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Familial Testotoxicosis

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Familial testotoxicosis is a rare variety of isosexual precocious puberty resulting from autonomous Leydig cell hyperplasia of testes(1). It is transmitted as a male limited autosomal dominant disorder(2).

Case Report

A 4-year-old boy was brought to us for

evaluation of masculine stature with inappropriately large penis (*Fig. 1*) observed for about 2 years. Thereafter, the patient gradually developed a masculine body habitus with moustache and adult like hoarse voice. However, expulsion of semen on one occasion—when the phallus was handled by the mother during a bath—made the parents anxious.

Nevertheless, the boy had no behavioral

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*Received for publication: November 25, 1992;
 Accepted: September 1, 1993.*

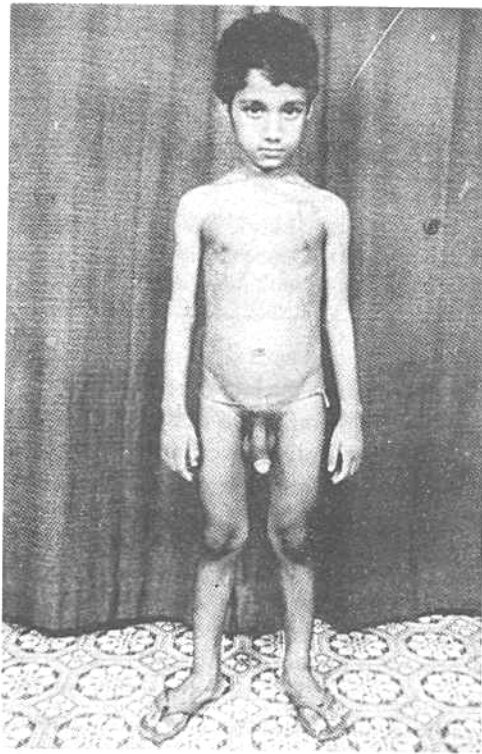


Fig. 1. Photograph showing physical features of the patient.

problems. There was no history of headache, convulsion nor dimness of vision suggestive of any pathology in the brain. *Hi* had no salt-craving nor any pigmentation related to adrenal disorder. He had no history of any relevant drug-intake. Surprisingly, the family history revealed the incidence of similar precocity in his father and one of his two paternal uncles. His only sib, an elder sister was found normal. There was no history of consanguinity nor any relevant history on the maternal side.

His height was 117 cm (>95th centile of ICMR standard(3), height-age: 7 years), weight 18 kg (weight-age: 5 years) and BE 94/60 mm Hg. Height (117 cm) was less

than arm-span (118 cm) and upper segment (59 cm) to lower segment (58 cm) ratio was greater than 1. Distribution of pubic hair was of Tanner Stage P₅; the penis was of adult size and shape with stretched penile length 9 cm. Both the testes were symmetrically enlarged with normal configuration and each had a volume of 8 ml. He had a muscular physique with moustache, yet no permanent tooth had erupted; however, all the deciduous teeth (twenty) were present. There was no gynecomastia and no abdominal mass was detected. Ophthalmoscopy was non-contributory.

Investigations showed bone-age 13 years (Greulich and Pyle's Standard) (4); semen analysis showed total count 25 million spermatozoa per ml with maturity 55%, motility 40%, abnormal forms 50% and viability after 4 hours 10-15%. Testicular biopsy demonstrated normal post pubertal histological pattern. X-ray chest was normal. Ultrasonography of abdomen was normal and without any adrenal enlargement. CT scan of whole brain—both plain and contrast films showed no intracranial lesion. The assay of serum hormones were done on two occasions (*Table I*). There was a definite rise of testosterone without any rise of FSH and LH *pan passu*. Moreover, a urinary pregnancy, coolor test (R) for HCG was negative.

Considering all these, it was diagnosed as a case of familial testotoxicosis and advised rationally after counselling the parents properly.

Discussion

Precocious puberty in boys refers to sexual development before the age of 9 years and is not a common condition; above all familial testotoxicosis is even rarer. The latter condition is known to result from

TABLE I—Summary of Hormone Assay

Hormone	Values on date		Normal level in boys	
	2.9.1992	16.4.1993	Pre-pubertal (childhood)	Pubertal and Post-pubertal
Testosterone (ng/ml)	3.8	3.9	<1	2.9-8.6
FSH (min/ml)	0.11	0.12	<3	1.5-9.0
LH (min/ml)	0.08	0.08	<1	1.5-9.0

autonomous Leydig cell maturation independent of any influence of gonadotropins(1,2,5). The exact mechanism is uncertain(6). Spermatogenesis in this condition may be initiated by the high intratesticular concentration of testosterone, even though the FSH level is prepubertal. Testicular histology also suggests that spermatogenesis develops subsequent to Leydig cell maturation and testosterone secretion(2).

Constitutional (idiopathic) variety of precocious puberty is caused by earlier activation of hypothalamic-pituitary-gonadal axis and there is pulsatile rise in serum level of gonadotropins as well as a brisk response to LHRH-stimulation(7). These two features could not be properly studied in our case because of financial constraints. In spite of these difficulties, the persistently low levels of gonadotropins in the face of elevated testosterone levels on two occasions rule out the possibility of gonadotropin-dependent precocious puberty. The normal cranial CT scan report, excluded the possibility of hypothalamic hamartoma too(8). Similarly, clinical features along with normal ultrasonogram report of abdomen ruled out congenital adrenal hyperplasia.

Above all, early age of onset (2 years in this case) together with occurrence of similar precocity in successive generations

(father and one uncle) affecting only the male family members of our patients with testicular maturation favored the diagnosis of familial male gonadotropin independent precocious puberty(9) (the other name of which being familial testotoxicosis).

Treatment of familial testotoxicosis includes psychological support and measures to prevent premature epiphyseal fusion. The latter is accomplished by drugs that inhibit synthesis of testosterone *e.g.*, ketoconazole(10) or, those that block testosterone action, *e.g.*, spironolactone(11).

Although spermatogenic dysfunction is reported in some, most affected individuals achieve adult pattern of LH secretion and fertility when they reach adulthood(2).

Acknowledgement

The authors wish to thank Prof. P.K. Mukherjee, Principal, Calcutta National Medical College and Hospital for granting them permission to publish the case report.

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Filarial Chyluria

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Chyluria is the passage of milky urine due to a lymphourinary fistula, the cause of which may be parasitic or non-parasitic. Filariasis is the commonest cause of chyluria.

Filarial chyluria has been reported to have a clinical incubation period of 15-20 years(1), while some authors have quoted it to be 5 years also. It is, therefore, uncommon in children, there being no report of its occurrence in children in a statistical analysis of more than 2000 cases of chyluria(2). We are reporting a case of microfilaricidal chyluria in a 10-year-old male child.

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Received for publication: December 19, 1991;
Accepted: August 14, 1993