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ARTICLE

Chromosomal analyses of 1510 couples who have experienced recurrent spontaneous abortions

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Professor Osman Demirhan was born in Konya, Turkey, in 1958. After completing elementary and middle school in the same town, he attended Hacettepe University Biology Department (Faculty of Science) and graduated in 1982. He began working at Çukurova University, Medical School, Department of Medical Biology, as a research assistant in 1983. He received his MSC degree in 1987, PhD degree in 1991 and was appointed Professor in the same department in 2004. He received professor degree at 2004, is still working at the same deparment. He also studied at the Department of Medical Genetics, Antwerp University, Belgium, in 1995.

Abstract In this retrospective study, karyotype results of 1510 couples with a history of recurrent spontaneous abortion were evaluated. The study was conducted at Balcalı Hospital in Adana region of Turkey. For all cases, peripheral blood lymphocytes were cultured for chromosome study using the standard method. Chromosome aberrations were detected in 62 couples (4.1% of all couples). At an individual level, chromosome aberrations were found in a total of 65 cases (41 females and 24 male cases), with structural chromosomal aberrations in 58 cases including balanced translocations in 30 cases, Robertsonian translocations in 12 cases, deletions in seven cases, inversions in nine cases and numerical chromosome aberrations in seven cases. The results of the study indicated that structural aberrations, particularly translocations, were the most common type of aberration observed among couples who had experienced recurrent spontaneous abortions and that these couples might benefit from cytogenetic analyses.
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Introduction

Recurrent spontaneous abortions that occur before the 24th week of gestation are observed among 1–3% of couples trying to have children. Various different definitions for recurrent spontaneous abortion have been proposed; certain studies defining recurrent spontaneos abortion as three or more losses of pregnancy (Coulam, 1991; Dubey et al., 2005; Flynn et al., 2014; Garrido-Gimenez and Alijotas-Reig, 2015; Grande et al., 2012; Mozdarani et al., 2008; Stirrat, 1990), whereas others

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define it as two or more failed pregnancies (Kochhar and Ghosh, 2013; Sugiura-Ogasawara, 2014; Van den Berg et al., 2012); these losses might be consecutive or non-consecutive. Nearly 15% of all clinically identified pregnancies end in spontaneous abortion (El-Dahtory, 2011; Exalto, 2005; Niroumanesh et al., 2011). Recurrent spontaneous abotions can be caused by a variety of factors, generally related to implantation, anatomic uterine defects, infections, autoimmunity, allo-immunity, endocrine abnormalities and genetics (Dubey et al., 2005). In 50% of couples, however, the exact cause of spontaneous abortion cannot be identified; in such cases, the spontaneous abortion is considered as idiopathic, or referred to as an unexplained spontaneous abotion (Flynn et al., 2014). A considerable proportion of couples who experience recurrent spontaneous abortions also exhibit chromosome aberrations. Numerous studies have been conducted to investigate the proportion of chromosome aberrations among couples experiencing recurrent spontaneous abortions, with some indicating that between 2.7–6.7% of couples experiencing recurrent spontaneous abortions are chromosome aberration carriers (Dutta et al., 2011; Goud et al., 2009; Stephenson and Sierra, 2006). Other studies have reported that, among couples who have experienced recurrent spontaneous abortions, the frequency of chromosome aberrations varies between 4 and 8% (El-Dahtory, 2011; Kavalier, 2005). Both numerical and structural chromosome aberrations have been observed, although the former type of aberration tends to be less common than the latter. Frequently observed chromosome aberrations include balanced translocations, Robertsonian translocations and inversions. The chromosome aberrations mentioned above might not necessarily result in the gain or loss of genetic material; such chromosome aberrations are defined as balanced rearrangements, which do not affect the viability or life of the individual.

During the segregation of chromosomes in meiosis, however, the presence of balanced rearrangements may lead to an unbalanced karyotype in the carrier's gametes, which in turn can result in spontaneous abortion, still birth or neonate congenital defects (Flynn et al., 2014).

The purpose of our study was to investigate 1510 couples who experienced recurrent spontaneous abortions, and to evaluate their karyotypes. We also aimed to obtain and identify significant statistical data that would contribute to research and help guide clinicians and genetic counsellors in counselling couples who have experienced recurrent spontaneous abortion. We believe that, owing to its evaluation of a large number of cases over a 28-year period, the present study and its findings are of considerable importance.

Materials and methods

The study was conducted retrospectively on couples who had experienced recurrent spontaneous abortion who were admitted to our institution between January 1982 and December 2010. The study was conducted at the cytogenetics laboratory of Çukurova University Balcalı Hospital's Medical Biology and Genetics Department, located in Adana, Turkey. All 1510 couples with a minimum of two pregnancy losses were referred by gynaecologists of the same hospital for chromosome analysis. After all other possible causes of spontaneous abortion were excluded, i.e. anatomic uterine defects, infections, autoimmunity, allo-immunity, and endocrine abnormalities, the couples were directed to chromosome analyses. Couples presenting diverse pregnancy loss histories were included in the study. As such, couples participating in the present study included couples who only experienced recurrent spontaneous abortions, couples whose recurrent spontaneous abortions were preceded by abnormal children or by stillbirths, and couples who had healthy children despite recurrent spontaneous abortions. A total of 2 ml heparinized (Vasparin, Defarma, Ankara, Turkey) venous blood was collected from each study participant. Slides were prepared using standard culture, swelling, and fixation procedures, and then stained according to the banding using the Trypsin and Giemsa GTG method. Chromosome analysis was conducted using CytoVision software. At least 20 metaphases were analysed for each case, whereas a minimum of 30 metaphases or 50 metaphases were evaluated in abnormal and mosaic cases, respectively. Solid staining was used for a few cases, specifically for cases who had applied to our laboratory during the early 1980s. When reporting the study result, the international system for human cytogenetic nomenclature (ISCN 1981 and 1985) at 500–550 band resolution was primarily used. Definition of any aberration at 500–550 band resolution paves the way for using molecular cytogenetic techniques, other molecular techniques, or both, and describing the aberrations at a molecular level. The chromosome aberrations were classified as either numerical or structural aberrations. The latter were further sub-divided as balanced translocations, Robertsonian translocations, inversions and deletions.

When planning the study, the necessary consultations were made with the ethics committee of the Çukurova University Medical Faculty.

Results

A total of 1510 couples were included in the study; 62 (4.1%) of the couples were identified as having chromosome aberrations and, in three of the couples, both the male and female were chromosome aberration carriers. As such, the total number of cases with chromosome aberrations was 65 (41 women and 24 men) (**Table 1**). The mean age of these cases was 29 years for women and 33.5 for men. At an individual level, a total of 3020 cases (1510 women and 1510 men) were evaluated. Among these cases, the mean age was 28.7 years for 1486 women/mothers, and 32.6 for 1474 men/fathers; records indicating age were absent for 24 women and 36 men. The proportions of structural and numerical aberrations were determined as 89% (58 cases) and 11% (seven cases), respectively. In addition, the proportions of balanced translocations, Robertsonian translocations, deletions and inversions were determined as 46%, 18%, 11% and 14%, respectively (**Figure 1**).

On the basis of the available medical records, the number of spontaneous abortions could be identified for 763 couples. As gynaecologists refer any couple with a minimum of two spontaneous abortions for chromosome analysis, the exact number of spontaneous abortions for the other 747 couples could not be determined, despite knowing that they had at least two spontaneous abortions. These couples were mainly those who had applied to our laboratory before 2000. The total numbers of spontaneous abortions varied between two and nine, with

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