

**A Novel Balanced Autosomal Translocation between Chromosomes 4 and 6 in a Couple
with Recurrent Pregnancy Loss**

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Recurrent pregnancy loss (RPL) is a heterogeneous condition defined as two or more consecutive spontaneous pregnancy loss before 20 weeks of gestation. Factors involving RPL are endocrine, uterine, genetic, immunological, anatomical, infections and psychological. Approximately 15% of all clinically recognized pregnancies results in miscarriage with an incidence of 1 in 300 cases. About 50% of conceptions are spontaneously lost prior to term. A non consanguineous couple of age 30 yr male and 24 yr female with a married life of 4 years were referred to Institute with a clinical history of two first trimester abortions. Physical examination of both the partners revealed normal phenotype. Cytogenetic analysis of peripheral blood lymphocytes of the couple was performed according to Moorehead *et al* (1960) protocol and standard GTG banding was done. 25 metaphases were screened using Olympus BX53 microscope and karyotype analysis was done using ASI spectral imaging software and results were interpreted as per ISCN 2016 nomenclature. Karyotype analysis revealed a balanced autosomal translocation between chromosomes 4 and 6 with 46, XX, t(4;6)(q35; q22) chromosomal constitution in female partner. Further, the karyotype analysis was carried out in female partner's parents and siblings. Interestingly, similar translocation was seen in her father and three sisters, where as her mother and elder sister shown a normal chromosomal constitution, indicating the paternal inheritance. Couples with translocations are at 20% risk of having children with unbalanced chromosomal rearrangement. The formation of balanced, unbalanced and normal gametes is dependent on the basis of the breakpoints and also on the chromosomes involved. Balanced chromosomal translocations may also lead to sequence rearrangements of the functional genes that may result in the reproductive errors accompanied by repeated pregnancy loss. Hence, karyotype analysis followed are genetic counselling is suggested for couples with RPL. Based on which prenatal diagnosis or in vitro fertilization with pre implantation genetic diagnosis are recommended.