Simple virilising congenital adrenal hyperplasia-late presentation
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Case report
A 32 year old female presented to the out-patient department of Sheri Kashmir Institute of Medical Sciences, Srinagar with a complaint of excessive hair growth all over the body. Her relatives confirmed the presence of some genital ambiguity at birth that was never evaluated due to social taboo and stigma associated.

On detailed examination she had a height of 130 cms, Body Mass Index of 26.9 Kg/m², Blood Pressure was 130/80 mmHg and male pattern of hair distribution all over the body [Ferriman Gallwey score of 30]. Breasts were poorly developed [Tanner I] and she was ammenorrhic since childhood. There was male pattern of baldness and genital examination revealed clitoromegaly of Prader stage IV, ruggated labial skin without palpable gonads (testes) in the folds.

Ultrasonography showed normal ovaries, uterus and fallopian tubes. Extreme elevation of 17-hydroxyprogesterone (17OHP), levels of testosterone approaching or exceeding the male range was observed with a karyotype of an ordinary female: 46, XX. Dyselectrolytemia was never documented after serial serum electrolyte analyses.

Fig. 1 shows male pattern of scalp baldness.
Fig. 2 shows male pattern of facial hair distribution with temporal balding.

Fig. 3 shows excessive hair growth on chest with Tanner stage 1 breasts.

Fig. 4 shows clitoromegaly Prader IV stage with partial labial fusion.

**Discussion**
Congenital Adrenal Hyperplasia (CAH) is the commonest etiological factor causing ambiguous genitalia\(^1\,^2\). The patients have ambiguous genitalia at birth with a normal female karyotype. Over 90% patients with CAH have 21 hydroxylase deficiency, with
buildup of 17OHP – a byproduct of pathway prior to the block\(^3\). A diagnosis of Simple Virilizing Congenital Adrenal Hyperplasia is usually confirmed by discovering extreme elevations of 17OHP along with moderately high testosterone levels. A cosyntropin stimulation test may be needed in mild cases, but usually the random levels of 17OHP are high enough to confirm the diagnosis.

The optimal treatment for CAH continues to be a challenge. Endocrinologists, gynaecologists specializing in reconstructive surgery, urologists, fertility specialists, dieticians, sex therapists, biochemists, geneticists, psychologists and clinical nurse specialists all have a role to play and need to be in close communication.

The primary goals of hormone replacement are to protect from adrenal insufficiency and to suppress the excessive adrenal androgen production. Glucocorticoids are provided to all children and adults with all but the mildest and latest-onset forms of CAH. The glucocorticoids provide a reliable substitute for cortisol, thereby reducing ACTH levels. Reducing ACTH also reduces the stimulus for continued hyperplasia and overproduction of androgens. In other words, glucocorticoid replacement is the primary method of reducing the excessive adrenal androgen production in both sexes. A number of glucocorticoids are available for therapeutic use. Hydrocortisone or liquid prednisolone is preferred in infancy and childhood, and prednisone or dexamethasone are often more convenient for adults. The glucocorticoid dose is typically started at the low end of physiologic replacement (6–12 mg/m\(^2\)) but is adjusted throughout childhood to prevent both growth suppression from too much glucocorticoid and androgen escape from too little. Serum levels of 17OHP, testosterone, androstenedione, and other adrenal steroids are followed for additional information, but may not be entirely normalized even with optimal treatment.

Mineralocorticoids are replaced in all infants with salt-wasting and in most patients with elevated renin levels. Fludrocortisone is the only pharmaceutically available mineralocorticoid and is usually used in doses of 0.05 to 2 mg daily. Electrolytes, renin, and blood pressure levels are followed to optimize the dose\(^4\). The mainstay of treatment is suppression of adrenal testosterone production by a glucocorticoid such as hydrocortisone. Mineralocorticoid is only added in cases where the plasma renin activity is high.

Recent additions have been made to the treatment protocols of CAH to preserve growth that includes aromatase inhibition to slow bone maturation by reducing the amount of testosterone converted to estradiol, and use of blockers of estrogen for the same purpose.

Surgery need never be considered for genetically male (XY) infants because the excess androgens do not produce anatomic abnormality. However, surgery for severely virilized female (XX) infants is often performed and has become a subject of debate in the last decade. Surgical reconstruction of abnormal genitalia has been offered virilized girls with a purpose to make the external genitalia look more female than male, help these girls to participate in normal sexual intercourse when they grow up, to improve their chances of fertility and to reduce the frequency of urinary infections\(^5\).
References


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