Adreno-genital Syndrome

An 8-year-old male child presented with fever. On routine examination it was observed that there was bilateral cryptorchidism and presence of pubic hair corresponding to SMR stage 3. The scrotum was well developed and the penis was small (with no prepuce) with an urethral opening at its tip (*Fig. 1*). There was generalized hyper-pigmentation and presence of acne. These findings prompted investigation for ambiguous genitalia. Abdominal CT scan revealed presence of uterus and its adenexa and bilateral adrenal enlargement. Karotyping revealed 46-XX genotype. The serum 17-hydroxyprogesterone (17-OHP) level was 2.8 ng/mL (normal: 0.2-0.5 ng/mL) and urinary 17-ketosteroids was 18 mg/d (normal: 1-4 mg/d). The clinical findings and investigations suggested the diagnosis of a female pseudohermaphrodite due to congenital adrenal hyperplasia resulting in adreno-genital syndrome. The family preferred to continue to rear the child as a male and therefore no treatment was offered.

Congenital adrenal hyperplasia is the most common cause of female pseudohermaphroditism. It can rarely be a result of virilizing adrenocortical tumors. 21-hydroxylase deficiency is the commonest cause of congenital adrenal hyperplasia, followed by 11b-hydroxylase deficiency. The clitoris may be enlarged to resemble a penis and labial fusion can resemble a scrotum, as the present case. The severity of virilization is greatest in salt losers. These children have premature development of pubic and axillary hair, acne, masculine voice and build. Children with 11b-hydroxylase deficiency in addition are often hypertensive and may have gynecomastia. Female pseudohermaphrodites due to CAH can be distinguished from those due to other causes by the elevated levels of serum 17-OHP and urinary 17-ketosteroids. Antenatal diagnosis is possible by detecting elevated 17-OHP in the amniotic fluid. The sex of rearing depends on the extent of virilization. If the family wishes to rear as females then appropriate surgical and endocrinal therapy will have to instituted.

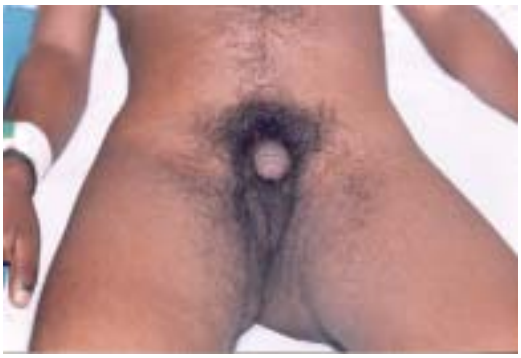

Fig. 1. Female pseudohermaphrodite with premature pubic hair, hypertrophied clitoris and fused labia.

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