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Abstract Topic: - Molecular and cytogenetic diagnostics

Abstract Title: - Cytogenetic Aberrations In Primary And Secondary Infertile Patients Seeking Assisted Reproductive Technology

Presenting author name: - Vasudha Sambyal

Presenting author institute: - GURU NANAK DEV UNIVERSITY AMRITSAR

Co-authors name: - Bhavna Sharma

Co-authors institute: - GURU NANAK DEV UNIVERSITY AMRITSAR

Aims: - In India over 27 million couples are infertile with varying rate of infertility (3.9%-16%) in different regions. In state of Punjab, North India fertility has decreased from 2 live births/woman in 2007 to 1.6 live births/woman in 2018; reported infertility rate 8.8%. Many infertility phenotypes are linked to specific genetic anomalies with multiple reports of chromosomal aberrations in infertile patients. In Punjab, infertility centers offer Assisted Reproductive Technologies (ART) to the infertile couple but cytogenetic analyses are often not recommended before beginning treatment. The present case-control study was conducted to identify the chromosomal aberrations associated with primary and secondary infertility in couples seeking ART.

Methods: - Study included 201 infertile patients (100♂, 101♀) and 201 age and gender matched healthy controls (100♂, 101♀) from Punjab, India. For each subject, standard PHA-M stimulated 72 hours Peripheral Lymphocyte cultures were set up from heparinized intravenous peripheral blood. Metaphase arrest at 70 hours with colcemid was followed by hypotonic treatment and fixation. The prepared slides were GTG banded and after karyotyping chromosomal aberrations were identified in all subjects. The metaphases were scored for aberrations as total metaphases showing any chromosomal aberration (TAM), showing only structural aberrations (TMSA), showing only numerical aberrations (TMNA) and showing both structural and numerical aberrations (TM(NA+SA)).

Results: - The cytogenetic aberration frequency was significantly high in both male and female patients as compared to controls (♂ TAM 28.37 ± 13.46 vs 11.28 ± 7.28 ; ♀ TAM 29.16 ± 14.02 vs 13.60 ± 7.03). Among primary infertility patients, in ♂ TAM 26.96 ± 12.52 vs 11.31 ± 7.02 ; TMSA 16.17 ± 10.74 vs 5.29 ± 5.06 and in ♀ TAM 27.22 ± 12.34 vs 13.44 ± 7.12 ; TMSA 16.74 ± 11.87 vs 5.42 ± 4.83 . The aberrations were significantly higher in 35 secondary infertility couples compared to matched controls and slightly higher than primary infertility patients. Infertile subjects had higher loss of chromosomes (30%) than gain (4%). Increased structural aberrations involving acrocentrics, chromosome X, 2, 7 and chromosome 9 variants were observed in patients with repeated IVF treatment failure.

Conclusions: - The frequency of both chromosome type and chromatid type cytogenetic aberrations was significantly high in infertile patients as compared to controls. The findings of the present study emphasize on importance of cytogenetic analysis in the patients opting for assisted reproductive technology to improve the ART outcome.

Keywords: - The cytogenetic aberration frequency was significantly high in both male and female patients as compared to controls (♂ TAM 28.37 ± 13.46 vs 11.28 ± 7.28 ; ♀ TAM 29.16 ± 14.02 vs 13.60 ± 7.03). Among primary infertility patients, in ♂ TAM 26.96 ± 12.52 vs 11.31 ± 7.02 ; TMSA 16.17 ± 10.74 vs 5.29 ± 5.06 and in ♀ TAM 27.22 ± 12.34 vs 13.44 ± 7.12 ; TMSA 16.74 ± 11.87 vs 5.42 ± 4.83 . The aberrations were significantly higher in 35 secondary infertility couples compared to matched controls and slightly higher than primary infertility patients. Infertile subjects had higher loss of chromosomes (30%) than gain (4%). Increased structural aberrations involving acrocentrics, chromosome X, 2, 7 and chromosome 9 variants were observed in patients with repeated IVF treatment failure.