

A CASE REPORT ON MATERNAL t (1;2) (q11; p11.1) AND ITS INHERITANCE



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ABSTRACT

Balanced translocation in one of the partners may form faulty gametes and so it may be associated with infertility or recurrent miscarriages. Although, if same balanced translocation gets inherited then it gives rise to normal child.

We report a case of non-consanguineous couple with the history of spontaneous abortion with bilateral dysplastic kidney in the foetus. Karyotyping of husband was normal and wife showed autosomal balanced translocation between q arm (11) of chromosome 1 and p arm (11.1) of chromosome 2. After doing genetic counselling for prenatal diagnosis, couple underwent prenatal diagnosis by chorionic villous and amniotic culture in two pregnancies respectively. In the first pregnancy, the female foetus inherited same translocation which mother carried and in the second pregnancy, male foetus inherited normal chromosomes from mother. Genetic counselling was done during both the pregnancies and couples continued the pregnancy.

Inheritance of balanced translocation and normal chromosomes in next generation from mother having balanced translocation is rare to happen. Proper genetic counselling for prenatal diagnosis will definitely help the couple to have normal children.

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1. INTRODUCTION

Translocation is structural chromosomal abnormality where exchange of genetic material takes place between two chromosomes. When such aberration is seen in acrocentric chromosomes then it is called as reciprocal translocation and when it involves autosomes then it is called as reciprocal translocation. Part of the chromosome gets separated because of cut in p or q arm. Part of the chromosome distal to breakpoint gets dislodged and gets attached to other chromosome which leads to



formation of derivative chromosome. Unless one or both of the chromosomal break-points involve an important functional gene, these balanced chromosomal rearrangements would not produce a significant phenotypic effect either [Paththivige \(n.d.\)](#). Parents will be phenotypically normal when there is absence of genetic dosage gain or loss in the process of translocation. Chromosomal reciprocal translocations frequently present with reproductive failure or recurrent pregnancy loss [Paththivige \(n.d.\)](#).

In approximately 2%–5% of couples with recurrent miscarriages, one of the partners carries a balanced structural chromosomal anomaly, most commonly a balanced reciprocal or a Robertsonian translocation [Dutta et al. \(2011\)](#). Carriers of balanced translocation may give rise to gametes which may inherit parental balanced translocation or normal chromosomes or unbalanced chromosome. Unbalanced gamete may consist of one normal chromosome and another derivative chromosome.

Karyotyping is being done to find out the cause of chromosomal aberrations in recurrent pregnancy loss. Genetic counselling and prenatal diagnosis in case of parental balanced translocation helps to have normal pregnancy outcome.

2. CASE

We report a case of non-consanguineous couple with the history of spontaneous abortion with bilateral dysplastic kidney in the foetus. Karyotyping of husband was normal and wife showed autosomal balanced translocation between q arm (11) of chromosome 1 and p arm (11.1) of chromosome 2. After doing genetic counselling for prenatal diagnosis, couple underwent prenatal diagnosis by chorionic villous and amniotic culture in two successive pregnancies respectively. In the first pregnancy, the female foetus inherited same translocation which mother carried and in the second pregnancy, male foetus inherited normal chromosomes from mother. Genetic counselling was done during both the pregnancies and couples continued the pregnancy and normal pregnancy was achieved.

3. METHODOLOGY

1. Chromosomal analysis of peripheral blood of husband and wife was conducted by conventional karyotyping
2. Prenatal diagnosis of 1st pregnancy was done by using chorion villous sample at 9 wks by direct culture method
3. Prenatal diagnosis of 2nd pregnancy was done by using amniotic fluid at 12 wks by flask method.

4. DISCUSSION

Balanced translocations are the most commonly detected chromosomal abnormality in couples with recurrent pregnancy loss [Pritti and Priya \(2018\)](#). Incidence of balanced translocation is 0.29% [Vasilevska \(2013\)](#). Couples with balanced translocation have 50% chance of having recurrent pregnancy loss and a 20% risk of having children with unbalanced chromosomal aberrations [Wirth \(1996\)](#). Inheriting normal chromosomes, balanced chromosomes and or derivative chromosomes in the offspring depends upon the gametes formed which undergo fertilization.

The formation of balanced, unbalanced and normal gamete is dependent on the basis of breakpoint and chromosomes involved. The larger imbalance will most likely lead to miscarriage whereas the subtle or smaller imbalance will increase the risk of having offspring with unbalanced karyotype [Farcas \(2007\)](#). This will lead to pregnancy outcome as either normal, carrier or abnormal progeny. Gametogenesis of the carriers is affected by forming trivalents (Robertsonian translocations) or quadrivalents (reciprocal translocations) between translocated and normal chromosomes. The type of gametes produced in meiosis depends on the mode of chromosome segregation [R. Gardner and Sutherland \(2004\)](#).

