

A CASE OF TURNER'S SYNDROME VARIANT WITH 45, X/46, X, FRAGMENT KARYOTYPE

(A Case Report)

by

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Introduction

Turner's syndrome is the most common chromosomal anomaly in the human being. This is also a common finding in 40% of those patients with cytogenetic abnormalities who present with primary amenorrhea. (Van Niekerck, 1978). In a systematic screening for chromosomal abnormalities in primary amenorrhea, we have found a rare variant of Turner's syndrome with 45, X/46, X, fragment karyotype. The case details are described and discussed.

Case Report

K.S., a 17 year old female came for primary amenorrhea and mental subnormality. She was the first born of normal and healthy parents. Family history revealed parental and paternal grand parental consanguinity (2nd and 1st cousins respectively). Age of the mother at the time of birth of this girl was 18 years and that of father 19 years. There were no issue after the birth of this girl except an abortion at 5 months of pregnancy. Prenatal, natal and post-natal histories of the patient were normal. There was slight delay in the development of milestones.

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Clinical Examination

Clear cut picture of Turner's syndrome was present with increased carrying angle and high arched palate (Fig. 1). Single palmar crease was present in the right hand. Secondary sex characters like breast development, pubic and axillary hair were completely absent. External genitalia were of normal female type. Ultrasonographic scanning of the lower abdomen showed absence of uterus.

Hormonal Findings

Serum gonadotropins were normal (FSH-20 mIU/ml., LH-15.0 mIU/ml). Prolactin-10.0 ng/ml. TSH-4.6 mIU/ml. All are falling under normal range.

Cytogenetic Findings

No peripheral chromatin was seen in the nuclei of buccal epithelial cells. Cultures of the peripheral lymphocytes revealed 45 chromosomes and an additional small piece of chromatin observed in 90% of the cells. The length of this piece was one fourth that of a G chromosome. The fragment was too short to demonstrate any informative banding pattern. The patient's mother had normal karyotype. Father was not available for study. Karyotype of the patient was 45, X/46, X, fragment.

Discussion

Jagiello *et al* (1966) and Zanoio *et al*

(1978) reported similar cytogenetic profiles in patients with Turner's syndrome. Clinically, their cases showed enlarged clitoris and histological examination of the gonads revealed the presence of Leydig cells and Sertoli cells without spermatogenesis. On the basis of their clinical and histological findings, these authors interpreted the fragment to be a portion of 'Y' chromosome. Sieber's *et al* (1973) also reported a similar finding in a patient with Turner's stigmata and streak gonads whose karyotype was interpreted as 45, X/46, X, i(Yp).

The present case differs from that of others in that clinically she was of normal female phenotype and also without clitoromegaly. On the basis of these clinical findings, absence of Y fluorescent body in the buccal epithelial cells and late replication studies of X chromosome, the fragment in the present study was interpreted as a part of the X chromosome. A critical role of X chromosome in primary determination of sex and fertility has recently been revealed. Any interference with the integrity of the regions Xp11—Xp21 (Therman *et al* 1976) and Xq13—Xq27

(Summit *et al*, 1978) in the X chromosome leads to Turner stigmata and gonadal dysgenesis. In our patient, the entire portion of the X chromosome was deleted, except for a small fragment, resulting in a complete Turnerian Phenotype.

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See Figs. on Art Paper VII

SYMPOSIUM

- (1) Changing trends in Fertility Control
- (2) Breast feeding and Benign Lesion of Breast
- (3) Monitoring of labour: Panel discussion

Indian College of Obstetricians and Gynaecologists with the committee for the study of Female Breast of the Federation of Obstetric and Gynaecological Societies of India and Bengal Obstetric and Gynaecological Society cordially invite you to a Symposium on "Changing trends in Fertility Control" on 29th March 1986 at Eden Hospital, Calcutta and Symposium on "Breast feeding and Benign Lesion of Female Breast and a panel discussion on "monitoring of Labour" on 30th March 1986 at Post Graduate Institute of Obstetrics and Gynaecology, Calcutta.

Detail Programme will follow.

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