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RESEARCH ARTICLE

An Unusual Presentation of 11β-Hydroxylase Deficiency: Gynecomastia with Precocious Puberty and Hypertension in a Child

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ABSTRACT

Background: Congenital adrenal hyperplasia due to 11\beta-hydroxylase deficiency is a rare autosomal recessive disorder caused by mutations in the CYP11B1 gene, leading to impaired cortisol synthesis. This results in excess adrenocorticotropic hormone (ACTH) stimulation, adrenal hyperplasia, and overproduction of adrenal androgens. Accumulation of 11-deoxycorticosterone (DOC), a potent mineralocorticoid, leads to hypertension and hypokalemia. It accounts for a small proportion of Congenital Adrenal Hyperplasia cases and may present with features of virilization and early puberty.

Case Presentation: A 6-year-old male child presented with bilateral breast enlargement and premature development of pubic hair for 2 months. There was no history of exposure to exogenous hormones or similar complaints in the family. On examination, the child had height >97th percentile for age with features of precocious puberty (Tanner stage II). Blood pressure was elevated for age. No genital ambiguity or testicular enlargement was noted.

Laboratory evaluation revealed low serum cortisol with elevated adrenal androgens. Further hormonal analysis showed markedly increased 11-deoxycortisol levels with suppressed plasma renin activity, suggestive of mineralocorticoid excess. Serum electrolytes showed hypokalemia . Bone age was advanced compared to chronological age.

Based on clinical features of precocious puberty with hypertension and supportive biochemical findings, a diagnosis of 11\beta-hydroxylase deficiency was made. The child was initiated on hydrocortisone therapy with advice for regular follow-up and blood pressure monitoring.

Conclusion: This case highlights an atypical presentation of 11\beta-hydroxylase deficiency with gynecomastia and precocious puberty. The presence of hypertension alongside androgen excess is a key diagnostic clue. Early recognition and treatment with glucocorticoid therapy are essential to prevent complications such as persistent hypertension, early epiphyseal closure, and compromised adult height.

Keywords: CAH,11-Beta Hydroxylase,Precocious puberty,Hypokalemia,Hypertention

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Introduction

Congenital Adrenal Hyperplasia (CAH) due to 11 beta-hydroxylase deficiency is a rare autosomal recessive disorder, accounting for approximately 5–8% of CAH cases. It is caused by mutations in the CYP11B1 gene, leading to impaired conversion of 11-deoxycortisol to cortisol. The resultant cortisol deficiency triggers increased adrenocorticotropic hormone (ACTH) secretion, causing adrenal hyperplasia and overproduction of adrenal androgens and mineralocorticoid precursors.

As per literature,21-hydroxylase is the commonest of all the CAH but absence of Ambiguous genitalia,virilisation and salt wasting and presence of Hypertension and hypokalemia rules out thr commonest presentation(21-Hydroxylase deficiency)

A characteristic biochemical feature is the accumulation of 11-deoxycorticosterone (DOC), which has potent mineralocorticoid activity, resulting in hypertension and hypokalemia, distinguishing it from other forms of CAH. Simultaneously, excess androgen production leads to precocious puberty, manifesting as early pubic hair, penile enlargement, and accelerated growth with advanced bone age in affected male children. Testicular size typically remains prepubertal, helping differentiate it from central precocious puberty.

Early diagnosis is essential to prevent complications such as persistent hypertension and reduced adult height due to premature epiphyseal closure. This case highlights the clinical and biochemical profile of 11\beta-hydroxylase deficiency in a male child.

Case Report

A 6-year-old male child presented to our hospital with a total duration of illness of 2 months, with chief complaints of bilateral breast enlargement and development of pubic hair. The symptoms had an insidious onset and gradually progressed. There was no history of exposure to exogenous hormones, chronic illness, headache, visual disturbances, or similar complaints in the family.

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On physical examination, the child was alert, cooperative, and oriented. Anthropometric measurements revealed:

Parameter	*Value*	*Interpretation / Reference*
Height	138 cm	>97th percentile for age
Weight	43 kg	>97th percentile for age
Blood Pressure	>95th percentile + 12 mmHg	Elevated (above age & height norms)

Above anthropometry suggestive of Accelerated growth and Hypertension.

