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45, XO karyotype in women with atypical Turner's syndrome presentation

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Abstract
Turner's syndrome (TS) is one of the important chromosomal disorders with loss of one sex chromosome in females. The characteristic features include short stature, webbed neck and poorly developed secondary sexual characters. Here we report four cases of TS (one asymptomatic and three symptomatic) who were admitted at Coimbatore Medical College Hospital with general health complaints. Further diagnosis was followed and discussed. Karyotyping was performed, which confirmed the presence of TS with presentation of 45, XO karyotype.

Keywords: Turner's syndrome; short stature; secondary sexual character.

Introduction

Classical Turner's syndrome (TS) is characterized by sexual infantilism, webbed neck, short stature, peripheral edema, lymph edema, renal and cardiovascular anomalies, gonadal dysplasia and learning disability (Zinn *et al*., 1993; Gicquel *et al*., 1998; Sybert, 1998; Ross *et al*., 2000). It effects at least 1:2000 live born girls and is therefore one of the most common sex chromosome abnormalities in humans (Nielsen and Wohlert, 1991). More than half of the TS patients have mosaic complement (Simpson, 1975; Saenger, 1993). The frequency of physical abnormalities in TS varies with the pattern of karyotype (Ranke, 1989). Women with TS show specific cognitive deficits in visuo-spatial processing as well as in selective aspect of attention, executive processing and memory functions. Specific neurophysiologic deficit that may affect adaptation includes four interacting areas of functioning: visual–spatial deficit (e.g., difficulty in driving), social–cognitive defect (e.g., failure to appreciate subtle social clues), problem with nonverbal solving (e.g., mathematics) and psychomotor deficit (e.g., clumsiness) (Rovet, 2004). One half of clinically identified cases possess part of a second X chromosome too, which is structurally abnormal, usually in association with some 45, XO cells (Jacobs *et al*., 1997).

Case 1

A 45-year-old woman was admitted in Coimbatore Medical College (CMC) Hospital with complaints of cough with sputum production and fever of 1 week duration. Fever was intermittent with no other symptoms such as rashes and joint pain. Bowel and bladder habits were normal. On physical examination she was found to be of short stature, with height of 140 cm and weight of 40 kg. She was febrile, and pallor was present. In this case, the woman had no webbed and short neck that forms a characteristic of TS. Breasts were underdeveloped and there was lack of pubic and axillary hair growth (Figure 1). Infantile external genitalia were present and no skeletal abnormalities were observed. Her IQ was normal with normal mental status. All physical and clinical investigations were carried out and the results were found to be normal. On ultrasound analysis, abdomen showed infantile uterus. Echocardiography was found to be normal. Skeletal survey was done and it showed mild scoliosis. The resting position was also found to be normal. Karyotyping study was carried out at Human Genetics Laboratory, Bharathiar University, Coimbatore and every metaphase plate was found to represent the 45, XO karyotype (Figure 2).

Case 2

A 27-year-old woman with severe abdominal pain for 2 weeks was admitted in CMC Hospital,

Coimbatore. Features observed included short stature, absence of secondary sexual characters, lack of menstruation cycle (primary amenorrhea) within puberty stages, severe abdominal pain and underdeveloped ovaries. Finally, karyotyping revealed the presence of 45, XO (TS) aberration (Figure 2).

Case 3

A 17 year female born to non-consanguineous parent had short stature, primary amenorrhea, absence of secondary sexual characters, developed pulmonary tuberculosis and took anti-tubercular treatment for 6 months. She then developed backache and spinal abnormality. She presented with anemia. Patient had exaggerated lordosis along with Gibbus deformity in lumbar region. Chest X ray postero-anterior (PA) view was normal; X ray thoraco-lumbar spine showed wedge compression and collapse of L3, L4, and L5 vertebrae. Sputum was acid-fast bacilli (AFB) negative, ultrasonography (USG) of abdomen showed streak ovaries. Karyotyping report showed 45, XO (TS) karyotype (Figure 2).

