TESTICULAR FEMINISATION

(Report of 3 Cases)

by

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The characteristic clinical picture of this syndrome is that of a female of normal height with well developed breasts but scanty or absent axillary and pubic hair. The external genitalia is completely feminine but the vagina is short and blind with absence of cervix, uterus and tubes. The gonads (testes) are found in the inguinal region in majority of the cases. These 'women with testes' report to the doctor with primary amenorrhoea, sterility or swelling in the inguinal region. (Morris, 1953; Morris and Mahesh, 1963; Scott, 1971 and Dewhurst, 1972).

Three cases of Testicular Feminisation including two sisters are being presented for its rarity.

CASE REPORTS

Case 1

Mrs. A, 20 years, was admitted to our department on 25-12-1974 with the complaints of primary amenorrhoea, sterility and swelling in the right inguinal region. She was married at the age of 14 and was leading a completely satisfactory married life.

At the age of 13 she noticed normal breast development and she had grown up gradually. Since the age of 14 she noticed a swelling in the right inguinal region which was increasing in size gradually. She had consulted several doctors without relief.

Family History: She is the third out of six children. Her parents have 4 girls and 2 boys. Her elder sister (second in order) is also having primary amenorrhoea. The first elder sister is married with children. Her younger sister has also attained menarche at the age of 14.

On examination she had a feminine body configuration. Height 5 ft. with average build. Face was smooth and there was no facial hair. Pulse rate, blood pressure and respiratory rate were normal. Her voice was feminine with good scalp hair and well developed breasts (Fig. 1) but areola and nipples were poorly pigmented and small. Axillary and pubic hair were sparse.

Cardiovascular and respiratory systems were normal. Abdominal examination revealed a smooth firm rounded swelling, about 3″ diameter at the right external inguinal ring (Fig. 2). This mass could be reduced into the abdominal cavity through the internal inguinal ring and reappeared on coughing. This mass was diagnosed as the gonadal swelling in the hernial sac.

External genitalia were normal. Clitoris was normal, vagina was 2″ deep with normal rugosity and ended in a blind pouch (Fig. 3). There was no uterus, cervix or any palpable adnexal masses.

Investigations: Buccal smear—Negative for Barr bodies. Peripheral smear—No drum stick pattern. Vaginal Cytology—Predominant intermediate cells 0/95/5. I.V.P.—Both kidneys were normally situated and functioning normally. Hormone assay—17 Kelosteroids—37 mg for 24 hours.

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On exploratory laparotomy uterus was absent. On the left side testis was visualised 2½" x 1" in the pelvis. There was no tube. Hydatid of Morgagani was present. On the right side testis was 3" x 1" and it had herniated through the inguinal ring. The testis on the right side was pulled back into the abdominal cavity and bilateral orchidectomy was done (Fig. 4) Herniorrhaphy was done on the right side from within and abdomen was closed in layers. Postoperative period was uneventful, wound had healed well and patient was advised to take Oestrogens.

Histopathological Report: Bilateral testicular tissue showing absence of germ cells.

Case 2

This patient is the elder sister of Case 1. She was 24 years old. She had primary amenorrhoea and was married for 8 years. She was leading a normal married life. Her breast development started at the age of 13 and was normal.

On examination, she was an average built female. Height was 5' 1". Breasts were normal with poorly pigmented areola (Fig. 5). External genitalia were normal and vagina was 2½" deep with normal rugosity. No cervix, uterus or adnexal masses were palpable on bimanual pelvic examination.

Investigations: Sex chromatin—negative. Vaginal smear—predominance of intermediate cells.

Case 3

C, 19 years, was admitted to the hospital on 25-5-1976 for primary amenorrhoea. She was the eldest of the 5 daughters of her parents. Her younger sister who is 16 years is menstruating regularly for the last one year and is taller than her.

On examination, her somatic growth was retarded. She was 4' 7" and appeared like a 12 years old girl. Her appearance was feminine with long scalp hair and smooth face. Breasts were poorly developed and appeared as small nodules with hypoplastic nipples (Fig. 6).

Local examination showed a moderately developed labia majora with normal looking clitoris. Vagina was hypoplastic with normal situation of the external urethral meatus. A small catheter could be passed upto 1" into the vagina where it ended blindly. There was no cervix or uterus. There was a swelling on the left labia majora of 2.5 cm x 2 cm which could be easily reduced into the abdominal cavity through the inguinal canal. A provisional diagnosis of testicular feminisation with herniation of the left gonad through the inguinal canal was made.

Investigations: Buccal smear—negative for sex chromatin. Peripheral blood smear—no drum stick pattern seen.

On exploratory laparotomy there was no uterus. There were bilateral testes with the left one herniating into the inguinal canal. Both gonads were removed and herniorrhaphy was done on left side. Both ureters were normally situated. The gonads on gross examination and cut section were looking like normal testis (Fig. 7). Histopathology showed hypoplastic seminiferous tubules with absence of spermatogenesis (Fig. 8).

Discussion

From the psychosexual aspect these patients are definitely females. The first 2 cases who were married were having a satisfactory and active sexual life. The sex urge of these patients was usually the same as those of other women. (Morris, 1953; Morris and Mahesh, 1963; Scott, 1971; Dewhurst, 1972; Vohra *et al*, 1970; Raju *et al*, 1970).

The first 2 cases being sisters points to the family incidence of this syndrome. The tendency for familial incidence transmitted through the maternal side is also reported by Morris (1953), Morris and Mahesh (1963), Zourlas and Howard (1965), Scott (1971), Dewhurst (1972), Raju *et al* (1970). The raised 17-Ketosteroid level in Case 1 shows that there is increased amount of circulating androgens. This is proved by Morris and Mahesh (1963).

Gonadectomy as part of management

Morris and Mahesh (1963) reported 1 case of malignant tumour arising from the gonads in a teen age patient and 2 malignant tumours in patients in twenties. In 50 reported cases of 30 years of age

or older there had been 11 malignant tumours, chiefly germinomas. In addition there were 15 tubular adenomas and 10 cysts arising from the gonads. This 22% incidence of malignancy appears sufficient reason to advocate removal of gonads. Overzier (1963) as quoted by Dewhurst (1967) reported 10 malignant cases among 128 cases. If the patient is less feminine it is better to remove the gonads as masculinisation is a real risk later on (Dewhurst, 1967). Zourlas and Howard (1965) have carried out gonadectomy in all their 6 cases including in a child of 4 years. Gonadectomy was part of the management advocated by Raju *et al* (1970), *Vohra et al* (1970), Nalini *et al* (1972) and Sreenivasa Rao (1974). Since the clinical picture was very typical of testicular feminisation in the first case and the secondary sexual characters were well developed to avoid the real danger of malignancy gonadectomy was performed. During the operation bisection of the gonad was very suggestive of being testis, frozen section was not done. In the third case which was more of an incomplete variety of testicular feminisation a gonadectomy was done to prevent the possibility of virilisation and malignant change later on.

Summary

(1) Three cases of testicular feminisation including two sisters are described.

(2) Clinical features, hormonal studies, pathogenesis, genetics and management are briefly discussed.

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See Figs. on Art Paper VI-VII