (58/366) in the control group $p < 0.005$. Among the acro ps+/- the prevalence of 22ps+ subtype polymorphism was significantly higher with the odd ratio OR (95% CI)- 2.35 (1.245-4.434).

Conclusions: - The presence of normal variant should be interpreted cautiously in the patient with recurrent pregnancy losses as they may play a significant role in prognosis and treatment. The incidence 21ps+ chromosomal polymorphisms are reported to be higher in women (7.8%) but were not significantly high compared to men (6.0%) in the study. The study instigates future research to explore for the possible association of chromosome polymorphism of acro ps+ region including 21ps+ and 22ps+ regions and high prevalence chromosome 21 aneuploidies in women with advanced maternal age and with history of recurrent pregnancy losses.

Keywords: - In total 21/700 (3%) couples showed chromosome abnormalities and chromosome polymorphisms in 471/1400 (33.7%) individuals. After analysis of different types of polymorphic variations in the study and the control group, the prevalence of acro ps+/- polymorphisms involving D/G groups of chromosomes was observed to be significantly higher in the study group with 13ps+/-, 14ps+/-, 15ps+/-, 21ps+/-, 22ps+/- observed in 23.5 % (330/1400) in the study group and 15.8% (58/366) in the control group $p < 0.005$. Among the acro ps+/- the prevalence of 22ps+ subtype polymorphism was significantly higher with the odd ratio OR (95% CI)- 2.35 (1.245-4.434).