Unbalanced gametes will lead to early pregnancy loss or congenital malformations in the foetus. Association of presence of balanced translocation in one of the partners with congenital anomalies in foetus [Dutta et al. \(2011\)](#). In the present case, first pregnancy loss was because of spontaneous abortion with bilateral dysplastic kidney in the foetus. Maternal inheritance of derivative chromosome in the gamete might have led to the formation of congenital abnormality i.e., bilateral dysplastic kidney in the foetus.

The couples which give history of pregnancy loss and one partner shows balanced reciprocal translocation, are strongly recommended to undergo prenatal diagnosis for next pregnancy. The risk of detecting an abnormality is higher at prenatal diagnosis than it is at the birth of live baby [M. K. Gardner et al. \(2012\)](#). Prenatal diagnosis can be done by culturing amniotic cells and or chorionic tissue.

In the present case the couple was counselled to undergo prenatal diagnosis. Couple underwent prenatal diagnosis by chorionic villous and amniotic culture in two successive pregnancies respectively. In the first pregnancy, the female foetus inherited same translocation which mother carried and in the second pregnancy, male foetus inherited normal chromosomes from mother. It was reported before where the daughter inherited same balanced translocation from mother [Vasilevska et al. \(2013\)](#). It was noted that if the same (balanced) karyotype found in the carrier parent was detected at prenatal diagnosis, there was no increased risk of phenotypic abnormality in the child [R. Gardner and Sutherland \(2004\)](#). It would be appropriate to advise continuing a pregnancy when the foetal karyotype is the same as that of carrier parent and with very considerable confidence of normal outcome [M. K. Gardner et al. \(2012\)](#). On the other side one case was reported where mother carrying balanced translocation had an abnormal child in whom the same apparently balanced

karyotype had been shown at prenatal diagnosis [Prontera et al. \(2006\)](#). This could be because of concept of position effect [M. K. Gardner et al. \(2012\)](#) in which a particular gene in the close vicinity of a translocation breakpoint may function normally in parent but in the child, the gene in question may be silenced, due to an effect of the adjacent chromatin of the other participating chromosome [M. K. Gardner et al. \(2012\)](#) and there are mechanisms where apparently balanced translocation may have phenotypic consequences in the progeny of translocation carriers. These are: cryptic unbalanced defect [Wagstaff and Hemann \(1995\)](#), post zygotic loss of a derivative chromosome in one cell line [Dufke et al. \(2001\)](#) and uniparental disomy.

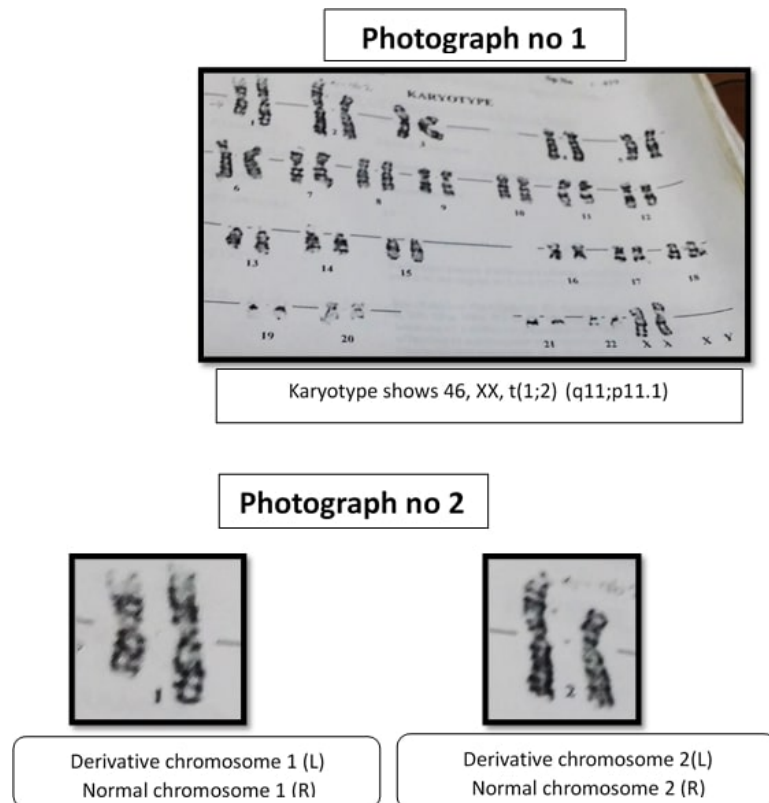


Figure 1

Vast literature search revealed translocations involving chromosome 1 and 2 but we have not come across balanced translocation between 1 and 2 chromosomes. This is very rare reciprocal translocation. Prenatal diagnosis and genetic counselling help the couple for achieving normal pregnancy outcome.

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