

Sexual Development Disorder Revealing 11-Beta-Hydroxylase Deficiency

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Case Report

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Abstract: Congenital adrenal hyperplasia (CAH) by 11-Beta-hydroxylase deficiency is a rare autosomal recessive disorder that accounts for 5-8% of CAH. We report the observation of one CAH discovered during a sexual development disorder with severe hypertension resistant to treatment in a girl aged 15 years, raised as a girl. A feminization surgery was proposed, a feminizing genitoplasty was performed without adrenalectomy because of the correction of blood pressure figures under medical treatment. Late diagnosis of CAH poses a problem of sexual orientation, so antenatal diagnosis and treatment are essential to avoid virilization of the female fetus.

Keywords: Congenital adrenal hyperplasia, adrenalectomy, Sexual development disorder.

INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a congenital, autosomal recessive disorder caused by a complete or partial deficiency of one of the adrenal steroid enzymes. There are several genetic forms whose clinical expression depends on the type and degree of deficit, sex and age of the patient at the time of diagnosis. In 95%, it is a 21 hydroxylase deficiency, followed by 11-Beta-hydroxylase deficiency in 5-8%. The incidence of 11 β hydroxylase deficiency is estimated at 1/200000 birth. It leads to the accumulation of DOC deoxycorticosterone, a decrease in the synthesis of cortisol with the raising of the ACTH feedback feed, the rate of which increases, an accumulation of 11desoxycortisol (compound S) and androgenic precursors responsible for various arrays.

Virilism in girls and precocious puberty in boys. This was the case of our patient where early puberty was the first revealing sign of this block in its unconventional form, the premature welding of the conjugate cartilage was responsible for the small size. With a severe hyper androgenism that was responsible for a sexual development disorder.

CASE REPORT

O.A, 15 years old, with no particular pathological history, admitted for hyperandrogenism:

acne, hirsutism (25 according to Ferriman and Gallwey's score), hypersudation, seborrhea, muscular hypertrophy with alopecia, anamnesis: Sexual development disorder evolving since infancy, with signs of precocious puberty and a delay statural, hypertension > 240 / 120mmhg, precocious puberty, premature pubarche and telarche. Examination of the external genitalia shows a penis of 3 cm (Prader IV). No sign of adrenal insufficiency.



Fig-I: clinical aspect before surgery

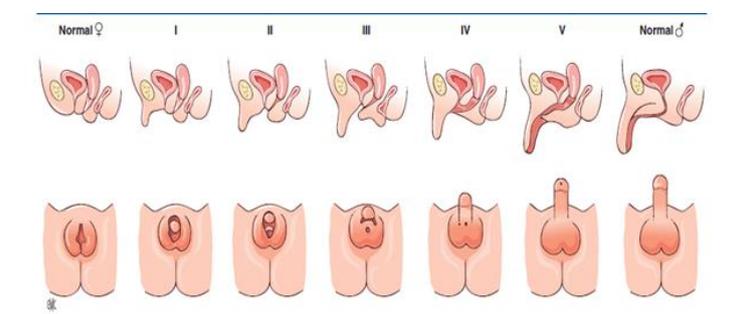


Fig-II : Prader classification

Biological results 17 OHP=29.6 ng/ml (normal value (NV): 0, 7 – 1,1) ,
 D4 androstenedione =69.3ng/l, total testosterone = 7.67 ng/ml (NV < 0, 8).cortisol = 173.6 ug/l, Aldosterone = 104pg/ml. ACTH= 97 ng/l (NV=9 – 52ng/l)

Determination of 11 deoxycortisol: results in progress.
 With the ionogram: hypokalemia at 1.9 mEq / l,
 Natremia at 144 mEq / l.

