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Short communication

X monosomy and balanced Robertsonian translocation in a girl with Turner Syndrome

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Abstract

We describe a case of X monosomy associated with a maternally inherited t(13;14) Robertsonian translocation in a girl with Turner syndrome. The girl's X chromosome was demonstrated to be maternally inherited, ruling out the hypothesis that the translocation exerted an interchromosomal effect on the origin of the monosomy. Chromosomes 13 and 14 showed biparental inheritance.

Key words: Robertsonian translocation, X monosomy, interchromosomal effect.

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It has been suggested that chromosomal rearrangements may disturb meiotic disjunction of chromosomes not involved in the rearrangement, resulting in an interchromosomal effect (Hamerton *et al.*, 1968; Subri *et al.*, 1980). We describe a case of X monosomy associated with a maternally inherited t(13;14) Robertsonian translocation in a girl with Turner syndrome. This study was approved by the ethics committee of our institution and informed consent was obtained from the parents of the girl.

The five-year-old female proband was referred to our clinic because of short stature and facial dysmorphism. Clinical examination revealed short stature (< 2.5 percentile), weight between 2.5 and 10 percentile, flat frontal, large and prominent low-set ears, papebral ptosis, high nasal bridge, micrognathia, long philtrum, high palate, clinodactyly of the fifth left digit, and hypoplastic toe nails. Neuropsychological development was appropriate for the age. Chromosome analysis showed monosomy X and a Robertsonian balanced translocation - 44,X,der(13;14)(q10;q10) karyotype. The girl's mother (who had a history of miscarriage) carried a balanced translocation but her father's karyotype was normal.

To verify whether or not the Robertsonian translocation influenced non-disjunction of the X-chromosome, we determined the parental origin of the girl's X chromosome and normal chromosomes 13 and 14. Polymerase chain reaction was performed using DNA extracted from peripheral blood lymphocytes. We analyzed the CAG repeat on exon 1 of the X chromosome androgen receptor (*AR*) gene (Bharaj *et al*., 1999) and polymorphic markers on chromosome 13 (D13S787 and D13S895) and 14 (D14S592, D14S608 and D14S617). Our analysis showed both maternal inheritance of the X chromosome (Figure 1) and biparental inheritance of chromosomes 13 and 14.

Three previous cases of monosomy X associated with a t(13;14) translocation have been reported. Salamanca *et al.* (1985) reported a case of maternally inherited t(13;14) translocation but did not investigate the parental origin of the X-chromosome non-disjunction. Laszlo *et al*. (1984) reported the case of a patient, his mother and sister, who all had a t(13;14), but found no evidence of an interchromosomal effect. Krajinovic *et al*. (1994) found that both the X chromosome and the t(13;14) translocation in one of their patients were paternally inherited and thus demonstrated that the translocation had no effect on X chromosome non-disjunction.

Kondo *et al*. (1979) compared the expected and observed frequency of the 45,X karyotype combined with unrelated balanced translocations and concluded that there was no causal relationship between the two chromosomal abnormalities

In our patient both the translocation and the X chromosome were demonstrated to be maternally inherited, ruling out a meiotic non-disjunction interchromosomal effect.

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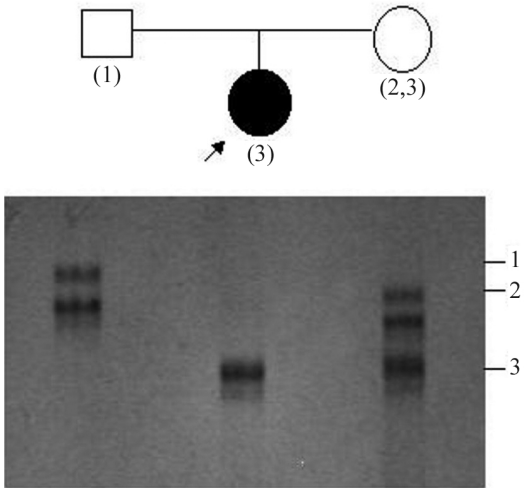

Figure 1 - The androgen receptor (*AR*) gene CAG repeats of a 45,X girl (arrow) and her parents were genotyped by PCR and electrophoresis using 6% denaturing polyacrylamide gel. The allele on the girl's X chromosome was inherited from her mother.

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