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Rare Disorder Causing Infertility: Robertsonian Translocation 13:14

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ABSTRACT

Background & aim: Robertsonian translocation is a chromosomal structural anomaly resulting in infertility and miscarriage. Robertsonian translocation includes balanced and imbalanced types. The balanced type presents a normal phenotype and the patient may have reproductive issues such as recurrent abortion and birth of neonates with chromosomal abnormalities. In unbalanced type, partial monosomy or trisomy will appear, which can cause miscarriage, and congenital and developmental disabilities may occur in surviving fetuses.

Case report: The present study reported a female infant with suspicious minor labial adhesion, and imbalanced adrenal hormones, who was found to be a 45xx (13; 14) (q10; q10) balanced Robertsonian translocation compatible to the normal female phenotype.

Conclusion: Therefore, it is recommended to the family for prenatal amniocentesis in each future pregnancy. As this translocation can be a reason for infertility, genetic counseling and karyotype analysis may be helpful in the assessment of infertile adults without other presentations except recurrent abortion in women and oligospermia in men.

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Introduction

Translocations are the most common type of chromosomal structural anomalies; which are the result of breakages in the DNA, followed by incorrect rearrangement of the fragments. This translocation includes balanced and unbalanced types. In balanced type, there is no obvious loss of genetic material and usually the phenotype is normal; but adults may have reproductive issues such as recurrent abortion and birth of neonates with chromosomal abnormalities. In unbalanced type, the genetic material is lost and results in partial trisomy or monosomy, which could lead to spontaneous abortion and congenital and developmental disabilities may occur in surviving fetuses (1, 2). Although the prevalence of ROB is approximately 0.1%, but its rate is higher in women with the history of premature termination of pregnancy (1.1%) and infertile men (3%) (3).

Robertsonian (ROB) translocations are typically seen in acrocentric chromosomes 13, 14, 15, 21, and 22 (4). ROB translocation between chromosomes 13 and 14 (der (13; 14) (q10; q10)) compromise 75% of all cases of this type of rearrangement (2). In balanced type of translocation, individuals became carriers with healthy and normal lifespan and the translocation be may never discovered. But some individuals with der (13:14) unbalanced translocation can have higher risks of infertility, recurrent spontaneous abortions or chromosomally unbalanced fetus (5-6).

In this study, a female infant with Robertsonian translocation der (13:14) with suspicious

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presentation of adrenal hyperplasia was reported.

Case Report

A 30 days old girl was referred to a pediatric hospital in Mashhad, Iran for endocrine assessment regarding hormonal imbalance. She was the second child of a relative marriage, and the mother has no pervious history of fetal/ neonatal death or abortion. In past medical history, the mother mentioned that her pregnancy first screening test in 12 weeks was positive in terms of Down syndrome risk (PAPP-A = 1519.00 mIU/L, and Free β-hCG= 80.00 IU/L). Therefore, amniocentesis was done in the second trimester for more assessment, which was negative and pregnancy continued to birth. A female newborn was born through vaginal delivery with 3165 gr weight. At first 24hours physical examination of newborn, a suspicious labia minor adhesion was observed (Figure 1).

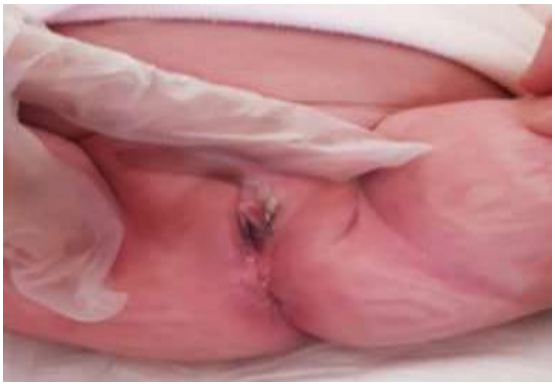

Figure 1. Suspicious minor labia adhesions

So hormonal tests was requested for adrenal hyperplasia evaluation. Some hormonal imbalances were observed, especially in the cortisol level (Table 1); thus, stimulation ACTH test, ultrasonography of abdomen, uterus, and adrenal glands, and karyotype analysis were ordered. Also, the patient referred to the pediatric endocrinologist. Labia adhesion was rejected in second physical examination. Further investigation showed normal levels of ACTH and cortisol (Table 1).

