

The Prevalence of X-Chromosome Aneuploidy in Recurrent Pregnancy Loss

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Abstract : Recurrent pregnancy loss (RPL), classically defined as the occurrence of two or more failed pregnancies, is a serious reproductive problem, in which, chromosomal rearrangements in either carrier are a major cause; mainly the chromosome aneuploidy. This study was conducted to determine the frequency and contribution of X-chromosome aneuploidy in recurrent pregnancy loss. A retrospective study was carried out among 100 couples with more than 2 miscarriages, referred to our genetic counseling. In all the cases the detailed reproductive histories were taken. Chromosomal analysis was performed using RHG banding in peripheral blood. Of a total of 100 couples; 3 patients with a detected X-chromosome aneuploidy were identified with an overall frequency of 3%. Chromosome abnormalities are as below: a Turner syndrome with 45, X/46, XX mosaicism, a 47, XXX, and a Klinefelter syndrome with 46, XY/47, XXY. These data show a high incidence of X-chromosome aneuploidy; mainly with mosaicism; in RPL. Thus, couples with such chromosomal abnormality should be referred to a clinical geneticist with whom the option of pre-implantation genetic diagnosis in subsequent pregnancy should be discussed.

Keywords : aneuploidy, genetic testing, recurrent pregnancy loss, X-chromosome

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