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TYPE OF ARTICLE: Case Report

TITLE: A novel reciprocal translocation t(2;14)(q11;q24) in a young woman with two times pregnancy losses: A case report

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TITLE: A novel reciprocal translocation t(2;14)(q11;q24) in a young woman with two pregnancy losses: A case report

ABSTRACT

Background
Recurrent miscarriage (RM) is defined as three or more loss of pregnancy before 24 weeks of gestation. It is one of the major regenerative problems all over the world, especially in young couples with history of pregnancy loss. The reciprocal translocations, especially balanced rearrangements are known to be one of the main causes of recurrent miscarriage.

Case presentation
We report results of clinical and cytogenetic analysis of a young couple with two pregnancy losses. Although the couple had a normal clinical study, the cytogenetic analysis of the wife revealed a balanced reciprocal translocation of t(2; 14) (q11; q24), whereas her husband has a normal 46, XY karyotype.

Conclusion
Based on literature search, this is the first report of t(2; 14) (q11; q24) translocation that is related to pregnancy loss. This translocation was found in couple with just two pregnancy losses. Early cytogenetic tests may help parents with pregnancy losses.

Keywords: Recurrent miscarriage, Reciprocal translocation, Karyotyping, G banding.
TITLE: A novel reciprocal translocation t(2;14)(q11;q24) in a young woman with two pregnancy losses: A case report

INTRODUCTION

Recurrent miscarriage is defined as three or more losses of pregnancy before 24 weeks of gestation[1]. It occurs in approximately 2% of fertile couples, so it must have been one of the main cause of sterility [2, 3]. Most women with recurrent miscarriage seem to have several risk factors for pregnancy loss.

Recurrent miscarriage has been directly associated with parental chromosomal abnormalities such as balanced translocations, maternal coagulation disorders like acquired maternal thrombophilia, anti-phospholipid antibodies syndrome (APS), severe preeclampsia, placental insufficiency and uterine structural anomalies. Recurrent miscarriage is also associated with maternal immune disorders such as excessive maternal immune response against paternal antigens, endocrine abnormalities like untreated hypothyroidism and poorly controlled diabetes mellitus[4–6]. Other important risk factors of RM are advanced maternal age, body mass index (BMI) > 25, BMI < 18, genital infections and environmental toxins[7]. However, these factors are present only in about 50% cases. In a vast majority of RM cases the pathophysiology remains unknown [8, 9].

We think that clinicians have extensive therapeutic choices for couples with RM such as medication, treat background disease, cytogenetic analysis or egg donation. We report a novel reciprocal translocation t(2; 14) (q11; q24), in a couple referred to our genetic clinic, for cytogenetic analysis and their diagnostic studies about cause of the RM.

CASE REPORT

We report a novel reciprocal translocation in a young couple, wife (20 years) and her husband (24 years). The couple was referred to our lab and the female partner gave a history of two miscarriages. Both miscarriages were in the first trimester of pregnancy. Both partners were physically and intellectually normal. The detailed family history was taken from the couple and revealed no significant positive genetic history (Figure 1). There was no history of abortion in the family of both husband and wife.
Clinical examination did not reveal any other abnormalities like anatomical defect, autoimmune, infectious and metabolic diseases.

Karyotype analysis was performed for both partners. Fifty Metaphase spreads were studied on the basis of GTG technique at high resolution banding (450–550 band), which revealed that female partner has a balance reciprocal translocation between the long arms of chromosome 2 and chromosome 14 as follow: [46, xx, t(2; 14) (q11; q24)] (Figure 2). The karyotyping of her husband showed that he was 46, XY (normal male karyotype).

**DISCUSSION**

Translocations are considered balanced if the transfer of genetic information between the chromosomes is reciprocal, and no genetic material is lost or gained[10]. When two non-homologous chromosomes break and exchange fragments, new chromosomes called derivative chromosomes are formed [11]. Balanced rearrangement is most frequent genetic disorders of human. It also appears in approximately 0.5% of normal population[12]. Reciprocal translocations are also the most common translocations in human and are found in about 1 out of 600 newborns[13].

The carriers of reciprocal translocation are usually unaffected because they have a normal complement of genetic material. However, they have an increased risk of having progeny with unbalanced karyotypes with interference in the meiotic segregation of their abnormal chromosomes[14]. Problems arise at meiosis because the involved chromosomes in the translocation cannot pair normally to form bivalents. Instead they form an unusual structure known as a pachytene quadrivalent[4] (Figure 3). There are few reports of t(2;14) associated with disease such as acute myeloid leukemia with t(2;14) (q22;q32)(15) and B cell lymphoma with t(2;14) (p11.2;q32)[16] and familial t(2;14) associated with autosomal dominant anterior polar cataracts[17].

**CONCLUSION**

In this case, we found a novel translocation t(2;14)(q11;q24), associated with recurrent miscarriage which has not been reported previously in literature. We think
that the new chromosomal rearrangement may be one of the causes of miscarriage in this young couple. We recommend that it is better to start genetic counseling in couples with two or more miscarriage so genetic studies should start earlier.

CONFLICT OF INTEREST
There is no conflict of interest in this case report

AUTHOR’S CONTRIBUTIONS
Mojtabahasanpour is the main analyzer. Mohammad Daghbash prepared the drafts. Mohamad Reza Farzanehis responsible for the case.

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REFERENCES

FIGURE LEGENDS

Figure 1: Pedigree of the affected family with the reciprocal translocation t (2; 14) (q11; q24).

Figure 2: The GTG banding Karyotype of illustrated case that shows reciprocal translocation t (2; 14) (q11; q24).

Figure 3: Possibilities for offspring of t (2; 14) reciprocal translocation carrier.

FIGURES

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