

# HUMAN GENETICS & GENETIC DISEASES

# MOLECULAR MEDICINE & DIAGNOSTICS

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## Chromosome studies in 55 infertile couples residing in the Bathinda district of Malwa region of Punjab

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**Background:** Malwa region of Punjab has recently been in news due to high incidence of cancer, birth defects and infertility but no systematic study has been carried out to find out the actual incidence and the cause of these disorders is caused by various factors including genetic as well as environmental factors.

**Objectives:** A pilot study was taken up to find out the incidence of chromosomal aberrations among infertile couples from the Bathinda district of Malwa region.

**Methods:** Conventional cytogenetic (from January 2016 to October 2016) study was carried out with blood samples from 55 couples with primary infertility (n=40) and spontaneous abortions (n=15). Karyotyping was done with the help of spectral imaging software (version 6.0). International system for Human Cytogenetic Nomenclature (ISCN 2009) was followed for the nomenclature.

**Results:** Two men were identified with translocation involving chromosomes 4 and 6, [46, XY, t (4; 6) (p15; p12)] and [46, XY, t (4; 6) (p14; p25)], respectively.

**Conclusion:** It is important to screen the couples using conventional cytogenetic technique to avoid unnecessary diagnostic procedures and treatment. The carriers thus identified may opt for ART accordingly to the disorder. More infertile couples need to be screened for chromosomal aberrations in order to establish this as an important causal factor of the disorder.

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