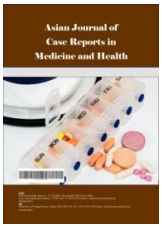

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A Rare De Novo Reciprocal Translocation 46,XX,rec(7;13)(p22;q32) Karyotype

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Authors' contributions

This work was carried out in collaboration between both authors. Authors STO and ME designed the study, collected the data and performed the statistical analysis, wrote the protocol and wrote the first draft of the manuscript. Author STO supervised the writing and critical review of the study. Authors STO and ME managed the literature searches. Both authors read and approved the final manuscript.

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Case Study

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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. We report an women with uncommon unbalanced Reciprocal translocation carrier 46,XX,rec(7;13)(q22; q32) chromosomal constitution. While her husband and her father showed normal 46,XY karyotype, her son and her mother showed same abnormal kartotype. Peripheral blood were taken from proband and family members, then performed with lymphocyte culture and stained by binded using Giemsa-banding method. According to the cytogenetic study results of first degree relatives of our proband, reciprocal translocation was maternally inherited in our case. Uniparental dysomia (UPD) is an abnormal condition in which a homologous chromosome pair is both from one parent and not from the other parent. The maternal inheritance of translocated chromosomes is same time compatible with UPD.

Keywords: Reciprocal translocations; carrier; chromosomal rearrangements; abortion; uniparental disomy.

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1. INTRODUCTION

Reciprocal chromosomal translocations (RCT) most common cause of pregnancy losses is chromosomal anomalies. According to Nielsen and van Dyke; The incidence of reciprocal translocations in the neonatal population was estimated to be 1:712 and their frequency was estimated to be approximately 1:250 in the examinations performed during prenatal diagnosis determined by amniocentesis [1,2]. It is accepted that the chromosomal rearrangements detected in the parents are an important etiological factor in spontaneous abortion, stillbirth or birth of a baby with malformation. Chromosomal abnormalities that occur with balanced rearrangements are one of the causes of reproductive failure or reduced reproductive abilities in humans. Often make couples unhappy and cause many marriages to end; it results in infertility, habitual disorders or the birth of children with an unbalanced karyotype [3].

During meiosis, chromosomally imbalanced gametes between translocated chromosomes and their normal homologues in reciprocal translocation carriers are distributed into daughter gametes as a result of different segregations. Balanced reciprocal translocation carrier meiosis I, when gametes form in the mother or father 4 chromosome to 2 daughter cell in division in various ways (Alternate, Adjacent 1, Adjacent 2) is dispersed and segregated and forms the gametes [4]. In alternate segregation, the translocated chromosomes segregate to one pole and the normal homologues to the other, producing balanced and normal gametes, respectively. Alternate two derivative chromosomes in segregation 2 normal chromosomes to the daughter cell, the other goes to the daughter cell. Resulting half of the gametes are balanced translocation chromosome carrier while the other half it has normal chromosome content. Adjacent formed in 1 and 2 segregation gametes have unbalanced chromosome content creates partial trisomic products. Gametes with normal or balanced chromosome formation arise as a result of alternative segregation only when the reciprocal translocation chromosomes are separated into each pole and the normal homologous chromosome is distributed to the other pole. There are 6 possibilities for reciprocal translocation in the 2: 2 distribution; It results in the formation of 1/6 completely normal, 1/6 stable carrier and 4/16 unbalanced-lethal gametes. Segregation can occur in three ways; one can result in 3:1 segregation (tertiary or exchange), 2:2 segregation (contiguous-1 or contiguous-2) and 4:0 and only unbalanced gametes are produced [5], (Fig.1, Fig. 2).

Abnormal phenotype is not observed in these translocations, but the real danger is that the silent translocation carriers will encounter varying degrees of risk for spontaneous abortions and the birth of children with anomalies, which they will encounter in their desired adulthood [6].

2. CASE REPORT

A non-consanguineous couple of age 25 years (male) and 22 years old (female) were admitted in the Afyonkarahisar Health Science University of Medical Genetics Department with a history of repeated abortions for cytogenetics evaluation. They were not related. The proband was phenotypically normal and normal menstrual cycles. There were two stories of recurring abortions in the last two years of their marriage. Their first abortion occurred four months after conception. There were no reasons such as illness, stress or weight lifting that could cause abortions. When the fetus was examined, decidua and edematous villus structures were detected. Her mother's sister had such recurrent abortion stories. Parents of proband were made cytogenetics analyses also. These family members do not have any characteristic disease (cancer, malformations, etc).The karyotype of the proband and his son (4 years old) was 46,XX, rec (7;13)(q22; q32), and the proband's husband had a normal karyotype (46,XY). Parents of proband were analysed, father of proband (55 years old) was 46,XY normal karyotype, while mother of proband (50 years old) was 46,XX, rec (7;13)(q22; q32) as seen in Fig. 3.

Genetic counseling was given to the family. Since the mother and grandmother are carriers of balanced reciprocal translocation, they can have only 1/6 healthy children because they will always have normal chromosomes from their spouse who has 46, XY normal karyotype. This couple has all pregnancy miscarriage, intrauterine fetal death, congenital anomaly baby, chromosomally balanced translocation carrier but normal baby with phenotype or completely healthy baby has the possibilities. Therefore, PGD(Preimplantation Genetic Diagnosis) was recommended for the mother's next pregnancy and when her son is going to have a child in the future.

