

## CASE REPORT

# Incidental diagnosis of 17 alpha-hydroxylase deficiency: a case report

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## Abstract

A 17 alpha-hydroxylase deficiency (17OHD) is a rare form of congenital adrenal hyperplasia (CAH). In this article we discuss a case of 8-year-old girl presented with upper respiratory infection symptoms and a history of hospital admission of fatigue and dehydration. She was incidentally found to have hypertension and hypokalemia. After an endocrine workup her biochemical tests showed: metabolic alkalosis, low levels of cortisol, high levels of adrenocorticotropic hormone (ACTH) and follicle-stimulating hormone (FSH) with normal female phenotype and (46,XY) karyotype. These findings led to the diagnosis of 17OHD confirmed by regression of hypertension and hypokalemia with hydrocortisone prescription. This case shows the importance of vital signs measurement, medical history and commitment to a systematic approach.

## INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a group of disorders resulting from defect of one of enzymes necessary for biosynthesis of cortisol. A 17 hydroxylase/17.20-lyase deficiency is a rare form of CAH, accounts about 1% of all cases [1]. It is an autosomal recessive defect in the cytochrome P450 17A1 enzyme which leads to impaired cortisol and sex steroid synthesis, with compensative elevation of adrenocorticotropic hormone (ACTH), gonadotropins [2], corticosterone and 11-deoxycorticosterone (DOC). In severe forms, patients usually have female phenotype and present with primary amenorrhea and absence of secondary sexual characteristics. Patients may have either a 46XX OR 46XY karyotype. We present a case of incidental diagnosis of (17 alpha-hydroxylase deficiency [17 OHD]) in a normal female phenotype child (46, XY karyotype) with hypokalemic hypertension.

## CASE REPORT

A 8-year-old girl presented in pediatric clinic with upper respiratory infection symptoms: fever, cough, rhinorrhea and

pharyngodynia, no other accompanying signs or symptoms, a medical history of hospital admission of dehydration and fatigue, family history: older 14-year-old female sibling with uninvestigated primary amenorrhea and absence of secondary sexual characteristics. On physical examination: weight: 28 kg (69.497th percentile), height: 130 cm (62.225th percentile), BP 150/90 mmHg (99th percentile plus 5 mm Hg), Tanner stage: breast papilla elevation only (Stage1), pubic Hair: no pubic hair (Stage1), normal female external genitalia. Laboratory investigations: sodium = 142.78 mmol/L, potassium = 2.6 mmol/L, arterial blood gas (ph = 7.49, PaCO<sub>2</sub> = 22, PaO<sub>2</sub> = 96), Cortisol A.M = 3.09 ug/dl (normal = 6.2–19.4), ACTH = 198.3 pg/ml (normal = 5–60), aldosterone (standing) = 335.9 pg/ml (normal: supine = 10–160, standing = 35–300), plasma renin activity (PRA) (erect) = 0.61 ng/ml/h (normal: erect 0.98–4.18, supine: 0.51–2.64), follicle-stimulating hormone (FSH) = 31.65 mIU/ml (normal = 0.3–11), normal thyroid functions, metanephrine in 24 h urine sample = 0.79 (normal: up to 1), DHEA-S (dehydroepiandrosterone sulfate) = 2.47 ug/dl (normal: males 6–9 years: 10–85 ug/dl, females 6–9 years: 13–114), 17-OH progesterone (17-OHP) = 0.411 ng/ml (normal prepubertal: 0.07–153).

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adrenal axis and hypothalamic pituitary gonadal axis. All these results in combination with hypokalemic hypertension led to the diagnosis of CAH with 17 OHD. Pelvic imaging revealed absence of uterus, ovarian tubes and cervix. Chromosome analysis revealed 46, XY karyotype. We were unable to perform plasma levels of DOC and 18 OHB which could confirm the diagnosis but as described earlier, clinical and available biochemical results supported the diagnosis of CAH due to 17OHD. Patients with 17 OHD have tall stature due to sex steroid deficiency causing epiphysis fusion failure; our patient's stature is within normal percentile similar to reported cases of 17 OHD patients with short or normal stature [10]. Hypertension usually resolves with glucocorticoid [6] therapy therefore the patient was treated with low-dose hydrocortisone (10 mg/m<sup>2</sup>/day) and a remarkable regression of high blood pressure, elevation of potassium serum within normal range, reduction of aldosterone level was noticed which supports the diagnosis of 17 a hydroxylase deficiency. At puberty age, development of female secondary sexual characteristic in 46,XY patients is induced by prescribing low-dose 17 beta estradiol patch (6.25 mcg/day) increased gradually every 6 months to reach target dose (25 mcg/day). Testes were not detected by ultrasonography. Detecting their position and performing gonadectomy is important, but in our case we were able to follow the patient for 3 months only, so we could not start sex steroid replacement therapy nor perform gonadectomy.

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### CONFLICT OF INTEREST STATEMENT

None declared.

### ETHICAL APPROVAL

Ethical approval is not required for a case report in Syria.

### CONSENT

A written consent was obtained from the patient's father because the patient is an underage child.

### GUARANTOR

Rasha Ammar and Ahmad Ramadan.

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