Case Report

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Pregnancy Outcome of a Case with a Rare Chromosomal Balanced Translocation 46XX t(4;13)

OBST & GYN Section

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ABSTRACT

Chromosomal translocations are rare. The unbalanced translocations may lead to structural malformations. The balanced translocations may not result in phenotypic alterations, as there is no loss or gain of genetic material. We are reporting here, a case of a balanced translocation carrier 46XX t(4;13) and her pregnancy outcome. Ultrasound at 11-12 weeks of gestation for determining the nuchal translucency thickness and for prenatal genetic testing might be considered as a part of the investigations for pregnancies with a parental balanced chromosomal translocation.

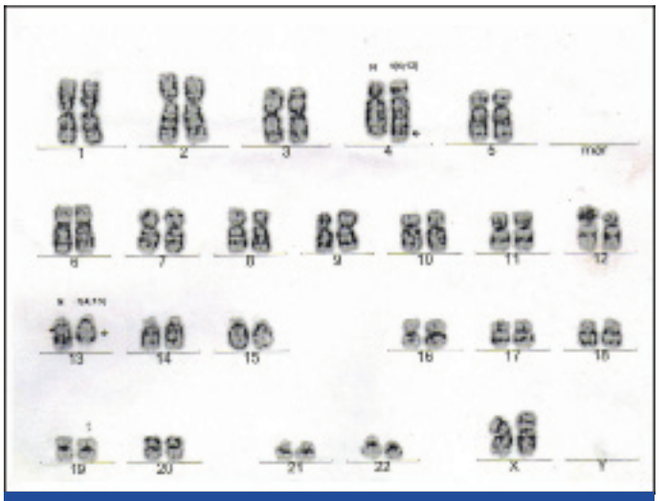
Key Words: Chromosomal translocation, Duplication 4q, Nuchal translucency

INTRODUCTION

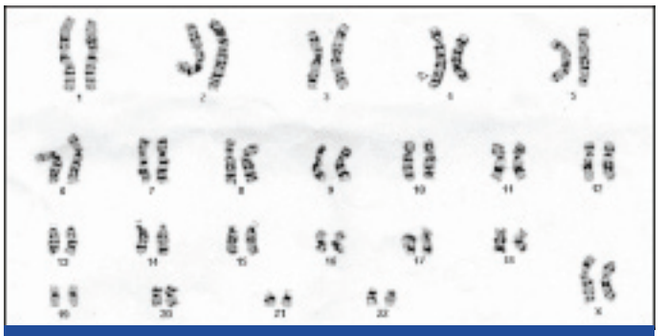
Chromosomal translocations are uncommon. They result from a chromosomal partition which is followed by a reunion in a different configuration. Chromosomal translocations may result in infertility, spontaneous abortions and congenital malformations. The balanced translocations may not result in phenotypic alterations, as there in no loss/gain of the chromosomal material. The unbalanced rearrangements may lead to malformations. We are reporting here, a rare case with a chromosomal balanced translocation t(4;13) and her obstetric outcome.

CASE REPORT

A 29-year-old, gravida-7, para-3, living-1, abortion-3 presented to the obstetric outpatients department on 18/02/2010, with two months of amenorrhoea. She had a non-consanguinous marriage to a 36-year-old man. Her first two pregnancies had resulted in spontaneous abortions during their first trimesters. Her third pregnancy had continued till term and she had delivered a male child with post-axial-polydactaly, an undescended testis, brachycephaly and a high arched palate, who died after a month of his birth. Karyotyping of the patient revealed 46XX chromosomes with a balanced translocation t(4;13). Her husband had a normal male karyotype. During the fourth pregnancy, an amniocentesis which was done at 16 weeks revealed extrachromosomal material on chromosome-4, with 44 autosomes and 2 sex-chromosomes. The couple refused termination of the pregnancy. A female child was born at term with postaxial polydactaly. The follow-up of the child revealed delayed physical and mental milestones. Her karyotype at one-year of age revealed 46XX 4q+, which was consistent with her amniotic fluid karyotype. During the mother's fifth pregnancy, ultrasonography (USG) which was done at 13 weeks revealed increased nuchal translucency (NT=4.8mm), polydactaly and an echogenic focus in the heart. The chromosomal analysis of the amniotic fluid showed extra chromosomal material on chromosomal. This pregnancy was terminated with the consent of the couple. Her sixth pregnancy continued till term as her USG at 12 weeks revealed no abnormality, and her amniotic fluid analysis showed a balanced translocation 46 **t(4;13)(q35;q22) with no apparent loss of the genetic material. The child was born as a phenotypically normal female baby. The present pregnancy was her



[Table/Fig-1]: Karyotype of the patient showing balanced translocation 46XX t(4;13)



[Table/Fig-2]: : Karyotype of patient's offspring showing extrachromosomal material on chromosome 4.

seventh pregnancy. An USG at 13 weeks revealed no abnormality. The patient refused chorionic villous biopsy and amniocentesis. Her subsequent scans revealed no abnormalities. She delivered a full term phenotypically normal female child on 11/10/2010.

DISCUSSION

Chromosomal translocations have been implicated in cases of infertility, recurrent spontaneous miscarriage and congenital malformations [1]. Chromosomal translocations result from a chromosomal partition which is followed by reunion in a different configuration. The clinical manifestations are severe in cases where there are unbalanced rearrangements, due to the altered amounts of the chromosomal material. Phenotypic consequences do not occur in balanced translocations, as there is no loss/gain of the genetic material, unless the break points affect a functional gene [2]. It may be appropriate to continue a pregnancy when the same balanced karyotype which is found in the carrier parent is detected in the foetus, as there is no increased risk for a phenotypic abnormality [3].

The production of a high portion of gametes with an unbalanced genetic complement may increase the risk of spontaneous abortions. Significantly, a higher number of miscarriages were noted in cases of male carriers as compared with the cases of female carriers [3].

Increased foetal nuchal translucency (NT) thickness is a useful ultrasonographic marker for the screening of foetal aneuploid conditions, particularly chromosomal trisomies. The increased foetal nuchal translucency (NT) thickness in foetuses with unbalanced chromosomal translocations is greater than the thickness in foetuses with a balanced translocation or a normal karyotype [3]. The NT thickness at 11-12 weeks of gestation may be a sensitive, first-trimester predictor of an unbalanced chromosomal translocation in the parental translocation carriers. The NT could serve as a reference ultrasound marker in these balanced translocation patients to undergo invasive prenatal tests for their diagnosis [3]. It has been recommended that pregnant couples in which one partner carries a chromosomal translocation have an early prenatal genetic testing by using chorionic villous sampling [4].

The most common findings in the duplication-4q cases according to a study were: low set and/or malformed ears, microcephaly, hypertelorism, a depressed/wide-bridged nose, a down turned mouth, malformation of the digits, urinary tract abnormalities and developmental/mental retardation [5]. In our patient, the findings which were caused due to the duplication of 4q from her records were: postaxial-polydactaly, delayed physical/mental milestones, increased-NT and an echogenic-focus in the cardia. A balanced translocation t(4;13) in the offsprings of the patient revealed no phenotypic abnormalities.

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