TURNER'S SYNDROME

(A Case Report with Review of Literature)

by

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and

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Turner described in 1938 a syndrome in post-pubertal females consisting of sexual infantilism, short stature, webbed neck and cubitus vulgus. It is gonadal dysgenesis complement of only one X and is written as 45, X. It appears to be one of the rare chromosomal anomalies. Maclean *et al* (1964) reported 0.4 per thousand female births. Carr (1965) reported that upto 5.5% of aborted foetuses have had 45, X-O karyotype.

The diagnosis of Turner's syndrome is usually evident after puberty owing to primary amenorrhoea and sexual infantalism. A chromatin negative buccal smear is confirmative although positive smear does not exclude the possibility. Since mosaicism or structural abnormality of X chromosome in the form of iso-chromosome may be present (as studied by various workers, Delachaplec, 1962; Mikkelsen *et al*, 1963; Jones *et al*, 1963; Lindstein, 1963; Ferguson Smith, 1964, 196; Srinivasa Rao *et al*, 1973).

Albright *et al* (1942) found elevated urinary excretion of gonadotrophins in these patients. Wilkins *et al* (1954) showed that affected individuals are phenotypic females with streak gonads that is all the fibrous tissue lacking follicles and ova.

We are presenting a case of Turner's syndrome with buccal smear showing positive sex chromatin. We do not have facility for chromosomal study so possibility of mosaicism is not studied.

CASE REPORT

Smt. A.B., aged 22 years, came to hospital for not having attained menarche, for neck deformity and for partially developed sex characters. She was also having hysterical fits off and on for the last 5 years.

Family History: Cosanguinity of her parents present. She has 2 sisters and 2 brothers. She is the eldest one and is married. Her both younger sisters and mother are of normal height and growth and are having normal menstruation.

She was of short stature, 127.5 cm in height, short webbed neck (Fig. 1) her carrying angle was wide. She was having 50 cm chest circumference, chest was flat with wide apart nipples, axillary and pubic hair were absent. External genitalia were infantile labia majora and minora were small and thin hymen was ruptured (Fig. 2). Vaginal introitus could admit 2 fingers with much difficulty vagina was of 5 cm in length with small cervix and small uterus which was acutely anteverted and anteflexed.

Her blood pressure was 110/70 mm of Hg.

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Cardiovascular and respiratory systems were normal. She was educated upto primary class, and was having I-Q below normal. Skiagram chest revealed old healed tubercular lesion. Her basal metabolic rate was —37%. Erythrocytic sedimentation rate 20 mm of Hg. Hemoglobin 9 gm/100 ml. Her urine examination was normal. Vaginal cytology study showed low intermediate cells. In buccal smear Barr bodies were positive. On gynaecography uterus was infantile. Dilatation and curettage was done, endometrial scrapping were scanty. Histopathological study of endometrial scrapping was few glands with proliferative phase.

Discussion

Wilkins *et al* (1954), Polani et al (1954), Decourt *et al* (1954) first reported that most individuals with Turner's syndrome are sex chromatin negative. A buccal smear can reveal quickly whether or not 2 chromosomes are present. If the patient has a second chromosome in the form of an Xqi (Isochromosome for long arms of an X) or an Xp—(Deletion of short arms of an X) however the buccal smear will be sex chromatin positive. These patients may have normal height and spontaneous menstruation. Mikkelsen *et al* (1963), Jones *et al* (1963), Lindstein (1963), Ferguson Smith (1964 and 1965) found streak ovaries with connective tissue with occasional atretic follicles.

Once an individual has been diagnosed as having gonadal dysgenesis, councelling and treatment with hormones are very important to the patient and her parents. Voorhess (1969) has presented a discussion on the psychologic councelling and hormonal therapy. In this case also as histopathologic study of endometrium scrapings obtained with endometrial biopsy, showed few glands with proliferative phase so hormonal therapy was started. Mikkelson *et al* (1963) studied the ovaries in one pair of XO/XX mosaic twins of years old and found that the child contained large number of ova. Although 45, X females are considered sterile there are 2 cases studied in the literature where the individuals have undergone parturition (Banner *et al*, 1960 and Nakashima *et al*, 1971). Courtbrown *et al* (1964) described a woman with an XO Karyotype who menstruated for 23 years. One case came after 1 month for follow-up and she had hormonal withdrawal bleeding. This gave her much satisfaction and confidence and she did not have fits after the start of treatment.

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See Figs. on Art Paper XII