

# Chromosomal abnormalities as a cause of recurrent abortions in Egypt

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**BACKGROUND:** In 4%-8% of couples with recurrent abortion, at least one of the partners has chromosomal abnormality. Most spontaneous miscarriages which happen in the first and second trimesters are caused by chromosomal abnormalities. These chromosomal abnormalities may be either numerical or structural.

**MATERIAL AND METHODS:** Cytogenetic study was done for 73 Egyptian couples who presented with recurrent abortion at Genetic Unit of Children Hospital, Mansoura University.

**RESULTS:** We found that the frequency of chromosomal abnormalities was not significantly different from that reported worldwide. Chromosomal abnormalities were detected in 9 (6.1%) of 73 couples. Seven of chromosomal abnormalities were structural and two of them were numerical.

**CONCLUSION:** Our results showed that 6.1% of the couples with recurrent abortion had chromosomal abnormalities, with no other abnormalities. We suggest that it is necessary to perform cytogenetic investigation for couples who have recurrent abortion.

**Key words:** Chromosomal abnormalities, cytogenetics, recurrent abortions

pregnancies end in spontaneous abortion.<sup>[1]</sup> In 4%-8% of couples with recurrent pregnancy loss, at least one of the partners has chromosomal abnormality that probably contains balanced chromosomal abnormalities.<sup>[2]</sup> These chromosomal abnormalities may be either numerical or structural.<sup>[3]</sup> This result from the production of gametes and embryos with unbalanced chromosome sets.<sup>[4,5]</sup> The clinical consequences of such abnormal gametes include sterility, repeated abortions, and giving birth to malformed children.<sup>[6,7]</sup> The majority of chromosome rearrangements are balanced reciprocal and Robertsonian translocations. It is known that such abnormalities cause no phenotypic effect on the carrier but lead to increased risk to produce unbalanced gametes. Therefore, they have not only high risk to give abnormal offspring with unbalanced karyotypes, but also have increased prevalence of miscarriages.<sup>[8,9]</sup>

The aim of this study was to determine the chromosomal abnormalities in Egyptian couples with recurrent abortion refer to Cytogenetic Unit of Children Hospital, Mansoura University.

## Introduction

Approximately 15%-20% of clinically recognizable

## Materials and Methods

After exclude immunologic effects, uterine malformations and other causes of recurrent abortion, 73 couples with at least two pregnancy losses were referred to Cytogenetic Unit of Mansoura University Children Hospital. The mean age of the females was 27 years, while it was 31 years for the males. Chromosomes were obtained from peripheral blood cultures according to Rooney and Czepulkowski.<sup>[10]</sup> Three to five milliliter of

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sodium-heparinized whole blood was collected from each patient and control individual. An amount of 0.5 cc of each patient and control individual's blood sample was added to 5 cc of a complete media containing RPMI 1640, fetal calf serum (10%), PHA (10 µg/ml), L-glutamate (2 mM), Penicillin (200 unit/ml), and Gentamycin (50 µg/ml). After 70 hours of incubation in 37°C, colcemide was added (0.2 µg/ml). After 90 min, the cells were harvested by centrifugation (150 × g for 10 min). Then, 5 ml of 0.075 M KCl solution was added and mixed and incubated at 37°C for 15 min. After centrifugation (150 × g for 10 min), hypotonic supernatant was removed. Then, 5cc cold, fresh fixative solution (3:1 methanol-acetic acid) was added drop-wise to the cell pellet. Centrifugation was done afterwards and the supernatant removed. These two latter steps were repeated until a clear pellet was obtained. Finally, cells obtained were dropped on distinct slides. Staining with Giemsa was performed for some of the slides prepared from each patient and analyzed by cytovision system.

