Case Report

Balanced Reciprocal Translocation: Multiple Chromosome Rearrangements in an Infertile Female

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Abstract

Double reciprocal translocations and triple-balanced reciprocal translocations multiple chromosome rearrangements are very rare events in the phenotypically normal individuals. Chromosome analysis with 500-band resolution was performed and analyzed in an infertile woman. Her karyotype revealed 46,XX, t(1;3) (q44;p11), t(2;14) (q11.2;q13), t(9;11) (p22;p15) pattern in all the metaphases. To the best of our knowledge, this is the first case of triple-balanced reciprocal translocations in an infertile woman.

Keywords: Balanced reciprocal translocation, chromosome analysis, phenotype

Introduction

Balanced reciprocal translocations are one of the most frequent chromosomal aberrations with the incidence of 0.16%–0.2%, i.e., 1/615–1/500.1,2 Individuals having balanced reciprocal translocations are phenotypically normal, but there is a high risk of progeny getting affected with meiotic segregation of abnormal chromosomes. Apart from simple reciprocal translocations, rarely there are multiple chromosome rearrangements, where double or triple reciprocal translocations with two-way exchanges between chromosomes occur.

Reciprocal translocations occur due to the exchange of chromosome material between two nonhomologous chromosomes. When the amount of genetic material is balanced, there is no phenotypic effect on the individual because of a balanced complement of genes.

Reciprocal translocations can be inherited or can be de novo. The risk of having de novo translocations is greater than inherited ones, which showed the incidence of 6%–9%.3

In the present study, we report a case with triple-balanced translocations in an infertile woman with cytogenetic finding 46, XX, t(1;3) (q44;p11), t(2;14) (q11.2;q13), t(9;11) (p22;p15) pattern. To the best of our knowledge, this is the first such case in an infertile woman as per literature.

Case Report

A 35-year-old woman (MT) whose height was 4.11 inches and weight 48 kg with multiple in vitro fertilization (IVF) failures from infertility center were referred to cytogenetics laboratory for chromosome analysis. She is physically and mentally normal.

Cytogenetic analysis was performed using peripheral blood lymphocyte cultures through the modified standard protocol of Moorhead et al.4 The banded metaphase chromosomes were examined at a 500-band resolution. At least 30 metaphases were analyzed, and 5 or 6 well-spread plates were photographed and karyotyped (Ikaros; Metasystems, Altussheim, Germany). This study was approved by the Institutional Ethics Committee.

Chromosome analysis of the infertile woman showed three balanced-reciprocal translocations as 46, XX, t(1;3)(q44;p11), t(2;14)(q11.2;q13), t(9;11) (p22;p15) [Figure 1] pattern in all the cells analyzed.

Discussion

The chromosomal anomaly is an important cause of infertility. Balanced chromosomal translocations are an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

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often identified as a cause of infertility.[5] Balanced translocation carriers account for 0.08%–0.3% of the general population.[6] Thus, genome-wide arrays should be used for patients with a de novo translocation and an abnormal phenotype. From these findings, we might assume that in about 60% of reciprocal translocations associated with abnormal phenotypes, the breakage or the abnormal expression of dosage-sensitive genes cause the pathological phenotype. Hence, apparently balanced translocations often hide complex chromosomal rearrangements (CCRs). Similar findings have been reported by Gribble et al.[7] and Ciccone et al.[8]

A number of double reciprocal translocations and complex chromosome rearrangements such as involving three chromosomes in a translocation were reported,[9] but a triple reciprocal translocation is a rare event. Till today, there is one case reported with three balanced-reciprocal translocations involving six chromosomes in a male child with a history of short stature who showed karyotype as 46,XY, t(l;7)(q42;q22), t(5;9)(q31;q32), t(I 3;1 6)(q21;q22) pattern.[12] In another case, a young boy had three chromosome abnormalities such as trisomy 21,XXY and de novo reciprocal translocation with 48,XXY, +21, t(6;10) (p22–24;p12) karyotype.[13] Simioni et al.[14] reported a patient with de novo two apparently balanced reciprocal translocations and two partial monosomies, Madan[11] reported 103 CCR cases of which 18 were related to infertility where complex chromosome rearrangement is due to three-way and four-way translocations involving three and four chromosomes and three to six break points.

In our study, we have a woman who had many IVF failures presented with triple-balanced reciprocal translocations. There is no theoretical explanation for the segregation ratios of human reciprocal translocations since the relative frequencies for the orientation is unknown. It is possible to have three separate reciprocal translocations in a single event arising either in zygote or meiotic error in the parental gametes.[12]

Further, genetic counseling and prenatal diagnosis would be essential to this woman if IVF treatment is successful.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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