On systemic examination, secondary sexual characteristics showed pubic hair development (Tanner stage II) and bilateral gynecomastia. There was no axillary hair. External genitalia were normal male. Testicular volume was 3 mL bilaterally, consistent with prepubertal status, suggesting gonadotropin-independent (peripheral) precocious puberty.

Skeletal assessment revealed bone age advanced beyond chronological age, indicating prolonged androgen exposure.

Laboratory investigations including complete blood count, serum electrolytes, renal and liver function tests were within normal limits. Hormonal evaluation revealed the following:

Test parameter	*Result*	*Reference range*
17-Hydroxyprogesterone (17-OHP)	1.44 µg/L	0.03 – 0.90 µg/L
Androstenedione	5440 ng/L	80 – 500 ng/L
Testosterone (Total)	1.73 µg/L (173 ng/dL)	0.02 – 0.20 µg/L
DHEA	6.79 µg/L	Not specified
Cortisol	63.60 µg/L	25 – 230 µg/L
11-Deoxycortisol	46.50 µg/L	0.20–2.50 µg/L
11-Deoxycorticosterone (DOC)	2.61 µg/L	0.02 – 0.34 µg/L
Progesterone	0.19 µg/L	0.22 – 1.10 µg/L
21-Deoxycortisol	0.03 µg/L	Not specified
(17-OHP + 21-Deoxycortisol) : Cortisol Ratio	0.02	< 0.53

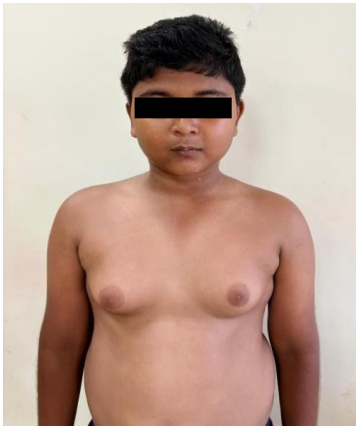

Fig 1: Bilateral Breast Enlargement

Fig 2: Pubic hair Development

Fig 3: X-ray showing advanced skeletal age.

The hormonal profile showed marked elevation of 11-deoxycortisol and 11-deoxycorticosterone along with significantly elevated adrenal androgens, consistent with 11\text{-}\beta hydroxylase enzyme deficiency.

Skeletal assessment revealed bone age advanced beyond chronological age, indicating prolonged androgen exposur

Based on the constellation of findings—precocious pubic hair, prepubertal testes, tall stature, advanced bone age, hypertension, and characteristic steroid profile—a diagnosis of congenital adrenal hyperplasia due to 11\text{-}\beta hydroxylase deficiency was made.

The child was initiated on glucocorticoid therapy (hydrocortisone) to suppress ACTH-driven adrenal androgen excess and was planned for regular follow-up with blood pressure monitoring, growth assessment, and hormonal evaluation.

Discussion

11 beta-hydroxylase deficiency is a rare subtype of Congenital Adrenal Hyperplasia caused by CYP11B1 mutation, resulting in impaired cortisol synthesis. This leads to increased ACTH stimulation, adrenal hyperplasia, and excess androgen production. Accumulation of 11-deoxycorticosterone (DOC), a mineralocorticoid, causes sodium retention, hypertension, and suppressed renin activity.

Clinically, affected males typically present with features of peripheral precocious puberty such as early pubic hair, accelerated growth, and advanced bone age. In the present case, the child had precocious pubic hair development along with bilateral breast enlargement, an unusual finding. Gynecomastia may be explained by peripheral aromatization of excess adrenal androgens to estrogens or altered hormonal balance.

Biochemical evaluation shows elevated 11-deoxycortisol and DOC levels with low/normal cortisol and suppressed plasma renin activity, which differentiates it from 21-hydroxylase deficiency. The presence of hypertension is a key distinguishing feature.

Management includes glucocorticoid therapy (hydrocortisone) to suppress ACTH, thereby reducing androgen and DOC excess. Early diagnosis is crucial to prevent complications such as persistent hypertension, early epiphyseal closure, and compromised adult height. This case highlights the need to consider this condition in children with precocious puberty and hypertension.

Lastly though 21 Hydroxylase deficiency is the commonest presentation however presence of hypertension,in the background of increased adrenal androgen,one must bring 11-beta hydroxylase deficiency as the next common entity.

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