中文題目：一個 17α-羥化酶缺乏症合併左側無功能性腎上腺腫瘤的病例報告

英文題目：A case report of 17α-Hydroxylase Deficiency with left non-functional adrenal tumor

作　　者：黃書恆[1,3]、黃尚志[1,2]

服務單位：高雄醫學大學附設中和紀念醫院[1]內科部 [2]腎臟內科 [3]內分泌新陳代謝科

Introduction

Congenital adrenal hyperplasia(CAH) refers to several disorders characterized by various genetic defects in the enzymes involved in cortisol and sex hormone biosynthesis in adrenal gland included steroidogenic acute regulatory protein (StAR)、20,22-hydroxylase、17 α -hydroxylase、21-hydroxylase、11b-hydroxylase and 3 β -hydroxysteroid-dehydrogenase.17 α -hydroxylase deficiency (17OHD) is a rare form (<1%) of congenital adrenal hyperplasia, usually presenting as amenorrhea, no development of puberty, hypertension, hypokalemia, and hypogonadism in female. It is due to the reduction of sex hormones and cortisol productions caused by genetic defect of 17α-hydroxylase and enhanced the production of aldosterone. We ever reported one case before, and this second case was promptly diagnosed from previous clinical experiences.

Case Presentation

A 31-year-old woman had past history of sicca syndrome under regular medical treatment with prednisolone and hydroxychloroquine for five years. She noted slightly high blood pressure as 140-150/90 mmHg since high school. There was no weakness, no excess sweating, or headache episode before. However, persistent hypertension after treated with three kinds of anti-hypertension drugs (atenolol 50mg and amlodipine 5mg/valsartan 160mg). So she received abdominal computed tomography examination, and a 1.5 cm mass over left adrenal gland was suspect as adrenal adenoma. Other associated symptoms included no menarche, lack of secondary sexual characters, and persistently growing in tall even at the end of puberty (176cm, in the top 3 %). Her parents were in normal percentile of height (Father:168cm; Mother:163cm) and denied any family history of delayed or absent puberty. Physical examinations revealed blood pressure 140/100 mmHg, no breast bud, no axillary hair, no pubic hairs, no stria, no plethora and no pigmentation. Laboratory tests showed hypokalemia (3.3mmol/L), Metabolic alkalosis (HCO3- :28.9 mmol/L), transtubular potassium gradient: 10.9, aldosterone (10.14 ng/dl) but low plasma renin activity (<0.36 pg/ml/h). Primary aldosteronism caused by adrenal tumor was impressed at first. However, further laboratory data showed low levels of sexual hormone and cortisol (estradiol: <20 pg/ml, progesterone: 6.3 ng/dl, cortisol <0.1 ug/dl), high FSH (98 mIU/ml) and LH (30.8 mIU/ml), and high ACTH (190.83 pg/ml). Primary hypogonadism and adrenal insufficiency were impressed. The clinical and laboratory results made us consider the possibility of 17α-hydroxylase deficiency from our prior experience. The further laboratory data showed low 17-hydroxypregnenolone (0.4ng/ml) and dehydroepiandrosterone (3.7ng/dl). Thus, congenital adrenal hyperplasia was impressed and gene sequence showed CYP17A1 genetic defects. She received hormone replacement therapy with cortisone acetate and spironolactone, she became more feminized and no further growth in height (176.5cm). Hypertension was also under controlled.

Discussion

17OHD　appears to be inherited as an autosomal recessive trait as a rare case of congenital adrenal hyperplasia. It is difficult to diagnose in such case because of neglect of history and careful physical examination on sexual characters. Biochemical evidence of a partial 17OHD could be observed without clinical manifestations of hypertension or hypokalemia. Besides, this adrenal mass in this case could also lead to misdiagnosis. We researched report that CAH with unilateral adrenal mass is even more rare phenotype. In this report, failure of hypertension control status post left adenectomy. Based on our prior experience, we can promptly catch the main points and do the right diagnosis to this patient. We also provide important experience to clinical worker to identify 17OHD with adrenal mass.