

INCOMPLETE TESTICULAR FEMINIZATION SYNDROME

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Incomplete testicular feminization syndrome is an important cause of male pseudohermaphroditism. Such patients either present with labial swellings in early childhood or with primary amenorrhea at puberty. End organ insensitivity to testosterone is the, basic causative factor.

CASE REPORT

A three and half year old girl presented to us with bilateral symmetrical swellings in the upper part of each labium majus. These were noted by her mother at birth. There was no history of any case of abnormal genitalia or inguinal hernia in her maternal aunts. Neither was there any record of intake of drugs like alcohol, hydantoin or cimetidine by the mother during pregnancy. On physical examination the girl was of normal height and weight according to her age. Abdominal examination was normal. Examination of the genitalia revealed a normal clitoris and labium minora except for two symmetrically placed, mobile, nontender and irreducible swellings 2x2 cm in the upper part of each labium majus. The depth of normally placed vagina was 2.5 cm. A separate urethral opening was present. Rectal examination was done and uterus was not palpable. The age-adjusted testosterone level was raised as compared to that for a normal girl, i.e., 20 ng/dl (normal prepubertal: 1-11 ng/dl). The Barr body test was negative and the karyo-type was XY. Ultrasonography failed to outline the uterus, cervix, fallopian tubes or ovaries. Intravenous urography showed normal appearance of the kidneys, ureters and bladder. The . swellings were explored surgically and on gross examination they appeared to be testes: They had epididymes and vas deferens. Wedge biopsies were taken from both sides. Histopathology confirmed the presence of testicular tissue. It consisted of variable sized juvenile seminiferous tubules lined by measurable number of sertoli cells which showed no functional activity. The intervening stroma was thick and fibrous and contained scattered leydig cells. Considering all these facts a diagnosis of incomplete testicular feminization syndrome was made and bilateral orchidectomy was performed at a later date.

DISCUSSION

The incomplete testicular feminization syndrome is inherited in X linked recessive fashion. The basic pathology is that there is a lesser degree or insensitivity to androgens in these patients compared to those with the complete syndrome. There is decreased binding of androgens to end organ cells and decreased nuclear uptake of dihydrotestosterone-cytsol receptor complexes. The fibroblast androgen receptor studies often show subtle qualitative abnormalities in the receptors in these patients¹. With clear cut clinical features, a confident clinical suspicion can be made on the very first visit of the patient. The important investigation is karyotyping which will show 46 XY. The Barr body test will be chromatin negative. The histopathology of patient's gonads is the same in both variants of testicular feminization syndrome. It resembles the histology of undescended testes. Patients of testicular feminization syndrome should be reared as females because the phallus is inadequate for male gender role². After puberty the gonads have a higher risk of developing malignancy. The incidence of malignancy in these gonads is 2.6% before the age of 15 years and about 33% by the age of 50 years³. Therefore bilateral orchidectomy is mandatory. The patients of incomplete testicular feminization

syndrome must be diagnosed early and treated before the age of puberty to avoid the risk of virilization at that stage⁴ some features of which, e.g., voice changes are irreversible. So bilateral orchidectomy should be done in infancy. After orchidectomy extra glandular conversion of adrenal androgens into estrogen is probably responsible for the satisfactory breast development⁵. At puberty exogenous estrogen supplementation may become necessary if satisfactory breast development does not occur⁶. So bilateral gonadectomy was done in this patient when she first reported. She will have a regular follow-up up till puberty. The patients with androgen insensitivity syndrome usually do not need surgical correction of external genitalia unless the adult vagina is too small for satisfactory coitus. Most of them can be treated with vaginal dilatation and they will end up with a functionally adequate vaginal cavity. Some may require vaginal replacement in late adolescence. The colon can preferably be used for this operation⁷. Patients with the incomplete variety may have clitoral hypertrophy and require clitoral recession.

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