Table1. Hormonal test results

7 days olds		35 days old	
Test	**Value**	**Test**	**Value**
Androstenedione	3ng/ml,	17OH	4.6ng/mL
Cortisol AM	1.5 µg/dL,	Progesterone	42 pg/ml
ACTH	39.82 pg/ml,	ACTH	56.0 pg/ml
Aldosterone	77.9ng/dL,	ACTH 30min	54.0 pg/ml
Renin	36.32 pg/ml	after injection	4.0 µg/dL
17OH	11.69 ng/mL,	ACTH 60min	
Progesterone	0.70 ng/mL,	after injection	
Testosterone	124 µg/dL	Cortisol	
DHEA SO4			

Ultrasonography of uterus, adnexa, and adrenal glands showed no significant findings suggesting adrenal hyperplasia, nor pathologic lesions in adnexa. Uterus and ovaries were normal in size and volume and there was no free liquid in pelvic. But an important finding was achieved in karyotype analysis, a 45xx (13; 14) (q10; q10) Robertsonian translocation compatible to normal female phenotype was revealed (Figure 2).

According to the above findings, and normal level of cortisol for the third time (Cortisol AM

10.5 µ/dL normal), she just followed-up without receiving any medication. Also, parental chromosomal analysis and prenatal amniocentesis were recommended in the future pregnancies.

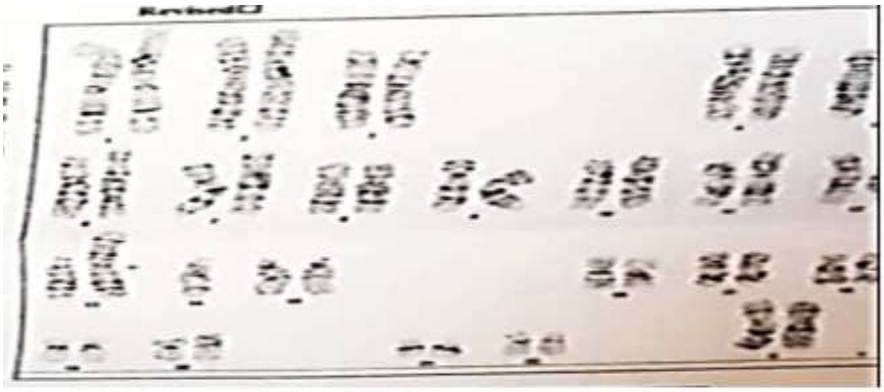

Figure 2. Chrosomal analysis

Discussion

Robertsonian translocation is a rare chromosomal anomaly which its carriers usually are phenotypically normal, but they can cause infertility problems including abortions in female adults and oligospermia in male adults (7, 8). Most of ROB translocations were inherited from normal phenotype mothers (2, 8), like the mother of the present case, who had normal phenotype without any history of abortion or child's death.

Also, ROB translocation is often discovered in adult patients when they were examined for infertility problems (1, 8). For example, in the largest discovered collection of balanced ROB in China, the main reasons for adults' referral were miscarriage and infertility. Moreover, in children cases, they were referred to be tested due to the presence of one or more of the following clinical findings: developmental delay, autism, dysmorphic features, seizures, or multiple congenital anomalies (8). But in the present case, this translocation was found when investigating the reason of suspicious labia adhesion and adrenal hormone imbalances, which could not be justified by adrenal hyperplasia. To our knowledge, ROB translocation der (13:14) in an infant with normal phenotype is a rare condition, and there is no pervious report of this rearrangement in Iran.

ROB carrier can produce offsprings with either a normal karyotype or unbalanced gametes through adjacent segregation of meiosis leading to increased risk of infertility, spontaneous miscarriage, and offsprings with unbalanced translocations (8). ROB translocations is a known chromosomal anomaly causing infertility with no treatment. Therefore, the present case was also discharged without any treatment and only was followed up until fertility period and future pregnancies. As with reciprocal translocation carriers, there is a risk of balanced Robertsonian translocations for producing unbalanced offspring (7), therefore, parents were recommended to investigate their chromosomal analysis and prenatal assessment by amniocentesis in each future pregnancies.

Conclusion

Since Robertsonian translocation is a condition which can cause abortion and infertility in normal phenotype carriers, it is suggested that a chromosomal analysis be done to identify this anomaly in females with recurrent abortion and males with infertility without any other pathological conditions.

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conflicts of interest

Authors declare no conflicts of interest.

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