5-ALPHA REDUCTASE DEFICIENCY SYNDROME

(A Case Report)

By

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SUMMARY

A rare case of male intersex due to 5-alpha reductase deficiency is rported. Differential diagnosis and the management has been discussed.

Introduction

Discrepancy at any level between chromosomal sex, gonadal sex, internal genital sex, phenotype sex or sex of rearing is known as Intersex. In the male intersex chromosomal sex is male while other types of sexes are anomalous, often typically female. 5-α reductase deficency syndrome is a rare variety of male intersex. In view of such rarity the present case is reported.

CASE REPORT

Mrs. A.G., 17 year old, married for 1 year, attended the Outpatient department of Obstetrics and Gynaecology, Eden Hospital, Medical College, Calcutta on 31st May, 1986 with the complaints of: absence of menarche, gradual enlargement of phallus for one year, recent development of breaking of voice for 3 months and dyspareunia. There was no history of drug intake. They were four sisters and two brothers. She was the third among the sisters. Two elder sisters were married and both of them were fertile. Younger sister was 14 years' old, had no physical abnormality and had her menarche one and half years back.

On examination she was slender, 150 cm tall, weight 40 kgm, breasts not developed at all and axillary and pubic hairs were scanty. Abdominal examination revealed bilateral firm masses (4 cm x 2 cm) at the level of the external inguinal ring (Fig. 1) with impulse on coughing. On pelvic examination (including EUA) enlarged clitoris (3.5 cm), narrow introitus and short blind vagina (4 cm) was found (Fig. 2). There was no uterus.

Investigations: Buccal smear-negative for chromatin, karyotyping-46 XY (Fig. 3), urinary 17-Ketosteroids—18 mg/24 hrs, plasma testosterone-4.5 ng/ml, plasma oestradiol-13 pg/ml and laparoscopic examination-neither uterus nor gonad was found in the pelvis.

Treatment: Clinical profile and investigations reports were suggestive of androgen insensetivity syndrome due to 5-alpha reductase deficiency. So bilateral gonadectomy with repair of inguinal hernia along with widening of introitus was done. This was followed by oestrogen therapy for six months. Histopathological examination of the removed gonads showed attenuated seminiferous tubules lined by immature germ cells, stroma studded with leydig cells and no evidence of malignancy.

Discussion

5-alpha reductase convert testosterone into dihydrotestosterone which is necessary for masculinization of cloaca and genital tubercle during intrauterine and

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early neonatal life. At puberty testosterone can act for dihydrotestosterone on the same organs. So in 5-alpha reductase deficiency infant is born as phenotype female but gradually tends to virilize at puberty (Imperato-Mcginely 1979).

It is an autosomal recessive disorder usually runs in family but may be also sporadic one as happened in this case.

The case was differented from female intersex by buccal smear examination and karyotyping and from true intersex by histopathological examination of the removed gonads (Van Niekerk, 1976). It was not a case of testicular feminization (cytosol receptor lack) as (a) in the complete variety—there is no virilization and besides there is breast development at puberty (Morris, 1953) and (b) in the incomplete variety usual presentation is of varying degree (according to severity) in a phenotype male (Dewherst, 1981).

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References

1. Dewherst, S. J.: Integrated Obstet. Gynec. for Post graduates Ed. Dewherst, S. J., 3rd Ed. 44, 1981.
2. Imperato-McGinley, J.: New Eng. J. Medicine, 300: 1233-37, 1979.
3. Morris, J. M.: Am. J. Obstet. Gynec. 65: 1192-1211, 1953.
4. Van Niekerk, W. A.: Am. J. Obstet. Gynec 126: 890, 1976.

See Figs. on Art Paper I