A CASE OF TESTICULAR FEMINIZATION SYNDROME

(A Case Report)

by

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Testicular Feminization Syndrome or feminizing testes is an uncommon but very interesting syndrome encountered occasionally in gynaecological practice. The first attention to this syndrome was drawn by Morris who introduced the term Testicular Feminization in 1953 and described the syndrome. The chief presenting feature in this syndrome is that a person of female phenotype attends the gynaecological service for primary amenorrhoea and on further investigations it is found that the sex gonad is testis. Externally there cannot be any suspicion that the person is a male pseudo-hermaphrodite. Morris described the syndrome in a case where the patient had female phenotype with well developed breasts and sparse axillary and pubic hairs. The external genitalia are like that of an adult or adolescent girl. The vagina is capacious but ends in a blind pouch. The uterus is absent. There may be an inguinal hernia with a gonad inside the inguinal ring or both the gonads may be intra-abdominal. Gonadal biopsy proves that the gonads are testes. This syndrome may occur in other sibblings or closely related persons. The psychologic orientation is like that of a female. There has been considerable discussion whether the person with the enlargement of the clitoris should be included in this syndrome. Morris considers that in the true or complete syndrome there should not be any manifestation of aberration of external genitalia. A typical case is presented here.

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Case Report

Mrs. S. F. aged 19 years, muslim, PO + O, married in 1967 was admitted in Eden Hospital on 16-8-69 with the complaints of primary amenorrhoea and consequent mental unhappiness of both husband and wife.

History of Present Illness:

The patient gave the history that she is now 19 years old. At the age of 13 she noticed normal breast development but there was no menstruation. She has grown up gradually and consulted many physicians for her amenorrhoea and some of them instituted hormonal therapy with no menstrual response. But, even after her marriage when she had no menstruation she attended the Eden Hospital.

Past History: Past history is non-contributory except fever in her childhood.

Personal History: She is a housewife of a middle class family. Her intelligence and mental attitudes are normal. She has also read upto class VI.

Family History: Her parents are first cousins. The patient has three sisters and one brother and out of the total five siblings she is the first in order. Her next sister, aged about 16 years is having normal

menstruation, menarche being at the age of 12 years. The others are still in their childhood and no sexual abnormality is detected in them.

Examination and General Survey:—

Face and body configuration—feminine. Height—5'-1". Weight—52 Kg. Span—5'-1½". Build—average. Nutrition—fair. Pulse—72/m. Resp.—20/m. B.P.—110/60 mm. Hg. Temp.—98°F. Her psycho somatic pattern is like that of an adult female.

Examinations of Sex Characters:—

Scalp hair—Growth of hair is good and the distribution is dense. Voice—feminine. Breasts—well-developed. Axillary hair—Scanty. Pubic hair—sparse.

P/V. & E.U.A.

External genitalia-normal, including normal clitoris.

Vagina—is capacious and is of normal length but ends in a blind pouch.

Neither the uterus, nor the adnexae were palpable by P/V or P/R examinations.

Heart & Lungs—N.A.D. Liver and spleen—not palpable. Per abdomen—N.A.D.

General Investigations:

Blood—Hb 10.1 G%; WBC—6,000/cu. mm.; poly 72%; lympho 25%; mono 0%; eosin 3%. Platelets—adequate. Urine—N.A.D.

Skiagram:

(a) Chest—Lung fields clear, cardiac shadow normal, no rib notching.
(b) Skull—N.A.D., suture lines normal.
(c) Long bones—skeletal age corresponds to the actual age.

Special Investigations:—

Buccal smear—As very few Barr bodies were detected, the slide was read as negative.

Leucocyte—Drum sticks negative in 100 polymorphs.

Vaginal Cytology—Her present maturation index is 0/97/3 with slight atypic and cyanophilic pattern.

Gynaecography—No uterine shadow, Left gonad appeared normal, right gonad enlarged.

I.V.P.—Both kidneys are normally situated and functioning. There is no radio-opaque shadow in any part of the urinary tract.

