Isochromosome Xq In A Girl Having Delayed Puberty

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We determined isochromosome Xq karyotype in a 14-year-old female patient having delayed puberty. The diagnosis of infertile Turner's syndrome was made later, based on the presence of the primary amenorrhea and additional features such as growth retardation (her height and weight were 132 cm and 45 kg, respectively) and unusual hormonal profile. She had high plasma gonadotropin and low ovarian hormone levels (LH: 42 mlU/mL, FSH: 14 mlU/mL, estradiol: 27 pg/mL, and progesterone: 0.6 ng/mL). Karyotypes in the peripheral blood cells were pure isochromosome Xq constitution; 46,X,i(Xq). Ultrasonography showed that uterus dimensions were 3x2x2 cm and ovaries were streak. The patient did not show other classical Turner's syndrome traits such as broad chest, neck webbing, low posterior hairline, renal and cardiovascular anomalies. [Journal of Turgut Özal Medical Center 1997;4(1):109-111]

Key Words: *Turner's syndrome, isochromosome Xq*

Puberte gecikmesi olan 14 yaşındaki bir kız hastada izokromozom Xq karyotipi

Puberte gecikmesi şikayeti bulunan 14 yaşındaki bir kız hastada izokromozom Xq karyotipi saptandı. Daha sonra, primer amenore yanında gelişme geriliği (boy: 132 cm, ağırlık: 45 kg) ve anormal hormonal bulguları da göz önüne alınarak hastaya izokromozom X' e bağlı Turner sendromu tanısı konuldu. Hastada plazma gonadotropinleri yüksek iken ovaryan hormon düzeyleri düşüktü (LH: 42 mlU/mL, FSH: 14 mlU/mL, östradiol: 27 pg/mL ve progesteron: 0.6 ng/mL). Periferik lenfositlerden elde edilen tüm metafazlarda 46,X,i(Xq) karyotipi tespit edildi. Ultrasonografi sonucu uterus boyutlarının 3x2x2 cm olduğu anlaşıldı. Hastada, geniş göğüs kafesi, yele boyun, düşük posterior saç bitim çizgisi, renal ve kardiyovasküler anomaliler gibi Turner sendromuna ait diğer klasik bulgular gözlenemedi. [Turgut Özal Tıp Merkezi Dergisi 1997;4(1):109-111]

Anahtar Kelimeler*: Turner sendromu, izokromozom Xq*

Turner's syndrome is an aneuploidy, whose most frequently observed chromosome constitution is 45, X0 with no second sex chromosome, either X or Y. However 46, X, i(Xq) is also seen with a frequency of 10% in all cases with Turner's syndrome.

The overall incidence of Turner's syndrome is approximately 1 in 5000 female births. The frequency in conception is much higher but over 90% of which are spontaneously aborted. The newborns with Turner's syndrome can often be identified at birth by their distinctive phenotypic features (1,2).

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The major traits of classical X0 Turner's syndrome are female phenotype, short stature, usually lower than 150 cm height, streak gonads, primary amenorrhea, hypoestrinism and sterility, short- broad neck with webbing and cardiovascular and/or renal anomalies. The chest is also broad with widely spaced nipples and secondary sex characteristics fail to develop spontaneously (1-3).

The chromosome constitution is also clinically significant in this syndrome. Patients with i(Xq) are like classical 45, X0 patients, whereas patients with a deletion of Xp have short stature and congenital malformations and those with a deletion of Xq have often only gonadal dysfunction (3).

In this paper, we aimed to present a case having 46, X, i(Xq) karyotype without clear features of Turner's syndrome except primary amenorrhea.

Figure 1. The case with i(xq) Turner's syndrome

CASE AND DISCUSSION

The case was 14 years old and phenotypically female having the basic symptoms growth retardation and absence of mens (primary amenorrhea). She was 132 cm tall and 45 kg weight (being in 3^{rd} and 10^{th} percentiles respectively) (Figure 1).

The bone age was found to be 12. Ultrasonographic findings showed that uterus dimensions were 3x2x2 cm and ovaries were streak.

Plasma gonadotropin levels were high (LH: 42 mlU/mL, FSH: 14 mlU/mL) but estradiol and progesterone were low (27 pg/mL and 0.6 ng/mL respectively). This hypergonadotropic hipogonadism phenomenon can be explained with absence of feedback mechanism due to lack of ovarian functions. On the other hand, the present case did not have broad chest, neck webbing, low posterior hairline, and renal or cardiovascular anomalies as in classic Turner's syndrome.

In cytogenetic analysis, 50 well-spreaded, G-banded metaphase plates were evaluated and all metaphases showed 46, X, i(Xq) karyotypic constitution (Figure 2).

Some reports have indicated that the patients with 46,X,i(Xq) karyotype demonstrated similar features as classical Turner's syndrome. It is claimed

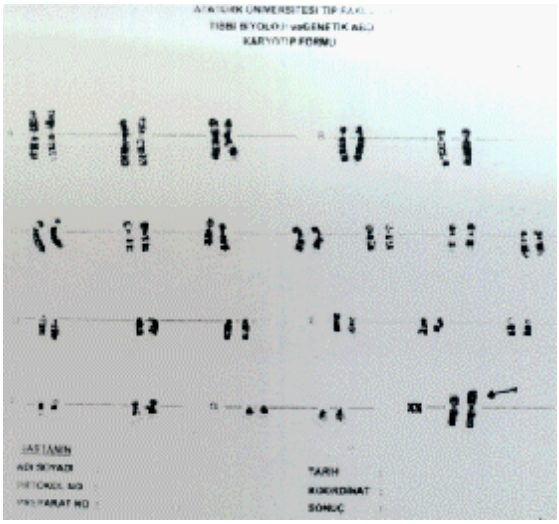

Figure 2. Karyogram of the case (arrow shows isochromosome Xq).

that hypothyroidism and slightly mental retardation risks were higher in these patients than in normal population (4,5). In addition, an association between inflammatory bowel disease especially ulcerative colitis and i(Xq) genotype was demonstrated (6). Correlation between acute monocytic leukemia and i(Xq) has also been reported (7). However, contrary to the classical Turner's syndrome where the risk of congenital heart disease is high, it was reported to be normal in i(Xq) (8).

In present case, none of the mentioned findings were observed except growth retardation and primary amenorrhea. Additively, in this case, mental status was normal. Considering this case, the symptoms in i(Xq) can be assessed to be more slightly than in classical Turner's syndrome.

The origin of isochromosome has been investigated in reported cases (9-12). Some of them have pointed out a significant relationship between the existence of i(Xq) and the paternal age (9,10). Using DNA markers it has been demonstrated that the origin of i(Xq) was paternal in 3 cases out of 5 and maternal in the other two (11). Considering the advanced age of her mother (41 years at giving birth), we suppose that the origin of i(Xq) in this case may due to a maternal centromer error in meiosis II.

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