



A 15; 15 Translocation in a couple with Repeated Abortions: Case Report

Azam Ahmadi Shadmehri^{1,2}, Javad Tavakkoly Bazzaz³, Mojtaba Darbouy², Mohammad Amin Tabatabaiefar^{4,5*}

¹ Department of Molecular Genetics, Marvdasht Branch, Islamic Azad University, Marvdasht, Iran

² Department of Molecular Genetics, Science and Research Branch, Islamic Azad University, Fars, Iran

³ Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences, Tehran, Iran

⁴ Department of Genetics and Molecular Biology, School of Medicine, Isfahan University of Medical Sciences, Isfahan, Iran

⁵ Pediatric Inherited Diseases Research Center, Research Institute for Primordial Prevention of Non-Communicable Disease, Isfahan University of Medical Sciences, Isfahan, Iran

***Corresponding authors:** Mohammad Amin Tabatabaiefar, **Address:** Pediatric Inherited Diseases Research Center, Research Institute for Primordial Prevention of Non-Communicable Disease, Isfahan University of Medical Sciences, Isfahan, Iran, **Email:** mamintab@yahoo.co.uk, **Tel:** +983137929023

Abstract

Carriers of structural chromosomal rearrangements such as Robertsonian or reciprocal translocations have an increased risk of spontaneous abortion and producing offspring with genetic abnormalities. Robertsonian translocations are present in 0.1% of the general population and 1% of the infertile population. Two types of Robertsonian translocations occur more frequently than all others, being 45,XX,rob(13;14)(q10;q10) and 45,XX,rob(14;21)(q10;q10) respectively. The history of repeated abortions could be the outcome of unbalanced gametes (either monosomy or trisomy) resulting during the meiotic segregation of the balanced heterozygote female carrier.

In the present report, uncommon Robertsonian translocation in a couple with spontaneous repeated abortions is reported. Cytogenetic analysis of a couple revealed the presence of 45, XY, t (15; 15) (10q; 10q) chromosomal constitution in the male partner.

The cytogenetic analysis of couples with repeated abortions is obligatory to identify any probable chromosomal aberrations. As far as we know this is the first instance reported in Iran.

Keywords: Carrier, Chromosomal rearrangements, Robertsonian translocations, Abortion

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Introduction

The human cell nucleus contains 23 pairs of chromosomes, on which lie 300,000 genes which carry the hereditary information (1). Chromosomal

aberrations lead to reduced fertility in both men and women (2). About 15% of pregnancies end up in spontaneous abortions mostly in the first trimester. The most frequent cause being represented by chromosomal

abnormalities, with an incidence of approximately 50 % in spontaneous abortions (3). Reciprocal and non-reciprocal translocations are commonly observed among the chromosomes. This process involves the exchange of different chromosomal fragments (4). It is estimated that 0.2% of individuals carry a symptomatic chromosomal rearrangement (5). A special type of non-reciprocal translocation is Robertsonian translocation. Robertsonian translocation is recognized to be the most common structural chromosomal abnormality in the population with an incidence of 1.23/1000 live births (6). Robertsonian translocations usually involve chromosomes from pairs 13, 14, 15, 21 or 22. The most frequent type of D/D translocation includes 13; 14 translocation, while 13; 15 and 14; 15 are rare structural rearrangements among Robertsonian translocations (7, 8). Robertsonian translocation is centric fusion of two acrocentric chromosomes. The resulting balanced karyotype has only 45 chromosomes including the translocated one, which in fact is made up of the long arms of two chromosomes (9). Robertsonian translocations are important in human genetics especially that it can be considered as a risk factor for Down's syndrome (10). The meiotic segregation pattern of this translocation heterozygote carrier results in trisomy or monosomy of the chromosomes (11-13). Balanced homologous Robertsonian translocations despite being rare have been reported as a cause of

recurrent spontaneous abortions in all the acrocentric autosomes (14, 15). In this study, we report a male with the 15q; 15q Robertsonian translocation, the couple had 3 recurrent spontaneous abortions.

Case report

A couple of age 33 years (male) and 26 years old (female) were referred to the Institute of Genetic Counseling, Birjand with a history of repeated abortions. They were related as first cousins. The wife had had four spontaneous abortions during the first trimester. The proband was phenotypically normal. His wife had normal menstrual cycles.

Chromosome Analysis

For karyotype analysis, 2 ml peripheral blood was sampled from both partners in 5 ml lithium heparinized tubes. Then 0.2 ml of the blood sample was added to 4 ml of HAM-F10 medium plus 1 ml of sterile fetal bovine serum, to which 0.2 ml of phytohemagglutinin was added. After 72 hours of incubation at 37° C, the cells were harvested at early metaphase by addition of 0.2 ml colcemid solution and prepared for GTG-banding (Gimsa Tripfin G-banding). The proband proved to be a carrier of chromosomal translocation and her partner's karyotype was found to be normal. The karyotype of the proband was 45, XY, t(15;15)(10q;10q) as seen in Figure 1.



Discussion

Chromosomal aberrations during embryogenesis are responsible for approximately 60 % of all spontaneous abortions in early pregnancies (16). The majority of pregnancy losses or neonatal deaths are attributed to the numerical chromosomal abnormalities especially trisomies of chromosome 13, 18, 21 etc. The structural aberrations could result in pregnancy loss and infertility (17). In a Robertsonian translocation, two of the five acrocentric chromosomes have broken at the beginning of the short arm near the point where it meets the long arm. The long arms have then fused together.

The commonest structural aberrations of the acrocentric chromosomes is Robertsonian translocations, the most common are the non-homologous forms (18). In general, carriers of homologous Robertsonian translocations are phenotypically normal. However, some carriers of t(15q15q) have been reported with Prader-Willi syndrome, putatively as the result of breakage in the region of band 15q11 or loss of the segment 15q11-15q13 (19, 20). The translocation between the D/D group is the most frequent one and studies of both spontaneous abortions and live births indicate a high predominance of 13;14 translocation. Translocations of Dq; Dq were about six times more frequent than Dq; Gqs (21).

This particular case of 15;15 Robertsonian translocation is very rarely seen in the postnatal or prenatal period (22, 23) and it is associated with infertility like in all Robertsonian translocations (24). In the present report, we found a unique Robertsonian translocation in chromosomes 15;15 which resulted in repeated abortions. As far as we know this is the first instance reported in Iran.

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