Case 4

A 26-year-old woman with complaint of abdominal pain for past 2 weeks was admitted in the above-mentioned hospital. Further investigations revealed absence of secondary sexual characters, primary amennorhea, widely spaced nipples, hypoplastic uterus, underdeveloped ovary, cardiac examination including the echocardiography revealed atrial septal defect of ostium secundum type, but no webbed neck. Karyotyping showed mosaic type of TS 45, XO/46, XX in metaphase plates (Figure 2).

Figure 1: Typical presentation of underdeveloped breasts with wide spaced nipples in a patient diagnosed with TS.

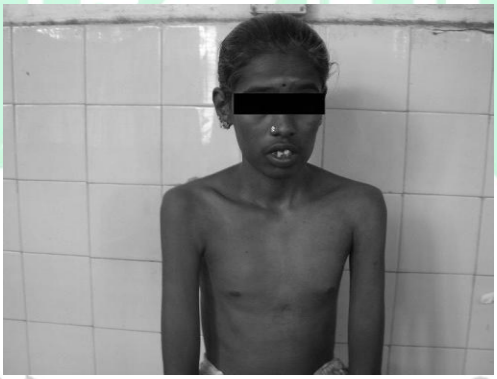
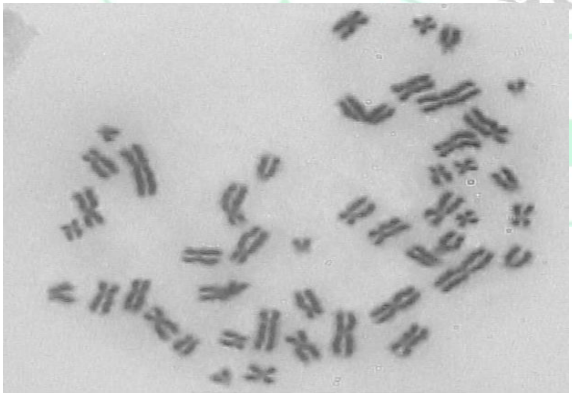
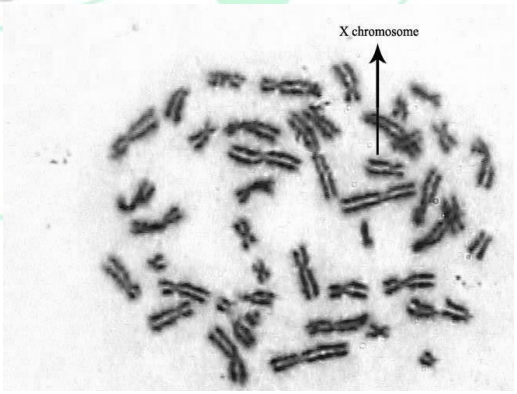


Figure 2: 45, XO/46, XX (mosaic) one of the cases confirmed by karyotype.



46, XX metaphase plate



45, XO metaphase plate

Discussion

Turner's syndrome is the result of complete or partial X chromosomal monosomy in phenotypic females and is associated with characteristic clinical features. The most consistent characteristics found in the cases studied were short stature and gonadal dysgenesis. The cases presented here lacked some of the other typical phenotypic characteristics like broad shield-like chest, webbed neck, high-arched palate, and puffiness of the dorsum of the fingers found in TS (Williams, 1992; Pasquino *et al*., 2000). In such presentations, the diagnosis is subsequently confirmed by chromosome study, with a karyotype of 45, XO. Turner's syndrome should be suspected when there is a combination of short stature, infantile genitalia, and infantile uterus. Although there is no pathognomonic clinical feature of this syndrome, any woman with short stature along with underdeveloped genitalia and uterus should be considered for diagnosis of TS with assistance of chromosomal analysis. The psychosocial impact of TS may be substantial in young girls and women. These effects may be caused by infertility, short stature and impaired development of sexual characteristics and most importantly, by lack of libido (Sutton *et al*., 2005). Physicians should elicit specific concerns from patients, addressing them individually and should recommend comprehensive school-based psycho-educational assessment (Noor *et al*., 2007).

Conclusion

The present study again reemphasizes the need for chromosomal analysis in a syndrome like Turner's, where more often the classical phenotypic manifestations are not found resulting in diagnostic dilemma.

Acknowledgement

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Ethical Approval

The patients' consent was taken by the authors before conducting this study.

Conflict of Interests

No funds were received for this study. Authors do not have any conflicting interests.

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