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A Case of 11β Hydroxylase Enzyme Deficiency Presenting With Precocious Puberty

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Abstract **Case Report**

Precocious puberty is a condition characterized by the development of secondary sexual characteristics before the median age for sex. It can be either central (gonadotropin dependent) also called true precocious puberty or peripheral (gonadotropin independent) also known as pseudo precocious puberty. 11β hydroxylase deficiency is the second most common cause of the peripheral precocious puberty. Here we present a 4 year male who presented with precocious puberty due to 11β hydroxylase deficiency
Keywords: Precocious puberty; Children.

INTRODUCTION

Precocious puberty (PP) is defined as the onset of secondary sexual characteristics before 9 years in males and before 8 years in females.This is due to excessive production of sex steroids which could be due to an activation of hypothalamic-pituitary-gonadal axis (GnRH dependent PP) also called as Central Precocious Puberty (CPP) or due to a nonhypothalamic mediated increase in sex steroid production (GnRH-independent PP)[1]. Out of these two types, GnRH dependent PP is the commoner one and accounts for more than 90% of girls and about 50% of boys presenting with PP [2].

GnRH-independent PPis due to congenital adrenal hyperplasia (CAH) which is a family of inborn errors of steroidogenesis, each characterized by a specific enzyme deficiency that impairs cortisol production. The majority of cases are caused by 21-hydroxylase deficiency (21-OHD) followed by 11*β*-hydroxylase deficiency (11*β*-OHD; OMIM _202010) in about 5–8% of cases [3,4].

Here we report a case of a male child who presented to us with precocious puberty and hypertension caused by11β-hydroxylase deficiency which is a rare cause of congenital adrenal hyperplasia. In females it presents with ambiguous genitalia at birth whereas in boys the diagnosis is most often delayed.

CASE REPORT

A-4-years- 1-month-old boy, 2nd born to 3rd degree consanguineous parents, presented to paediatric out-patient care with complaints of acne and appearance of axillary and pubic hair for the last 1 year. Blood pressure was 130/90 (> 99th percentile). Anthropometric measurements showed tall stature (height 122.5 cm, > 97th percentile, height age: 7y4m) and a weight of 22 kgs (weight age 7 years) with normal BMI (14.6 kg/m2). Head to toe examination shows acne over the face, facial puffiness, and downward slant of palpebral fissures, short 4th and 5th metatarsals, pectusexcavatum and vellus upper lip hair. He had left gynecomastia and muscular appearance. SMR was A4P4 with stretched penile length of 8.5 cm. Right testis was 2 cc with mild nodular consistency whereas left testicular volume was 3cc with moderate nodular consistency. There were no signs of raised ICT or hypothyroidism. Patient had hypokalemia (serum potassium: 3.3 mEq/l). Bone age estimation was 13 years.

Thyroid function tests were normal. Luteinizing hormone (0.04 mIU/ml) and follicle stimulating hormone (0.05 mIU/ml) were low with elevated testosterone (232 ng/dl). βhCG was 0.22. Steroid profile was performed which was suggestive of 11β-hydroxylase deficiency.

Table-1

Steroid Profile	Observed Values
Cortisol (µg/dl)	0.71
Aldosterone (ng/dl)	0.97
Androstenedione (ng/dl)	>2000
Total testosterone (ng/dl)	263.13
Estradiol (pg/ml)	16
17-hydroxyprogesterone(ng/dl)	334.73
DHEA (ng/dl)	107.13
DHEAS (µg/dl)	28.12
Progesterone (ng/ml)	0.28
Deoxy cortisol	>1000
Corticosterone (ng/dl)	>5000

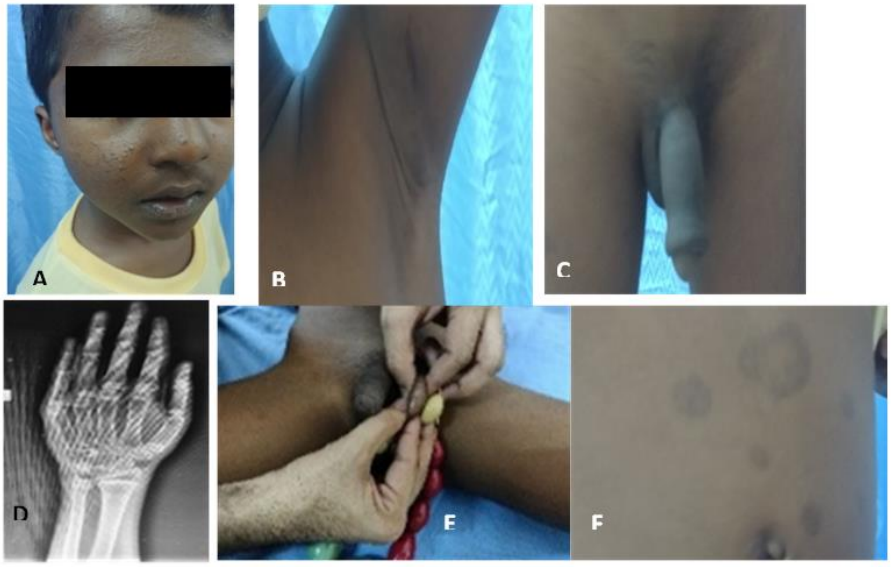

Fig-1: A.acne over the face of the patient. B.axillary hair C. genitalia D. xray for bone age. E. testicular volume measurement F. Skin

Plasma direct renin was suppressed (0.5 µIU) and 2D-Echo revealed global hypokinesia of left ventricle with mild systolic dysfunction (EF: 45%). A final diagnosis of GnRH independent precocious puberty and hypertension due to 11β-hydroxylase deficiency was made. Child was treated with oral Hydrocortisone10mg/ day (5mg-2.5mg-2.5mg), along with amlodipine 2.5 mg/day. Parents were counselled for stress dosage of glucocorticoids and advised for regular follow-up.

DISCUSSION

11β-hydroxylase deficiency accounts for about 5-8% of congenital adrenal hyperplasia. Conversion of 11-deoxycortisol to cortisol and deoxycorticosterone to corticosterone is partially blocked leading to increased levels of ACTH, accumulation of 11-deoxycortisol (which has limited biological activity) and deoxycorticosterone (which has mineralocorticoid activity) and overproduction of androgens (androstenedione and testosterone)[5,6].

The diagnosis of 11β-hydroxylase in boys is most often delayed due to lack of ambiguous genitalia and infrequent adrenal crisis in them.[6]The effect of androgen excess manifests during childhood in the form of precocious puberty as observed in our patient. The presence and severity of hypertension varies among patients with 11β-hydroxylase deficiency.[7] Boys with 11β-hydroxylase deficiency rarely present with complications of hypertension most often remains undiagnosed [8, 9], which is seen in our patient. Hence, all children who present with androgen excess should be checked for hypertension.

CONCLUSION

Our case showcases the importanceof early diagnosis, counseling and management of a young patient presenting with precocious puberty and hypertension. Our patient was treated successfully with steroids, anti-hypertensive medications, and is currently undergoing rehabilitation.

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