## Results

A total of 73 Egyptian couples with history of recurrent abortion were examined. Their ages ranged from 20 to 46 years, with a mean of 29 years. The number of recurrent abortion varied from 2 to 10 abortions/couple, Chromosome abnormalities were found in (2/27)=7.4% of the couples with a history of two abortions, in (3/23)=13% with three abortions, and in (4/23)=17.39% with four or more abortions. Among these 73 couples, 9 (6.1%) were found to be carriers of different chromosomal abnormalities, five females (3.42%) and four males (2.74%). Seven of chromosomal abnormalities were structural and two of them were numerical. These abnormalities included three balanced reciprocal translocations, one Robertsonian translocation, two inversions, one case of mosaic X-chromosome monosomy, one trisomy X-chromosome and one deletion of short arm of chromosome X [Table 1].

## Discussion

Several studies have been carried out to determine the prevalence of chromosomal aberrations among

couples with recurrent abortion. This was found to be 4.7%, 8.6%, 9.92%, 5.34%, 6.7%, and 5.3% in six of the largest reviews that were reported by De braekeleer *et al.*,<sup>[11]</sup> Makino *et al.*,<sup>[12]</sup> Tsui k *et al.*,<sup>[13]</sup> Fryns *et al.*,<sup>[14]</sup> Al Husein *et al.*<sup>[15]</sup> and Azim *et al.*,<sup>[16]</sup> respectively [Table 2]. In our study, we found that the incidence of chromosomal abnormalities among couples with recurrent abortions was 6.1%, which is not significantly different from this global incidence.

Kiss *et al.* found that, Chromosome abnormalities were found in 5% of the couples with a history of two abortions, in 10.3% with three abortions, and in 14.3% with four or more abortions,<sup>[17]</sup> as a similar to this, in our study we found that (2/27) = 7.4% of the couples with a history of two abortions, in (3/23) = 13% with three abortions and in (4/23) = 17.39% with four or more abortions.

The structural chromosomal abnormalities that we encountered were divided into balanced reciprocal chromosomal translocations (3/9), Robertsonian translocation (1/9), inversions (2/9) and deletion (1/9). The distribution of structural chromosomal rearrangements in our study is similar to that reported worldwide by.<sup>[18-20]</sup>

Autosomal reciprocal translocations have been proposed as the most common chromosomal changes in couples who have recurrent abortion.<sup>[21-23]</sup> In the same way, reciprocal translocations were the most common

**Table 1: Cytogenetic findings, number of abortions and age in cases with abnormal karyotype**

Karyotype	No. of abortions	Age
46,XX,t(14;X)(q11;q12)	4	26
45,XY,t(13;14)(q11;q11)	2	32
46,XY,t(3;22)(q11;p11)	2	38
46,XY,t(16;X)(q24;q23)	3	39
46,XX,inv(2)(p25;q14)	5	28
46,XY,inv(3)(p26;q21)	7	42
46,X,Xq-	3	23
46,XXX	4	27
46,XX/45,X	3	33

**Table 2: Global studies of chromosomal abnormalities found in couple with recurrent abortion**

Country	Authors	Year	Percent of chromosomal abnormalities
Canada	De braekeleer M <i>et al.</i> ,	1990	4.7
Japan	Makino T <i>et al.</i> ,	1990	8.6
China	Tsui kM <i>et al.</i> ,	1996	9.92
Belgium	Fryns Jp <i>et al.</i> ,	1998	5.34
Saudi Arabia	Al Husein M <i>et al.</i> ,	2000	6.7
Pakistan	Azim M <i>et al.</i> ,	2003	5.3

abnormalities (2.05%) in our studies as reported in literature.

Numerical chromosomal aberrations are less frequently encountered among couples with repeated abortions. Those aberrations are usually in the form of sex chromosomal aneuploidy, and they occur in a low frequency (<0.15% of cases).<sup>[24]</sup> We encountered one case with X-chromosome mosaicism and one case with trisomy X.

In conclusion, our results showed that, 6.1% of the couples with recurrent abortion had chromosomal abnormalities, with no other abnormalities. We suggest that it is necessary to perform cytogenetic investigation for couples who have recurrent abortion.

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