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Impact of Chromosomal Heteromorphisms on Recurrent Miscarriages

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Abstract

Introduction: Chromosomal Heteromorphisms are the inherited variations at specific chromosomal regions without a proven impact on the phenotype of an individual.

Material and Methods: We studied and compared the chromosome Heteromorphisms in the Karyotypes of two groups. The first group consisted of 320 individuals of 160 couples with more than two miscarriages and no live birth and the second group consisted of 412 individuals of 206 normal couples with more than 1 normal child birth.

Results and discussion: Twenty Nine individuals (9.06%) with a history of recurrent pregnancy loss were found to have chromosomal heteromorphisms, whereas thirteen individuals (3.1%) from the normal group were having the chromosomal heteromorphism. The difference between the two groups was statistically significant ($p<0.0001$)

Conclusion: These results are consistent with other similar studies that suggest the yet undefined relationship between chromosome heteromorphism and recurrent pregnancy loss.

Keywords: Recurrent pregnancy loss; Chromosome heteromorphism

Introduction

Repeated DNA sequences in the genome are subject to heteromorphism [1]. Chromosomal heteromorphisms are structural chromosomal variants that are widespread in human populations and have no known effect on phenotype [2]. These heteromorphic regions may be identified by several methods; each of these methods reveals a typical staining pattern implying constitutional differences in heterochromatin. Common chromosomal polymorphisms detected by Giemsa banding are considered as heteromorphisms and include heterochromatin regions on short arms of Acrocentric chromosomes and regions of chromosomes 1, 9, 16 and Y. Impact of chromosome heteromorphism has been studied previously on infertility and recurrent miscarriages [1,3]. There seems an increased incidence of chromosome heteromorphism in infertility and recurrent miscarriages; however, the underlined mechanism needs to be clearly defined yet.

We aimed to study the impact of chromosomal heteromorphisms of Acrocentric chromosomes on recurrent miscarriages in the present study, by comparing the frequencies of heteromorphic regions in Karyotypes of normal individuals and the couples with recurrent pregnancy losses.

Material and Methods

We studied and compared the chromosome Heteromorphisms in the Karyotypes of two groups. The first group consisted of 160 couples with more than two first trimester miscarriages and no live birth that were referred to our Genetic Health & Research Center between October 2010 to December 2011 for Karyotyping and the second group consisted of 412 individuals of 206 normal couples with more than 1 normal child births and absence of history of abortions.

Chromosomal analysis was done in cytogenetic laboratory using standard methods. Peripheral blood samples from individuals of both the groups were cultured for 72 hours and chromosomes were studied after Giemsa- Trypsin banding.

At least 20 metaphases were analysed for each case and heteromorphisms were reported according to ISCN 2005.

Prominent stalks and satellites of chromosomes 13, 14, 15, 21 and 22 were included in the study. The findings were considered heteromorphic if the chromosome region of interest was greater than the same region of its homologue.

All the detected heteromorphic Karyotypes were examined under light microscope by two independent laboratory technicians in the laboratory to avoid uncertainty and variability in the results.

Statistical analysis was done by two- tailed Fisher's exact test.

Results and Discussion

In couples with recurrent pregnancy loss, twenty nine individuals (16 males and 13 females; 9.06%) were found to have chromosomal heteromorphisms in the acrocentric chromosomes. In males the frequency of heteromorphism was 10% and in the females 8.12% (Table 1).

Most of the spontaneous miscarriages occur because of chromosomal abnormalities in the embryo or foetus [4-6]. Several early studies [5,7-10] suggested that the variation in size of heterochromatic regions on human chromosome might have deleterious effect. Hong Y et al. [11] has shown that the outcome of infertility treatment (IVF/ICSI) in non-carriers and carriers of polymorphic chromosome variants does not differ; however, the early miscarriage rate of the male carriers tends to be higher.

The chromosomal polymorphisms of short arms of acrocentric chromosomes have been reported in humans by Podugolnikova et

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Chromosomes	Number of heteromorphic chromosomes in Group I (320)			Number of heteromorphic chromosomes in Group II (412)		
	Male (16)	Female (13)	Total	Male (6)	Female (7)	Total
13	2	2	29 (9.06%)	1	1	13 (3.1%)
14	2	2		0	1	
15	7	5		2	3	
21	1	1		1	1	
22	4	3		2	1	

Group I- Couples with more than 2 first trimester miscarriages and no live birth, Group II- Couples with more than 1 normal child and absence of history of abortions

Table 1: Acrocentric Chromosomal heteromorphism observed in two groups.

al. [12]. According to the present study, the most frequently affected chromosomes are chromosome 15 and 22 as compared to chromosomes 13, 14 and 21, which is consistent with the findings of Purandare et al. [3]. No specific functions have been reported to be associated with the satellite segments (ps+); however, such variations in the couple may make the fetus susceptible to translocations which may lead to fetal wastage [3]. Karyotyping 842 individuals with primary infertility or repeated miscarriages by Madon et al. [10] showed polymorphic variants in 28.82% of males and 17.19% of females, which was quite high as compared to 10% males and 8.12% of females in our study. The present research finding shows that the incidence of heterochromatic variations on the short arms of acrocentric chromosomes is higher in couples with early miscarriages.

Conclusion

Present study shows that the prevalence of acrocentric chromosomal heteromorphisms is higher in coupes with early miscarriages and is consistent with figures described in several populations across the world. This could be an important reason for early miscarriage. However; the number of carriers of acrocentric chromosomal variations in our study was insufficient. Moreover, the chromosomal analysis method in the present study had 450 Giemsa banding resolution. Therefore, to establish a definite relationship, more sample size and more sensitive techniques are needed.

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