TESTICULAR FEMINIZATION SYNDROME

(A Case Report)

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Introduction

A rare case of TFS is reported. A genetically male presenting with female habitus and primary amenorohoea. As the patient was aged 21 years, bilateral gonodectomy was done followed by estrogen replacement therapy. Following gonadectomy, plasma testosterone level as determined by RIA was 0.55 ng/ml.

Case Report

Mrs. S.P. aged 21 years female, University Student, attended Vani Vilas Hospital on 4-6-1983 with history of not attaining menarche.

Family History

She was the first born among 6 children and had 3 younger normal sisters.

General Examination

Phenotype Female, Ht. 157 cm, arm span 162 cms Wt. 40 kgs carrying angle 18°, axilary and pubic hairs absent. Breasts normally developed Tanner stage V (Fig. 1), smell and vision normal. No impairment of visual fields. No thyroid swelling and a low occipital hair line present.

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Gynaecological Examination

Labia Majora, minora and clitoris were infantile. Speculum examination, blind vagina, rugosity present, cervix not seen. Vagina admitted 1 finger easily upto 4 cm length. Cervix and uterus not felt on rectal examination, uterus not felt, no mass felt.

All investigations were within normal limits. Buccal smear, only 2% cells were chromatin positive. Serum gonadotrophins By RIA, FSH 130 MIU/ml., LH 136 MIU/ml. Total serum testosterone by RIA, 2.1 ng/ml (Normal Female range 0.12 to 0.84 ng/ml), Karyotype, 46 XY, Vaginogram, Shows blind vagina.

Diagnostic Laparoscopy Done on 15-6-83

Uterus and tubes on both sides were absent. Both gonads were seen near the internal inguinal rings pearly white in colour with smooth surface and no cysts seen. Lower poles of the gonads were connected, to the internal inguinal ring with strong muscular bands.

Laparotomy Done on 26-7-83

Uterus and Tubes were absent. Both sides pearly white gonads with smooth surface measuring 3 cm x 2 cm x 1 cm oval in shape were present. Lower pole was attached gubernaculum like structure, Frozen, section biopsy from left gonad showed testicular tissue. Gonadectomy was done on both side. Patient, had an uneventful post operative recovery.

Biopsy Findings

Testicular tubules separated by bands of connective tissue, many tubules show hyaline thickening of basement membrane. The tubules no spermatogenic activity. A few tubules

shows spermatocytes. The interstitial cells are not remarkable. Epididymis showed ductules of small size (Fig. 2).

Treatment: Bilateral orchidectomy done. are lined by cuboidal and columnar cells with Patient was discharged after advising estrogen, stillbesterol 1 mg/day as maintainance therapy.

See Figs. on Art Paper V