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Precocious Puberty in a Severely Virilized 46, XX Child: Diagnostic and Therapeutic Challenges

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Abstract

The most common cause of peripheral precocious puberty (PPP) is congenital adrenal hyperplasia (CAH) due to 21-Hydroxylase deficiency resulting in excess androgen production independent of activation of the hypothalamic pituitary gonadal axis. A 6-years and 6 months-old child, 2nd issues of non-consanguineous parents presented to us with appearance of pubic hair and gradual enlargement of penis for last 6 months. The child was born at term and raised as male but had absent gonad in scrotum since birth. Physical examination revealed normal vitals, height between 75th to 90th centile. Examination of external genitalia revealed pubic hair stage-3, stretched penile length- 5 cm with urethral orifice at the tip of penis (Prader stage 5) and no palpable gonad in scrotum. Investigations revealed elevated testosterone and 17-hydroxyprogesterone. Karyotyping revealed 46, XX genotype. Bone age was markedly advanced. MRI of abdomen revealed rudimentary uterus but ovaries not clearly visualized. Based on the clinical manifestations, laboratory findings, and radiology imaging, patient was diagnosed with PPP due to CAH. The condition was well explained to parents of the patient. However, they were unwilling for our patient to change gender and treatment was started using hydrocortisone at a dose of 10 mg/m^2 body surface area. The most common form of CAH is 21-Hydroxylase deficiency, accounting for approximately 95% of cases. Androgen excess in CAH in 46, XX individuals leads to a range of clinical manifestations, including ambiguous genitalia at birth, enlarged clitoris, heterosexual precocious puberty etc., even the external genitalia may appear fully male (Prader 5). Early recognition is crucial to prevent compromised adult height and long term complications. This case highlights the importance of considering CAH in children presenting with precocious puberty particularly when testis is absent or prepubertal despite virilization. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S69]*

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