

Introduction

Recurrent pregnancy loss (RPL) is always destructive for the couple and consist a common dilemma of the reproductive community. Fetal development is multifarious process which requires equilibrium of all hormonal environmental and genetic factors, abnormality in either factor can lead in the abnormal development of embryo or fetal loss. Pregnancy loss is actually nature's quality control for selecting genetically normal offspring. Recurrent pregnancy loss is described as two or more consecutive pregnancy losses before the 20th week of gestation, though it over all distresses 3% of the couples attempting to begin a family. The etiology is often uncertain and may have several factors, like anatomical, immunological, endocrine, infectious, nutritional, environmental and genetic factors, without excluding that the vast majority of cases remain unexplained. Around 10–15% of clinically recognized pregnancies result in miscarriage, and about 50% of early pregnancy losses have chromosome abnormalities. Prenatal cytogenetic analysis could be beneficial for the couples having recurrent pregnancy loss to rule out the possible rearrangement of chromosome consequently genetic counseling. Cytogenetic investigations have been the gold standard technique for decades in several countries to investigate pattern of chromosomal anomalies among the couples having recurrent pregnancy loss. Thus, cytogenetic analysis of spontaneous miscarriages is essential to establish the etiology of fetal losses and to assess patients with risks of recurrence in future pregnancies. Our objective was to examine the type and frequency of chromosome abnormalities in recurrent early pregnancy losses and to analyzed the cytogenetic results from our series of first trimester miscarriages.

Materials and Methods

The presented study was conducted at the Department of Obstetrics and Gynecology, General Hospital of Chania, Crete, Greece. All women presenting with vaginal bleeding and a positive pregnancy test in the first trimester of pregnancy were offered a transvaginal ultrasound examination. Where an early pregnancy failure was diagnosed, with retained products of conception, management options, conservative versus surgical, were discussed with the couple and where surgical management was selected, the women were asked to participate in the study. A total of 111 consecutive chorionic villi samples were obtained by transcervical sampling before evacuation, minimizing external microbial contamination and obtain karyotyping. Results for numerical chromosome abnormalities were analyzed, classifying pregnancies according to maternal age and gestational weeks. Conventional cytogenetic was performed on the blood samples of couples to rule out the possible chromosomal abnormality more precisely chromosomal rearrangement. Every patient was interviewed and consent was taken from the couples.



Results

In 111 samples, the overall and individual frequencies of the different types of chromosome abnormalities were established, including placental mosaicisms, and their relationship with maternal age and gestational weeks was assessed. An abnormal karyotype was detected in 70.2% of the samples, a high detection rate, probably due to the exclusive selection of first trimester miscarriages. Single autosomal trisomy was the most frequent abnormality (64.1% of the abnormal cases), followed by triploidy (12.8%) and monosomy X (9.1%). Chromosome rearrangements were found in 5.3%, combined abnormalities in 8.7%, and placental mosaicism in 3.1% of the cases performed. Individual trisomies behaved differently with respect to maternal age and intrauterine survival. The comparison of age and the number of abortions between male and female carriers shows us that there were no statistical difference in the ages of the female and male carriers, the number of abortions in the case of male carriers were significantly higher than that in the case of female carriers.

Conclusion

Our study offers reliable information on the incidence and type of chromosome abnormalities and placental mosaicism in miscarriages and contributes to define the cytogenetic implication in their etiology and allows providing accurate genetic counseling to families. In future pregnancies the probability of healthy child birth depends on the chromosome number and the type of rearrangement found among the couple. When one partner has the structural chromosomal abnormality, it is highly recommended to perform embryonic karyotyping by amniocentesis, or chorionic villus sampling to avoid the possible chromosomal abnormality among their offspring.

References

1. Priya PK, Mishra VV, Roy P, Patel H. A Study on Balanced Chromosomal Translocations in Couples with Recurrent Pregnancy Loss. J Hum Reprod Sci. 2018 Oct-Dec;11(4):337-342.
2. Najafi K, Gholami S, Moshtagh A, Bazrgar M, Sadatian N, Abbasi G, Rostami P, Khalili S, Babanejad M, Nourmohammadi B, Faramarzi Garous N, Najmabadi H, Kariminejad R. Chromosomal aberrations in pregnancy and fetal loss: Insight on the effect of consanguinity, review of 1625 cases. Mol Genet Genomic Med. 2019 Aug;7(8):e820.
3. Hanif MI, Khan A, Arif A, Shoeb E. Cytogenetic investigation of couples with recurrent spontaneous miscarriages. Pak J Med Sci. 2019 Sep-Oct;35(5):1422-1427.
4. Nikitina TV, Sazhenova EA, Zhigalina DI, Tolmacheva EN, Sukhanova NN, Lebedev IN. Karyotype evaluation of repeated abortions in primary and secondary recurrent pregnancy loss. J Assist Reprod Genet. 2020 Mar;37(3):517-525.
5. Bhatt RK, Agarwal M. Study of Spectrum of Chromosomal Rearrangements in Recurrent Pregnancy Loss. J Obstet Gynaecol India. 2020 Jun;70(3):189-194.