Urinary hormone assay report

Date	Volume of 24 hrs. urine	PH of the urine	Urinary total oestrogens assay by Ittrich (1960) method	Urinary 17 Ketosteroids assay by Vesten Gaard (1951) method
2-7-69	1500 ml.	PH 5.4	Total oestrogens 6/ug/24 hrs.	Total 17 Ketosteroids 1.7 mg/24 hrs.

Result—Excretion values for total oestrogens and total 17 Ketosteroids fall within the lower part of postmenopausal range.

Sex factors in the patient:

Sex Factor	*Findings.*
Genetic	
Nuclear chromatin ♂ (Negative) by tissue culture.	Genotype male Phenotype female
Chromosomal pattern	not done
Gonad	Testis (♂)
External Genitals	Feminine (♀
Internal genitals	Absent.
Sex hormone status	Hypogonadal
Sex of rearing	female (♀)
Sex role	female (♀)
Gender role	female (♀)

Laparotomy:

An exploratory laparotomy was performed on 3rd September, 1969. The uterus and oviducts were found absent. Both the gonads were resembling testis. They were attached to the lateral pelvic wall by the round and infundibulopelvic ligaments. So bilateral gonadectomy was performed. Exploration of the other organs were done. No abnormality was detected in adrenals.

Gross appearance of the specimen and the histopathological report:

A pair of oval tissue, size 2″ x 1½″ ¾″. On cut surface, irregular whitish more or less circular areas were found which were firmer than the surroundings—Section from whitish area and from the intervening darkened area shows structure of testicular tissue. The tubules shows no evidence of spermatogenesis.

Post Operative:

The post-operative course was uncomplicated. She was discharged from the hospital on the 16th Sept. 1969 with the advice that she should have to maintain oestrogen therapy daily.

Conclusion:

The reported incidence of Testicular Feminization is infrequent. Certainly more cases occur than those reported in the literature.

These patients cannot be properly classed as "Pseudo-hermaphrodite" as by the appearance they look exactly like a female. There is no suspicion as regards their sex from inspection of external genitalia and secondary sex characters. If a thorough examination is done when the patient seeks consultation for her primary amenorrhoea the existence of such syndrome is suspected and gonadal biopsy finally establishes the diagnosis.

It is generally believed today that the hormonal excretion pattern in this syndrome is that of a normal man but non-development of male secondary sex characters is due to the non-responsiveness of target organs to androgens because of some inherent autosomal dyscrasia and not due to sex chromosome defect. Burno has made a through study of the hormone pattern of these cases and found hormone excretion pattern is like that of a male.

In this particular case the hormone pattern did not compare to that of an adult male but hormone assay has been done by crude methods. The vaginal cytology proved hypo-oestrogenic.

The other point in such a case is about the development of the vagina when all other Mullerian tissue is suppressed. Full development of vagina is a strong evidence of Meyer, Vilas and Fulham's theory of development of vagina from the epithelium of urogenital sinus. The generally accepted belief is that the lower third of vagina is developed from urogenital sinus and the upper two-thirds from Mullerian ducts.

The gonadectomy is advised in these cases to prevent the development of malignancy in intraabdominal gonad and to render the person more suitable for the roll of a female.

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References

1. Burno, A.: Am. J. Obst. & Gynec., 84: 710, 1962.
2. Fluhmann, C. F.: Obst. & Gynec., 15: 62, 1960.
3. Meyer R. Zur: Frage der Entwicklung der menschlichen vagina, Teil IV, Arch. Gynae., 165: 504, 1938.
4. Morris, J. Mcl: Am. J. Obst. & Gynec., 65: 1192, 1953.
5. Morris, J. Mcl and Mahesh, V. B.: Am. J. Obst. & Gynec., 87: 731, 1063.
6. Pion, R. J. et al: Am. J. Obst. & Gynec., 93: 1067.
7. Vilas, E. Uber: die Entwicklung der Manlungsgesch; 98: 263, 1932.

See Figures on Art Paper III