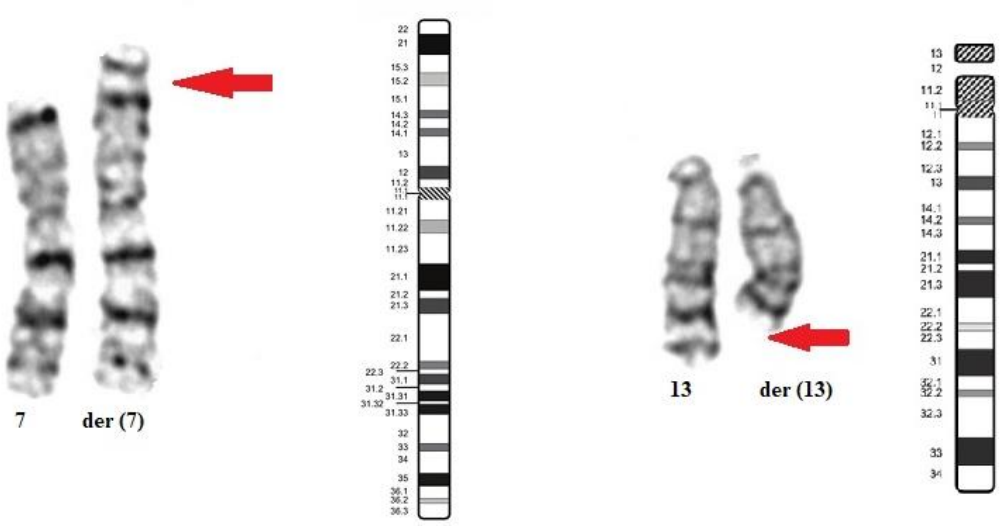

Fig. 1. Kartoype of proband, chromosome establishment is 46,XX,rec(7;13)(q22;q32)

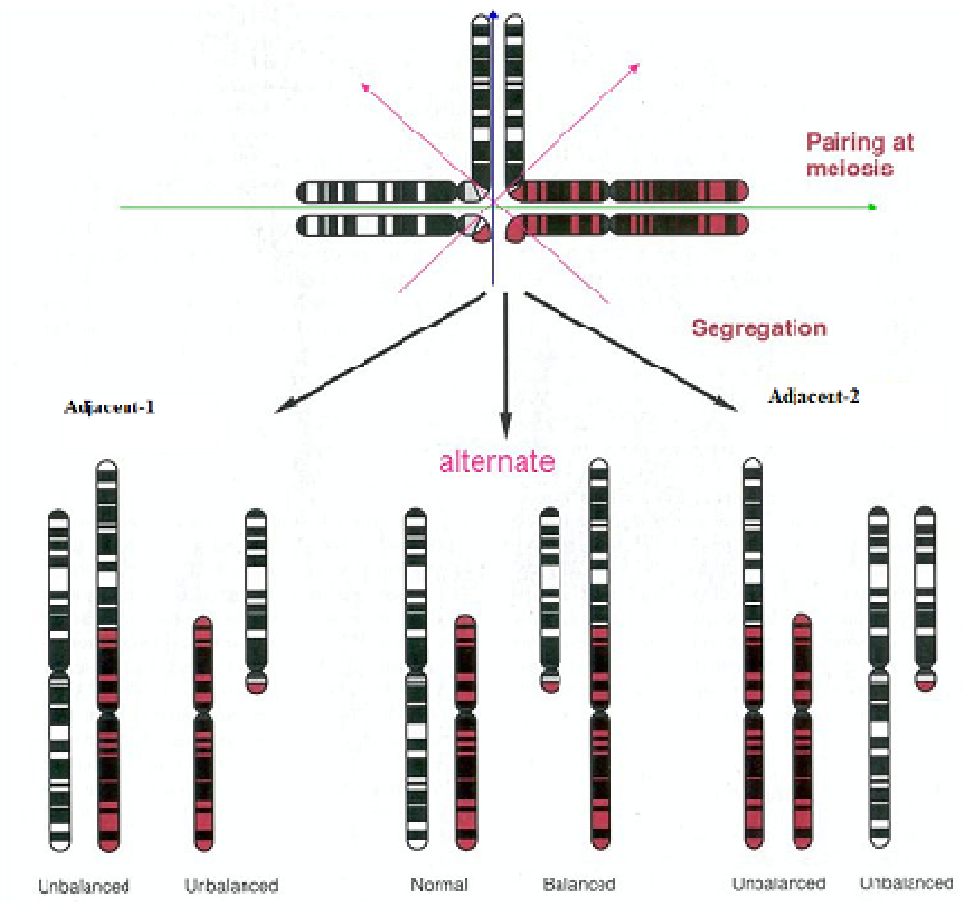

Fig. 2. Possible segregation consequences of reciprocal translocation [9]

