A rare balanced nonrobertsonian translocation involving acrocentric chromosomes: Chromosome abnormality of t(13;15)(p11.2;q22.1)

ABSTRACT

BACKGROUND: Balanced non-robertsonian translocation (RT), involving acrocentric chromosomes, is a rare event and only a few cases are reported. Most of the RTs are balanced involving acrocentric chromosomes with the breakpoints (q10;q10).

MATERIALS AND METHODS: Chromosome analysis was performed as per standard procedure – Giemsa-trypsin banding with 500 band resolution was analyzed for chromosome identification. RESULTS: In the present study, we report a balanced non-RTs involving chromosomes 13 and 15 with cytogenetic finding of 46, XX, t(13;15) (p11.2;q22.1). CONCLUSION: To the best of our knowledge, this is the first such report of an unusual non-RT of t(13;15) with (p11.2;q22.1) break points.

KEY WORDS: Chromosome analysis, nonrobertsonian translocation, preimplantation genetic diagnosis

INTRODUCTION

Robertsonian translocations (RTs) are the most common balanced structural chromosomal abnormalities in the population involving (often nonhomologous and rarely homologous) acrocentric chromosomes with an incidence of 1.23/1000 live births. In RT, the pericentric regions of two acrocentric chromosomes fuse to form a single centromere or two. The resulting balanced karyotype has only 45 chromosomes including the translocated one which is the result of a fusion of the long arms of two acrocentric chromosomes. Balanced translocations have been determined as 0.2% in the neonatal population, 0.6% in infertile couples, and 9.2% in cases who have recurrent miscarriages. Phenotypically, carriers of balanced reciprocal translocation will be normal. These individuals have a high reproductive risk of having abnormal embryos due to chromosomal imbalances during meiosis, leading to birth of affected offspring or recurrent miscarriages.

Balanced non-RT involving acrocentric chromosomes is a rare event. In the present study, we report a case of rare non-RT involving chromosomes 13 and 15 with cytogenetic finding of 46, XX, t(13;15) (p11.2;q22.1). To the best of our knowledge, this is the first report of t(13;15) with this breakpoint, which is not usual RT.
RESULTS

A couple, with 35-year-old wife and 37-year-old husband, was referred to our center for chromosome analysis with the history of two miscarriages and one congenitally abnormal fetus. The couple was normal physically and intellectually.

Chromosome analysis revealed normal male karyotype in husband, whereas the female partner showed balanced reciprocal translocation as 46, XX, t(13;15)(p11.2;q22.1) [Figure 1]. This translocation is different from the usual RT of acrocentric chromosomes. The satellite region of chromosome 13 translocated to long arm of chromosome 15 and part of chromosome 15 translocated to chromosome 13.

DISCUSSION

Couples with balanced reciprocal translocation and RTs have 50% chance of having spontaneous abortions and 20% risk of having children with abnormal genetic makeup.

Usually, we find acrocentric chromosomes 13, 14, 15, 21, and 22 as RT in couples with a bad obstetric history. In the present study, we observed acrocentric chromosomes 13 and 15 as a rare balanced non-RT, where satellites of chromosome 13 translocated to chromosome 15 and part of chromosome 15 were translocated to chromosome 13. Non-RT involving acrocentric chromosomes is a rare event, and only a few cases have been yet reported.

Frikha et al.[6] reported a familial non-RT between the long arm of chromosome 15 and the long arm of chromosome 21 with 46, XY, t(15;21)(q21;q21) in an infertile man and his hypofertile brother whose wife had a history of repeated pregnancy loss.

Baruffi et al.[7] reported Down syndrome features case with non-RT involving chromosomes 15 and 21 as 46, XX, der (15) (15pter→15q26.2:21q11.2→21qter). Abeliovich et al.[8] described the non-RT karyotype t(15;21)(q15;q22.1) pat in two siblings. Mangelschots et al.[9] reported a case with 46, XX, t(13;15)(q12;q13) where she gave birth to a child with karyotype 46, XX,-13, +der (15) (13q13;15q12) mat. Most of the RTs are balanced involving acrocentric chromosomes with the breakpoints (q10;q10).

The formation of normal, balanced, and unbalanced gametes is dependent on the chromosomes involved and on the location of breakpoints. The configuration of the quadrivalent at the pachytene stage may alter the pattern of segregation, resulting in normal, balanced, and unbalanced gametes. Balanced chromosomal translocations may alter the expression of the functional genes which could result in the reproductive errors.[10] Further analysis of the breakpoints and molecular characterization of genes involved might enlighten the understanding of the basis of repeated abortions.

To detect structural chromosomal abnormalities in couples having assisted reproductive treatment, preimplantation genetic diagnosis method has become very important as an alternative to invasive prenatal procedures to diagnose the structural abnormalities in the offspring transmitted from one or both of the parents.[11] Cytogenetic analysis is the gold standard method to detect chromosome abnormalities. Advanced molecular cytogenetic techniques such as spectral karyotyping have opened up an era in detecting cryptic translocations, insertions, and deletions.[12]

Acknowledgement

The authors are grateful to the SRL Limited for providing infrastructure facilities.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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