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CASE REPORT

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Reciprocal autosomal translocation t(5;14) (q32;q32) in a patient with secondary amenorrhea

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Introduction

Secondary amenorrhoea (SA) is a disorder characterized by the absence of menstrual period extending over 6 months in women with regular menstrual cycle or over a period of 1 year in women reporting irregular periods. The incidence of SA is reported to be about 1%. SA is known to have association with structural anomaly of X-chromosome or mosaicism involving X-chromosome. Temocin et al[1] reported an instance of mosaicim (46,XX/ 47,XXX) in a patient with SA. Another case of SA described by Tarkane et al [2] exhibited the karyotype, 46,X,t(X;X). Calvano et al [3] reported a case of SA showing complex mosaicism. Instances of X-autosomal translocations are also on record. Phelan et al [4] reported t(X;4), Sauer et al [5] observed t(X;7), Fournier et al [6] reported t(X;1) and Banerjee et al [6] recorded t(X;22) in instances of SA. Association of X-chromosome with SA is known. A woman with recurrent miscarriages exhibited reciprocal insertions between chromosome 7 and 14 (Wang et al [7]. Translocation between autosomes associated with SA appears to be a rare phenomenon. Philip et al [8] reported a case of SA showing t(14;21). We describe here a unusual case of SA exhibiting a translocation between two autosomes 5 and 14.

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Case report

A 21-year-old woman with SA was referred for chromosomal evaluation. She was born to nonconsanguineous parents when her mother was 27 year old and father 36 year old. She attained menarche at the age of 14 years and developed amenorrhea at the age of 20 years. Her two elder sisters were normal. She exhibited normal secondary sexual characters and there was no family history of Turner syndrome, amenorrhea, infertility, recurrent abortions, exposure to medications. Her height was 147 cm, a little less than the average for females viz., (150 to 170 cm). On sonography the size of the uterus was 6.1 x 3.3 cm. The right ovary was normal in size and the left ovary was hypoplastic measuring 1.5 x 0.7 x 0.6 cm. The level of follicular stimulating hormone estimated from a pooled sample was 71.24 IU/ L.

Chromosomal analysis was done by leucocyte culture method. Peripheral blood was collected in a 5mL heparinized syringe. 0.5mL of the blood was added to 5mL of RPMI medium with 1 mL of AB serum, antibiotics and 0.2 mL of phytohemagglutinin and was cultured at 37°C for 72 hours. 0.01 mL of 0.01% colchicine was added to arrest the dividing cells at metaphase. Chromosomal preparations obtained were subjected to giemsa banding technique [9,10]. Analysis of chromosomal preparations revealed the presence of a reciprocal autosomal translocation between autosomes 5 and 14 [46,XX,t(5;14)(q31;q32)]. (Figure 1).

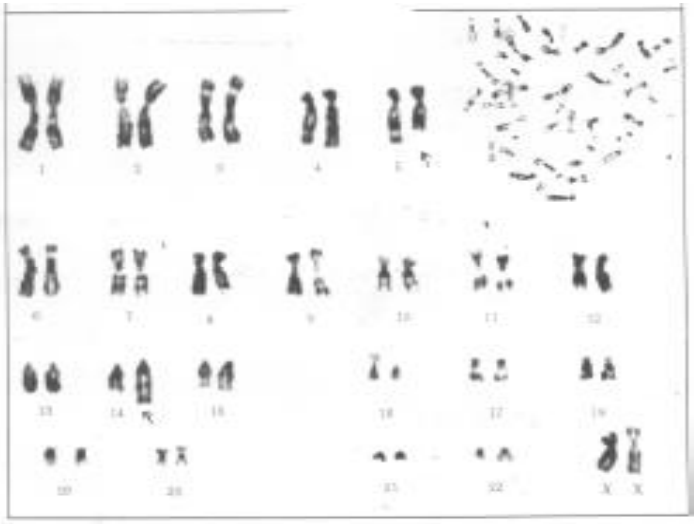

Figure 1. A reciprocal autosomal translocation t(5; 14) (q31; q32).

Discussion

Most reported cases of SA exhibited either a structural anomaly in the X-chromosome or a translocation between X-chromosome and an autosome. The only other report available on translocation between autosomes associated with SA was that described by Philip et al [8], involving chromosome 14 and 21 in a Robertsonian translocation. In the absence of good sample size, it may be premature at present to implicate this reciprocal translocation as having an etiological role.

The role of autosomal involvement in the etiology of SA needs to be elucidated with a large number of samples. Our patient had no plans about marriage. She was counseled about having normal married life, low chances of pregnancy, and need for hormonal support and prenatal diagnosis in case of pregnancy.

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