TESTICULAR FEMINISATION WITH CLITOROMEGALY

by

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It was Morris and Mahesh (1963) who proposed the syndrome of Testicular feminisation be divided into two groups on the basis of clitoral development. In their opinion the person who is affected with male hermaphroditism with normal female external genitalia is to be distinguished from the person who has clitoromegaly. They designated the former as complete testicular feminisation which is caused by androgen insensitivity of the target organs and the latter as incomplete testicular feminisation, the cause of which is obscure.

But Teter *et al,* (1966) reiterated that the term incomplete testicular feminisation be applied to those who manifest enuchoid body proportions, no breast development and 'shield' chest and who should be excluded from the syndrome of complete testicular feminisation but there are other patients who in spite of clitoral enlargement, possess almost complete female secondary sex features. Thus, Teter *et al* categorised feminising testes Syndrome cases into those hairless women with testes from feminising testes syndrome with clitoromegaly and pubic and Axillary hair with complete female secondary sex features. Gonads may be palpable in the inguinal hernia. At puberty, affected patients usually feminise but may have varying degrees of masculinisation. At any rate sexual hair does appear at puberty and the voice may deepen. Its inheritance appears to be identical to the complete form. The etiology of this condition also appears to be end-organ insensitivity to androgen, but the insensitivity is only partial according to Summitt (1972).

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Case Report

Smt. A., aged 22 years, unmarried, was admitted in King George Hospital, Visakhapattam on 21-4-1968 at 7-15 p m. as an emergency case with signs and symptoms of obstructed inguinal hernia on the right side. The swelling was irreducible, painful and tender, associated with vomiting. A clinical diagnosis of strangulated right inguino-labial hernia was made and emergency operation was done the same night.

History: An unmarried woman of 22 years, bilateral inguino-labial swellings present since childhood. She is the only child in the family.

The patient was tall of 5′ 4″ with enuchoid features. No hirsutism, voice feminine, no acne, well developed breasts and no periods at any time since puberty. Pubic and Auxiliary hair present (Fig. 1).

Abdominal Examination: Slight distension of lower abdomen. An obstructed irreducible tended right inguinal hernia. In the left inguinal region also there was a soft freely movable, reducible hernia, size of mass of 1½″ × 1″, which disappears into the external abdominal ring on pressure and reappearing on coughig. Testicular sensation present suggesting the mass to be testes.

Bimanual Examination: Hypertrophy of clitoris, labia major and minor normal and well formed (Fig. 2). Urethral opening in normal situation, vagina admits two fingers freely upto 1½″ depth, cervix not made out.

Speculum Examination. A blind vagina with normal rugosity.

Rectal Examination: Uterus and gonads not made out.

Investigation: Hb%: 9 gms%. Buccal mucosal cells for sex chromatin study—negative for Bar bodies suggesting genetic sex to be male. Peripheral blood culture for karyotype analysis showed 46/XY male karyotype.

Vaginal Cytology: 2/70/28 oestrogenic smear.

Gross Pathology: The removed gonads were testes measuring 4 cm × 3 cm × 1". The right gonad was haemorrhagic due to strangulation. The investigating capsule was white tunica albuginea. Each testis was connected by a tortuous spermatic cord with recognizable epididymis, was deferen and dilated plexus of vessels (Fig. 3).

Histopathology: Thickened tunica albuginea with thickening of the basement membrane and sclero-hyalinosis of many tubules. Ocassional tubule shows lipid laden Sertoli cells. No spermatogenesis seen. Leydig cells show marked hypertrophy and hyperplasia in areas showing nodular formations (Fig. 4).

Follow-up: She married 3 months later and is leading a family life with no complications. Her general and local conditions are absolutely normal. Immediately after discharge from the hospital she was maintained on stilboestrol but later discontinued.

Discussion

These cases are reported to have a strong familial tendency and this syndrome is hereditary with maternal transmission, the carriers usually being normal females. The oestrogenic effect of this syndrome is thought to arise from hyperplastic Sertoli cells of the tubules. Castration results in a menopause-like state with hot flushes, elevation of gonadotrophins and a decrease in oestrogen secretion as noted by vaginal cytology and atrophy of breasts. Morris and Mahesh (1963) commented that there is strong evidence of this syndrome to be an endocrine abnormality, essentially related to end-organ insensitivity to androgen effect.

Teter *et al* (1966) differentiated the testes of the two groups as follows:

Hairless Women with Testes

1. No thickening or sclero-hyalinisation of tubules.
2. Immature Leydig cells closely encircle the tubules in the manner of cuff.
3. Small tubules lined with undifferentiated Sertoli cells.

Women with Clitoromegaly with Testes and Presence of Sexual Hair

(1) Sclero-hyalinosis of tubules.
(2) Leydig cells more mature and are grouped in triangular islands between the tubules.
(3) Large tubules lined with lipid laden Sertoli cells.

Patients with feminising testes syndrome can be described as showing total underlying 'end-organ' resistance to biologically active androgen, though they are able to synthesize and maintain normal male levels of testosterone. However, Bruchorsky and Wilson (1968) demonstrated that the biologically active androgen in normal males was not testosterone itself but 5L-Dihydro-testosterone which is formed from testosterone by the action of 5L-reductase. But it is lately known that it is not due to 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 5L-reduction. If that is correct then this clitoromegaly and development of pubic and axillary hair in these cases may be due to increased amounts of unbound testosterone available to the skin for 5L-reduction Mauvais *et al*, (1970).

This explanation fits well with the clinical features that are noted in these cases with the histopathological features of testes studied.

The facilities for estimation for unbound testosterone levels in plasma were not available for confirmation and that may throw more light in understanding these cases.

Summary

1. A rare case of Testicular feminisation syndrome with clitoromegaly and presence of sexual hair, with complete feminisation features is reported.

2. Male Karyotype of 46/XY and testes showed sclerohyalinosis of tubules with large islands of Leydig cells were noted.

3. These types of cases should be distinguished from the Incomplete Testicular feminisation cases of Morris and Mahesh (1963).

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See Figs. on Art Paper VIII