TESTICULAR FEMINIZATION SYNDROME

(A Case Report)

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Introduction

Testicular feminization is a term suggested some years ago for a hereditary syndrome characterized by individuals with testes who present female phenotype. Females are carriers. Many terms are used for this syndrome. Some of the recently used terms are "Psuedohermaphorodifismus masculinus with complete feminization" Oles, (1958), "an inherited type of intersex in apparently female individuals of male gonadal and chromosomal sex, without a uterus or body hair", Prader, (1957), "Hairless women with testes" Wilkins, (1965).

CASE REPORT

S.B.D. 17 years, female admitted in Civil Hospital, Ahmedabad, with history of primary amenorrhoea.

Menstrual history: Not attained menarche since puberty.

Marital history: Unmarried.

Family history: Her parents have 5 children, 2 daughters and 3 sons, the patient being the second child. One brother is elder to her and is mentally retarded. Two brothers are younger to her, one is 10 years and the other 7 years old. Both are normal. Sister is younger to her, 4 years, and has hypertrophied clitoris. Mother is healthy and has normal cycles. Mother has 3 brothers and 2 sisters. All are married having children. Grandmother is healthy.

General Examination: The height of the patient was 148 cms. weighing 42 Kg. No facial hair. Breast not well developed. Axillary hair normal. No hair on the abdomen or chest. Cardio-vascular and respiratory systems clinically normal.

Gynaecological Examination: Pubic hair of feminine distribution. Labia majora on both sides contained oblong swellings. Testicular sensation present on both sides. Swelling on right side was 2 cm x 2 cm in size and was easily reducible. Left sided swelling was 1½ cm x 1 cm in size and was also easily reducible. Internal abdominal ring on the left side admitted the tip of index finger. Clitoris and urethera normal.

Speculum Examination: Cervix not visualised and vagina ends blindly.

Vaginal Examination: Vaginal length about 1.5 cm., rugose. Uterus and ovaries not felt.

Rectal Examination: Uterus and ovaries not felt. No other masses felt.

Investigations: Hb% 12 grams; urine—Nil abnormal, E.S.R. 5 mm/hour, Android type of pelvis on X-Ray, I.V.P. right side, showing

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pyelonephritis, X-Ray skull, normal, X-Ray chest, nil abnormal. Vaginal smear: M.I. 2/55/43. Buccal-smear: Sex chromatin negative E.C.G. normal; fasting blood sugar 76.5 mg.%, Blood urea: 27.5 mg.%.

Management: Laparotomy was done on 12.4.1973. The uterus and ovaries were completely absent.

Bilateral gonadectomy was done at the level of superficial inguinal ring after ligating the spermatic cords. The gonads were sent for histopathological examination.

Histopathology Report: Two masses removed from labia consist of testes, consisting of arrested spermatogenesis. Few sertoli cells and leydig cells are seen. Patient was put on hormonal line of treatment as a maintenance therapy.

Discussion

Incidence of testicular feminization varies with different authors, ranging from 1:2000 Hauseret *et al,* (1957) to 1:62100 males Jagiello, (1962). According to Netter *et al* (1958), 15.20% of intersexual individuals with other than gonadal dysgenesis are cases of this syndrome.

There are various views regarding the etiology of testicular feminization. The exact genetic mechanism of this disease is not known. Upto now, we do not know whether the hereditary defect is primarily in the testes or in the pituitary or the hypothalamus. The defect might be in the target organs for testicular inductors and hormones. A combination of factors may exist.

The syndrome is hereditary with transmission through the maternal line. The carriers are usually normal females, but decreased axillary and/or pubic hair has been noted in otherwise apparently normal mothers, grandmothers, aunts and sisters by a number of authors. Late menarche has also been maintained as familial trait Puck *et al,* (1960). The mode of inheritence of this disorder is due to either a sex linked recessive gene or a sex linked autosomal dominant gene Grumbach and Barr, (1958).

Regarding line of management, it is clear that when the child is grown up, it is embarassing for the parents and the child to change over the sex, therefore, to avoid psychological trauma the child should be set in the sex which is most adaptable. These gonads may be left till puberty for better development of secondary sex characters, as there is evidence that these testes secret estrogens and androgen. Morris and Mahesh (1963) quoted incidence of neoplasia as 22%. According to them only one malignant tumour has been reported among teenage patients and 2 in those in their twenties, but there have been 11 malignant tumours in 50 cases of 30 years or above. Therefore, the incidence of neoplasia appears sufficient to continue to advocate removal of gonads. Some times after castration patient develops menopausal symptoms. In such cases oestrogens can some times be given. These patients usually have a strong female libido and some may be frustrated because of the short vagina. In such cases, surgical correction should be done. Clitoridectomy is also recommended in patients having enlarged clitoris.

Summary

1. A case of testicular, feminization syndrome is presented in this paper.

2. Some aspect of physiology and etiopathogenesis are discussed.

3. Some points regarding management are discussed.

References

1. Grumbach, M. M. and Barr, M. L.: Recent Progr. Hormone Res., 14: 255, 1958.

2. Hauser, G. A., Keller, M., Koller, T., Wenner, R. and Gloor, F.: Schweiz, Med. W. Schr., 87: 1573, 1957.
3. Jagiello, G. and Atwell, J. D.: Lancet, 1: 329, 1962.
4. Oles, H.: Zbe. Gynaek., 80: 1877, 1958.
5. Prader, A. S.: Med. Wschar., 82: 278, 1957.
6. Puck, T. T., Robinson, A. and Tgio, J. H.: Proc. Soc. Exp. Biol. Med., 103: 192, 1960.
7. Wiekins, L.: The Diagnosis and Treatment of Endocrine Disorders in Childhood and Adolescence. 3rd Ed. Springfield, Thomas, 1965.

See Figs. on Art Paper VII-VIII