Case Report

A Novel t(10; 13) (q24; q14) in a female with recurrent miscarriage

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ABSTRACT

This case report presents a novel finding of 46, XX, t(10;13) (q24; q14) in a 35-year-old female with five spontaneous abortions. The only other abnormal finding was a low testosterone level. This report provides further information on the influence of balanced reciprocal translocations in recurrent miscarriage.

KEY WORDS: Recurrent miscarriage, Balanced reciprocal translocation, Testosterone.

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INTRODUCTION

Recurrent miscarriage (RM) is described as three or more consecutive spontaneous miscarriages before 20 or 28 weeks of gestation, and affects about 1–3% of women.[1] Causes of RM include genetic disorders, uterine pathologies, endocrine dysfunctions, autoimmune diseases, acquired and inherited thrombophilia, as well as environmental factors.[2] Parental chromosome abnormalities are reported in 4% of couples with RM but in only 0.2% of control couples.[3] The most common chromosome abnormality in RM couples is a balanced translocation, including reciprocal and Robertsonian translocations, which allow a normal parental phenotype but chromosomally unbalanced gametes.[4]

Here, we describe clinical and cytogenetic findings in a female with five spontaneous abortions. To our knowledge, this is the first report of an association of t(10;13) with RM.

CASE REPORT

A 35-year-old woman with five spontaneous abortions was evaluated at the Center for Reproductive Medicine. The First Bethune Hospital, Jilin University, China. Her family history was negative for hereditary disorders and her body mass index was 21.76 (weight: 55kg, height: 159cm). All the investigations were performed according to the established clinical criteria for RM.[4] A gynecological examination including transvaginal ultrasonography, hysterosalpingography, and endometrial biopsy revealed no abnormalities. Results of endocrine function testing, including the thyroid hormone and sex hormones of the follicular phase, are shown in Table-I. Immunological factors such as antiphospholipid antibodies, thyroid antibodies, and circulating immunocompetent cells, Prothrombotic studies, infectious factors, and basal serum glucose level were all normal. G-banded karyotypes of the patient and her partner were

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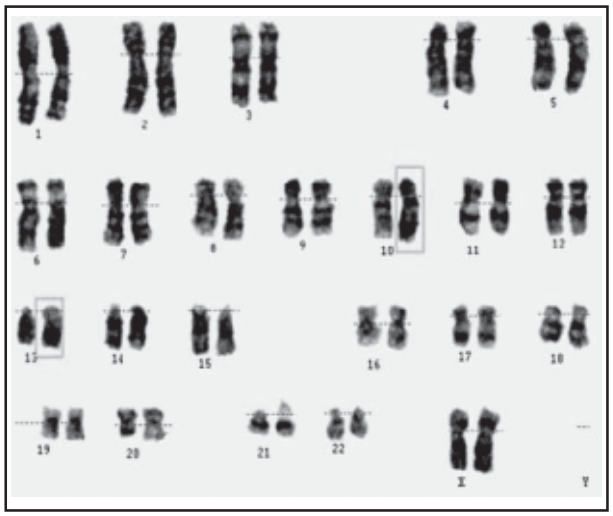

Figure-1: Karyotype of RM patient with t(10;13)(q24;q14) (magnification 1000X). The chromosomes involved in the reciprocal translocation are framed.

prepared from peripheral blood lymphocytes. The chromosomal abnormality was described according to the International System for Human Cytogenetic Nomenclature.

The only notable findings were the testosterone level (0.02 ng/ml), which was below the normal range of 0.06-0.82 ng/ml, and the karyotype, which was found to be 46, XX, t(10;13)(10pter?10q24::13q14?13 qter;13pter ?13q14::10q24?10qter) (Fig-1).

DISCUSSION

Recurrent miscarriage (RM) is a vexing complication of pregnancy. Although many factors were found associated with it, it still remains unexplained in nearly 50% of RM patients.[3] Chromosomal abnormality accounts for only 4% of RM cases, with the most common abnormality being balanced translocation.[4] In this report, we describe a balanced reciprocal translocation, t(10;13)(q24; q14), in a woman with RM.

One of the sites involved in this translocation, 10q24, has been reported in a man with t(10;17)(q24;p13) whose wife experienced RM.[5] The other site involved in this translocation, 13q14, has been reported in another RM case, a woman with t(7;13)(p22;q14).[6] These observations suggest that translocation of chromosomes 10 and 13 may influence normal gene expression, leading to RM.

A balanced reciprocal translocation is an exchange of material between two nonhomologous chromosomes, without loss or gain of material and balanced translocation carriers are usually healthy and phenotypically normal. During meiosis, the translocation chromosomes take on a quadrivalent conûguration.

Table-I: Sex hormone levels in the RM patient and normal range values.

Hormone	*RM patient*	*Normal range*
PRL (μIU/ml)	245	102 - 498
FSH (mIU/ml)	5.4	2.4 - 12.6
LH (mIU/ml)	4.5	3.5 - 12.5
E_2 (ng/ml)	0.089	0.0125– 0.166
T (ng/ml)	0.02	0.06 - 0.82
P (ng/ml)	0.6	0.2 - 1.5

PRL, Prolactin; FSH, follicle simulating hormone; LH, leutinizing hormone; E_2, estradiol; T, testosterone; P, progesterone.

The meiotic behavior of the quadrivalent configuration leads to the production of different proportions of balanced and unbalanced gametes, which may result in an adverse pregnancy outcome.[7] They have a higher risk of reproductive problems including infertility, IVF failure, spontaneous abortion, and having children with serious birth defects.[8] As the development of the artificial supplementary reproduction technique, IVF plus preimplantation genetic diagnosis (PGD) can increase the percentage of successful pregnancies in these couples.[9] Therefore, genetic counseling is important for these couples and their extended families.

High testosterone levels have been associated with RM. In this subject, however, the testosterone level (0.02 ng/ml) was below the normal range of 0.06 - 0.82 ng/ml for adult females. Women with low testosterone may have problems with libido, sense of well-being, or anemia, but there are no reports of an association between low testosterone and RM.[10] Our patient did not report any problems typically associated with low testosterone, and there is no evidence of a link between low testosterone and her abnormal karyotype.

This is the first report of the balanced translocation t(10;13)(q24;q14) and low testosterone in a subject with RM. The link between the translocation and low testosterone, if any, remains unrecognized.

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Authors Contribution:

Ru-Lin Dai and *Rui-Zhi Liu* made their contribution to the conception and analysis of the case.
Hong-Liang You, Chao Fu and *Yuan Dong* made their contribution to the acquisition, analysis and drafting the article.