"TESTICULAR FEMINIZATION" SYNDROME

(A Case Report)

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Among the most interesting forms of intersexuality is that of phenotypic female with testes. Morris (1953) coined the term "Testicular Feminization" for chromosomal and gonadal males, presenting with complete female urogenital sinus development, lack of development of both Mullerian and Wolffian duct systems, female secondary sexual characteristics, and scanty or absent sexual hair. More cases of "testicular feminization" syndrome are diagnosed due to the greater awareness of the condition and its genetic implications and the better facilities available for hormone assays and sex-chromatin studies. This syndrome has been reviewed extensively. (Mahesh, 1969; Rao, 1974). Morris and Mahesh (1963) called attention to an incomplete form of testicular feminization syndrome with clitoromegaly and incomplete female secondary sexual characteristics.

The purpose of this report is to present a typical case of "Testicular Feminization" with absent wollfian and mllerian development, female secondary sexual characteristics and inguinal testicles.

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CASE REPORT

An 18 year-old Hindu female was admitted to the Medical College Hospital, Kottayam on January 14, 1976 for primary amenorrhoea and bilateral swellings in the inguinal region. She was the only 'female' in the family.

Pertinent physical findings revealed a pleasant, healthy, well-developed, tall thin woman of very feminine appearance. Her height was 5 feet and 5 inches. Her breasts were well developed (Fig. 1). Axillary hair was absent; hair on the mons and labia majora was scant. There were bilateral oval swellings in the inguinal region. The oval masses could be easily brought down to the labia majora. No obvious inguinal hernia was present. The labia majora, minora and clitoris were moderately developed (Fig. 2). The introitus was admitting one finger and the vagina was shortened to one cm. The cervix and uterus were absent and no adnexal masses were palpable.

Routine laboratory studies were normal. A buccal smear for sex chromatin study was negative. 17-ketosteroid values for 24 hours urine were, pre-operative level 9 mgms, post-operative (8 days after operation) level was 3 mgms.

On January 23, 1976 the patient underwent bilateral gonadectomy and laparotomy. Exploration of the inguinal region revealed bilateral testes—like gonads with rudimentary epididymis and vas deferens. The absence of uterus and tubes were confirmed by laparotomy.

The histologic diagnosis was: Bilateral immature testes with seminiferous tubules showing hypoplasia of the germinal line and absent spermatogenesis. There was mild Leydig cell hyperplasia; and the Leydig cells were forming smali clusters situated in the 'triangles' between

the tubules. The patient's postoperative course was uncomplicated and she was discharged on the 10th postoperative day. The gonadal histology is shown in Fig. 3.

Discusion

Jost has postulated (1958) and it has been further shown that two inductor substances produced by the foetal testes namely mullerian duct inhibitor and wolffian duct stimulator act locally in determining gonaduct differentiation. Mullarian inhibitor factor is neither androgenic nor steriodal, whereas the stimulator of the wolffian ducts and external genitals is a steriodal androgen. Patients with testicular feminization syndrome produce enough androgen from the testes; (Jeffcoate *et al* 1968; Morris and Mahesh, 1963) but show underlying 'end organ' resistance to the 'biologically active androgens'. Failure of the androgenic male organizer, due to end organ resistance, results in deficient wolffian duct differentiation in an individual in whom mullerian duct regression, produced by the second male organizer, does occur (French *et al* 1970). The end organ insensitivity to androgen is responsible for the female urogenital and external genital developments. In our case, the 17-ketosteroid value had fallen from 9 mgms to 3 mgms after gonadectomy, indicating that the major source of androgen was the testes.

Bruchovsky and Wilson (1968) demonstrated that the biologically active androgen in normal males was not testesterone itself but 5 alpha-dihydrotestosterone (DHT) which is formed testosterone by the action of Delta-4-5 alpha-reductase. Initial observations that the skin of patients with testicular feminization syndrome was unable to convert testosterone to DHT were later found to be not due to a deficiency of the specific reductase but, rather, to increased plasma levels of testosterone-binding protein which decreased the amount of unbound testosterone available to the skin for 5 alpha reduction.

The characteristic female appearance of the patient with testicular feminization, accompanied by abundant breast tissue in the presence of male gonads is very familiar and is well demonstrated in our case. The exact source and nature of the oestrogens produced by these individuals, however, has not been studied or defined as comprehensively as for the androgens.

The testes in this patient were removed because of the neoplastic potential, the magnitude of which increases with age (Mahesh 1969). Since the oestrogen production by the adrenals are under the influence of the gonads, gonadectomy is preferably postponed until the breast development is completed; otherwise oestrogen-progestogen therapy should be instituted as the individual approaches puberty.

Summary

A case of 'testicular feminization' syndrome (complete variety) is presented with review of literature.

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See Figs. on Art Paper VII