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(CASE REPORT)

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Balanced translocations 46, XY, t (8;17)(p23;q11) in brothers with azoospermia

Mirela Mačkić-Đurović [1,*], Nejla Đikić [2] and Dunja Rukavina [3]

[1] *Center of Genetics, Faculty of Medicine, University of Sarajevo, Bosnia and Herzegovina.*
[2] *Faculty of Medicine, University of Sarajevo, Bosnia and Herzegovina.*
[3] *Department of Biology, Veterinary Faculty, University of Sarajevo, Sarajevo, Bosnia and Herzegovina.*

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Abstract

Infertility is one of the most significant human health problems of the reproductive years. The causes of infertility are diverse and numerous-including non-genetic and genetic factors.

A review of this case confirmed this. A balanced translocation was found in two siblings diagnosed with azoospermia. After being unable to conceive, the older brother and his wife had two *in vitro* fertilization (IVF) failed. At the same time, his younger brother and his wife had one IVF. After the cytogenetic analysis was performed in both pairs, it was shown that the cause of their infertility was the same balanced translocation in the brothers. The female showed a regular (46, XX) karyotype, whereas the male was found to carry balanced reciprocal translocation [46, XY, t(8;17)(p23;q11)].

This case support that cytogenetic analysis is still the first and basic diagnostic analysis of patients with azoospermia and other reproductive problems.

Keywords: Azoospermia; Balanced translocation; Karyotype

1. Introduction

According to the World Health Organization, infertility is a disease of the reproductive system defined by the failure to achieve a clinical pregnancy after 12 months or more of regular unprotected sexual intercourse (1). 20-30% of infertility was found in men (2). 50% of male infertility is currently unexplained and much of this likely is has a genetic basis. It is considered that 15% of men with azoospermia have abnormalities in chromosomal number or constitution (3). Zhang H et al. suggest that this condition can be the result of balanced translocations (4). Balanced translocations are chromosomal aberrations where one part of the chromosome broke off and reattached to another chromosome without losing any genetic material. Individuals with this translocation are called carriers of balanced reciprocal translocations. Studies have compared the chromosomal constitution of a normozoospermic carrier and azoospermic carrier of a balanced translocation. They revealed that synaptic pairing abnormalities in prophase are more frequent in azoospermic carriers (5). That means that azoospermic carriers have more chances to experience infertility or recurrent miscarriages. The present study reports cytogenetic analysis of a male patient (two brothers) with azoospermia and a carrier of an uncommon balanced translocation. According to the available literature, this translocation 46, XY, t(8;17)(p23;q11) was not found in men with reproductive problems.

* Corresponding author: Mirela Mačkić-Đurović
Center of Genetics, Faculty of Medicine, University of Sarajevo, Bosnia and Herzegovina.

2. Case report

Two brothers have come to the Center for Genetics with their wives. The older brother was 44 years and the younger 34. According to the doctor's findings, they could not get pregnant. According to the physician, both brothers had azoospermia, and their wives had never conceived. They also have two younger sisters and one older sister who had three miscarriages, but now she has a child. Since one of the leading causes of azoospermia is genetic, both brothers analyzed the microdeletion of the Y chromosome (AZF azoospermia factor region), and the finding showed that they do not have a microdeletion of that area. After this analysis, they decided on in vitro fertilization (IVF).

IVF is a method of assisted reproduction. In these couples was done by separating mature eggs and sperm, fertilizing in a laboratory under controlled conditions, and then transferring a pre-agreed number of embryos to the uterus through a painless ultrasound procedure.

After failed IVF in both couples, the physician instructed them to do a cytogenetic analysis as well. This analysis was performed on the peripheral blood, which was cultured for four days.

The karyotype of the couples was prepared using the G-banding technique with trypsin and Giemsa staining (GTG). Obtained the karyotype is discovered that both brothers have the same balanced chromosomal translocation between the short arm of chromosome 8 and the long arm of chromosome 17 [46, XY,(8;17)(p23;q11)] Figure 1. Their wives showed a normal karyotype 46, XX It is assumed that this translocation is familial. The other family members were invited to come to do their cytogenetic investigation to prove this assumption, but they never showed up. The patient has given written consent to the publication of her case in the newspaper. Of course, without the publication of her data and images.

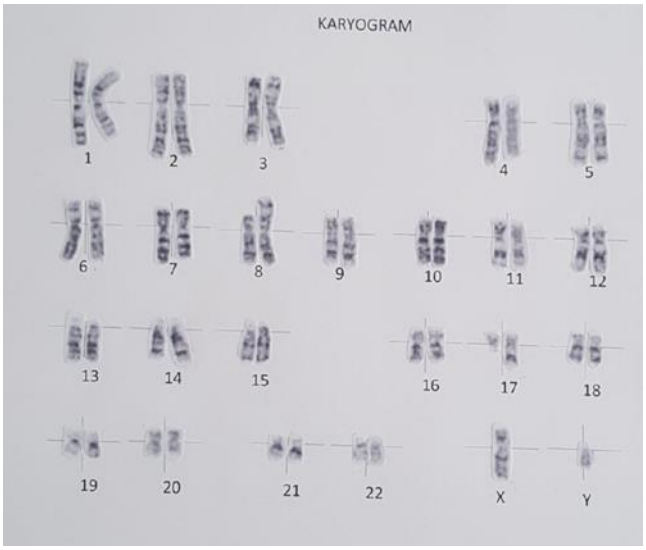

Figure 1. Karyotype showing balanced translocation 46, XY, (8;17)(p23;q11)

3. Discussion

Structural chromosomal abnormalities are a prevalent cause of recurrent miscarriages in men with infertility. The studies agree that the most common structural rearrangement is a translocation, found in about 5% of couples experiencing repeated losses (5,6). Also, these aberrations were primarily observed in men diagnosed with azoospermia and oligospermia. The effect of these aberrations is to block spermatogenesis, stop meiosis, and affect dosage-sensitive genes that stop spermatogenesis and cause infertility (7). Reciprocal translocations are related to azoospermia was also confirmed by our case in which translocation t (8; 17) was found in two brothers diagnosed with azoospermia. This is the first time that this translocation has been described in men with infertility. In the literature, this translocation (46, XX, t(8; 17)(p23; q21)) has been described as hereditary because it was a woman who had multiple miscarriages, and her siblings had the same translocation and problems with conception (8). Also, the study of couples from Northeastern Iran mentioned one 27-year-old woman who had 46, XX, t (8; 17) (q24.3; q21), and two abortions (6). Since it is a balanced translocation, the carriers of balanced translocation can produce phenotypically normal offspring. The report of scientist Zhang confirms this. He and his team reported a familial case of t (3; 6) where

the male carrier got a normal infant after two miscarriages (9). That can be an explanation of why the older sister, in our case, has a healthy living child.

In cases where men have azoospermia, as well as when women have recurrent miscarriages, they should do genetic counseling, as well as to determine their karyotype.

One of the solutions for carriers of balanced translocations is the preimplantation genetic diagnosis (PGD). PGD provides benefits for couples with high-risk translocations by reducing the risk of miscarriage and avoiding pregnancy with an unbalanced form of translocation (10).

4. Conclusion

In conclusion, our study supports the correlation between balanced translocation and azoospermia. Therefore, patients with azoospermia and other reproductive problems should do cytogenetic analysis and genetic counseling, allowing them to have healthy offspring.

Compliance with ethical standards

Disclosure of conflict of interest

There is no conflict of interest.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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