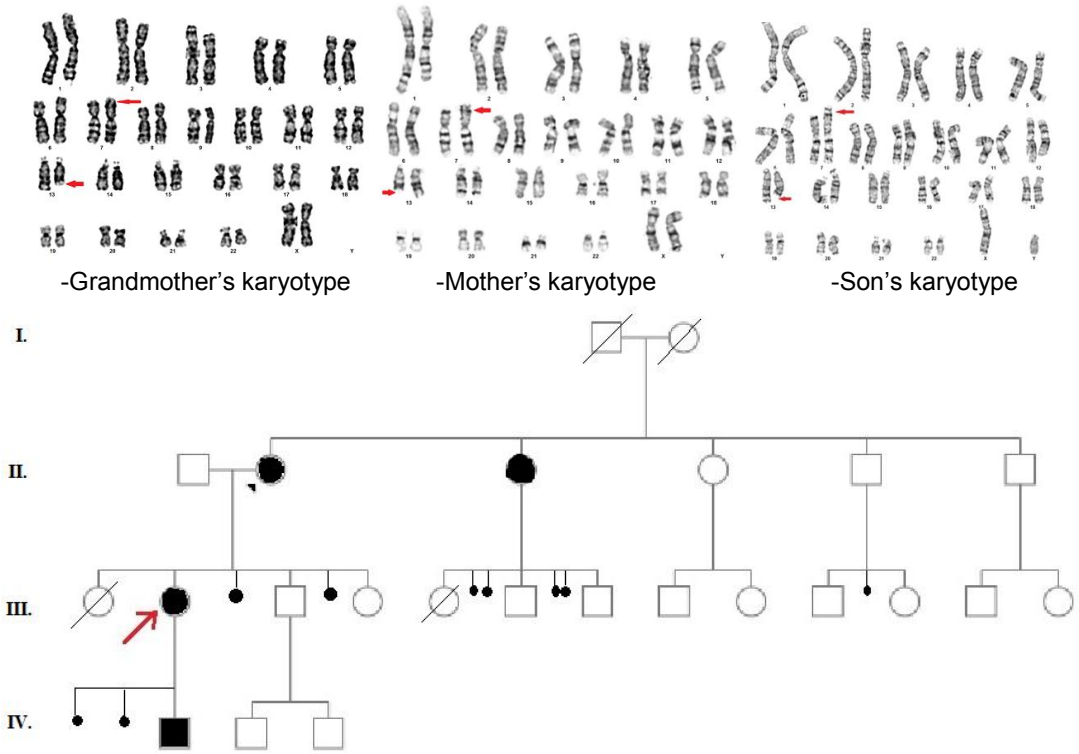

Fig. 3. The pedigree and karyotype of the grandmother, proband and her son were 46,XX, rec(7;13) (p22;q32)

3. CHROMOSOME ANALYSIS

For cytogenetic analysis for karyotyping analysis, 2 ml peripheral blood samples with sodium heparin were collected from both partners. RPMI 1640 medium was prepared for lymphocyte culture, supplemented with 0.2 ml PHA, and cultured blood samples were incubated at 37 ° C for 72 hours. After fixation using Cornoy's fixative (3:1 methanol-acetic acid), G-banded metaphases were prepared for cytogenetic analysis and chromosome study was performed. Twenty-five metaphases for each partner were analyzed and the karyotype was interpreted using Applied Imaging Software. Chromosomes were identified and classified according to the International System for Human Cytogenetic Nomenclature (ISCN, 2016) guidelines, karyotyping, and numerical and structural abnormalities recorded.

4. DISCUSSION

In this case report, we present a patient with de novo 46,XX, rec(7;13)(q22;q32) balanced reciprocal translocation carrier a female. Our proband transferred the chromosome establishment with reciprocal translocation she received from her mother through maternal inheritance to her son. Our proband's husband and father had normal karyotype, therefore paternal inheritance was not considered. Balanced translocations are inherently passed unnoticed down from generation to generation and can sometimes even be carried in an entire family without any anomalies, such translocations are called "familial translocations". This condition, which does not comply with Mendelian inheritance, is called maternal (maternal) UPD if both homologous chromosomes are inherited from the mother, and paternal UPD if inherited from the father [7]. There is maternal uniparental dysomia in our case.

According to Warburton (1993); Individuals with balanced reciprocal translocation are phenotypically normal, but risk having children with chromosomes in subsequent generations Reciprocal translocations occur due to the exchange of chromosomal material between two non-homologous chromosomes, there is no phenotypic effect on the individual due to the balancing of the normal chromosome and the

amount of genetic material from the partner. Reciprocal translocations can occur either inherited or de novo. The risk of having de novo translocations is greater than those that are inherited, showing an incidence of 6-7% [8].

5. CONCLUSION

It constitutes an important group in prenatal cytogenetic diagnosis indications, since parents with balanced chromosomal anomalies have a 10-15% risk of carrying fetuses with unbalanced chromosomal anomalies. The reciprocal translocation detected in the proband, mother and son of the proband reveals the necessity of prenatal cytogenetic diagnosis in all future pregnancies of the family.

CONSENT

Consent form was obtained from the patients since cytogenetic analysis were studied from peripheral blood.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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