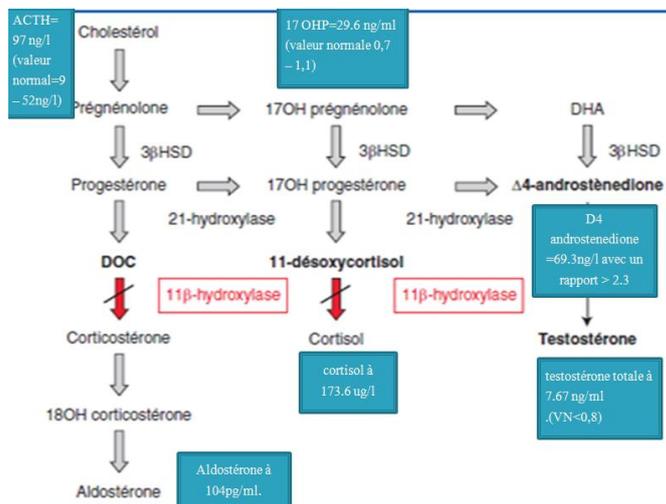


Fig-III: Biological results

Adrenal MRI showed bilateral nodular adrenal hyperplasia, karyotype: a female morphotype 46 chromosome XX. Imaging shows internal genitals of

the female type. Regarding the genetic study looking for the CYP 11 B 1 mutation, the results are in progress.



Fig-IV: Adrenal MRI

The treatment consisted of a braking of androgen production by hydrocortisone at a dose of 10 mg / m² body surface area equivalent to 20 mg / day. Concerning arterial hypertension, the treatment consisted of a triple therapy with spironolactone at a high dose of 100 mg / day, nicardipine and a central antihypertensive drug. The psychiatric report states that the child is oriented towards the female phenotype.

A feminization surgery was proposed by multidisciplinary consultation staff, a feminizing genitoplasty was performed without overrenalectomy before the correction of blood pressure figures under medical treatment. With nutritional and psychological care.



Fig-V: feminizing genitoplasty

DISCUSSION

Congenital adrenal hyperplasia (CAH) is an inherited autosomal recessive genetic endocrine disease that results from the deficiency of one of the steroidogenesis enzymes responsible for corticosteroid synthesis. 21-hydroxylase deficiency is the most common form. There is also the deficiency in 11 β -hydroxylase, 3 β -hydroxysteroid dehydrogenase, 17 α -hydroxylase and the Block in StAR[1,2].

This deficiency is at the origin of a lack of synthesis of cortisol and aldosterone, an accumulation of upstream metabolites: compound S and DOC, an excess synthesis of adrenal androgens by the only possible metabolic pathway [3-5]. Since DOC has a mineralocorticoid action [6], its excess leads to high blood pressure. And the increased synthesis of androgens during embryonic and fetal life is responsible for the virilization of female fetuses[7,8].

Hypertension is usually perfectly controlled by glucocorticoid treatment. [8,9] This is not the case in our patient, or it has evolved on its own and in this case, the introduction of a spironolactone at maximum dose in combination with a calcium channel blocker is mandatory until the correct blood pressure figures are obtained in relation to the targets set for age[6], if the goal is not reached under dual therapy, the addition of a central antihypertensive drug: methyl-dopa or rilmenidine may give good results as noted in our patient, some teams advocate the addition converting enzyme inhibitor or b cells blocking if blood pressure goal is not reached under triple therapy[10], otherwise bilateral adrenalectomy will be reserved for patients whose hormonal balance is difficult to obtain and / or hypertension resistant to medical treatment.

CONCLUSION

Late diagnosis of CAH poses a problem of sexual orientation, so antenatal diagnosis and treatment are essential to avoid virilization of the female fetus.

Abbreviations

- CAH : congenital adrenal hyperplasia;
- DHEA : dehydroepiandrosterone;
- DSD :disorders of sexual development;
- GC :glucocorticoid;
- GnRH :GnRH agonist;
- HC :hydrocortisone;
- 11 β -HSD2 :11 β -hydroxysteroid dehydrogenase type 2;
- NCCAH :nonclassic CAH;
- 17-OHP :17-hydroxyprogesterone;

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