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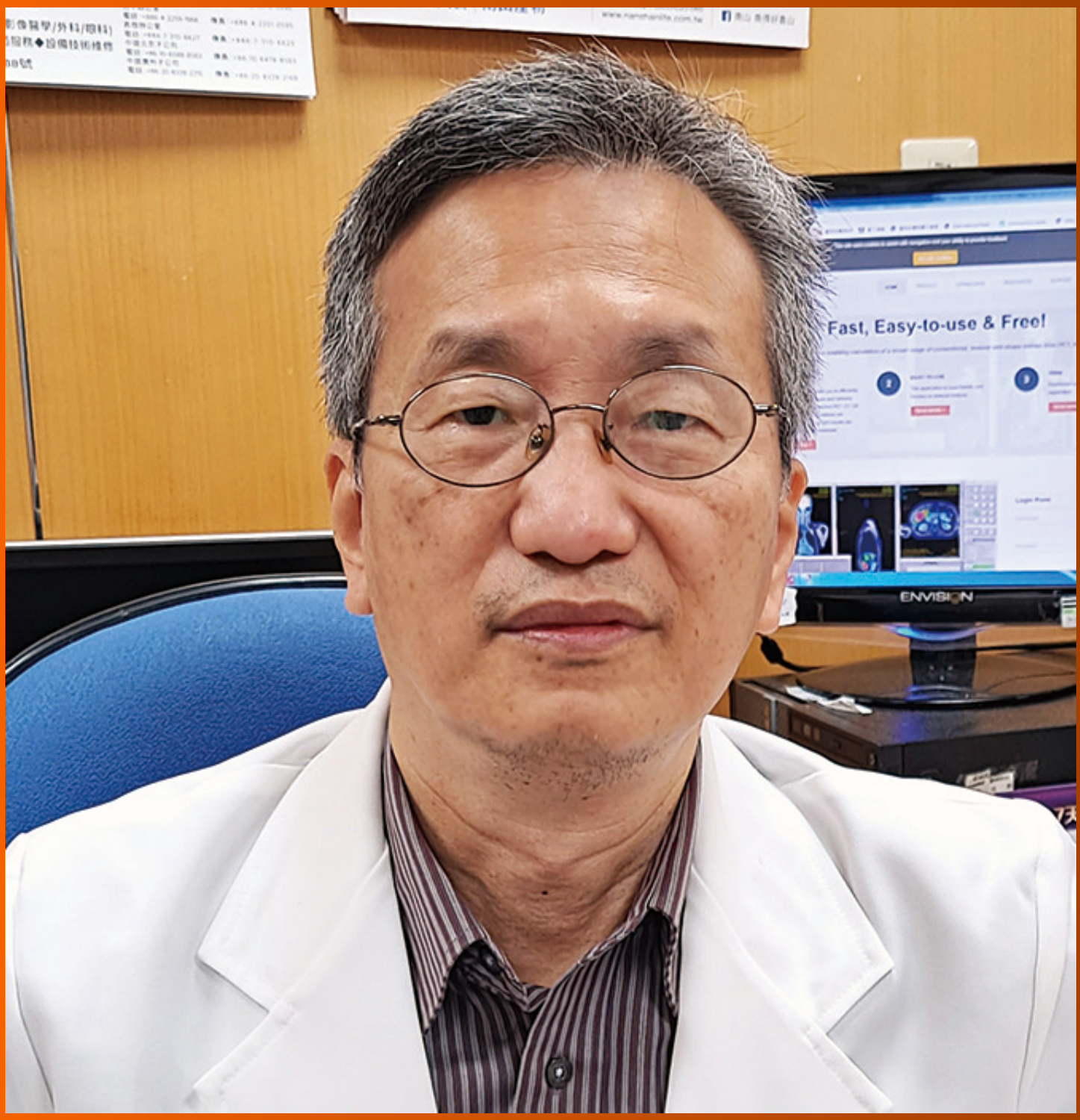

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CASE REPORT

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ABOUT COVER

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CASE REPORT

Cytochrome P450 family 17 subfamily A member 1 mutation causes severe pseudohermaphroditism: A case report

Yu Gong, Fang Qin, Wen-Jia Li, Le-Yu Li, Ping He, Xing-Jian Zhou

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Abstract

BACKGROUND

17α-Hydroxylase deficiency (17-OHD) is a rare form of congenital adrenal hyperplasia, characterized by hypertension, hypokalemia, and gonadal dysplasia. However, due to the lack of a comprehensive understanding of this disease, it is prone to misdiagnosis and missed diagnosis, and there is no complete cure.

CASE SUMMARY

We report a female patient with 17-OHD. The patient was admitted to the Department of Neurology of our hospital due to limb weakness. During treatment, it was found that the patient's condition was difficult to correct except for hypokalemia, and her blood pressure was difficult to control with various antihypertensive drugs. She was then transferred to our department for further treatment. On physical examination, the patient's gonadal development was found to be abnormal, and chromosome analysis demonstrated karyotype 46,XY. Considering the possibility of 17-OHD, the cytochrome P450 family 17 subfamily A member 1 (*CYP17A1*) test was performed to confirm the diagnosis.

CONCLUSION

The clinical manifestations of 17-OHD are complex. Hormone determination, imaging examination, chromosome determination and *CYP17A1* gene test are helpful for early diagnosis.

Key Words: Congenital adrenal cortex hyperplasia; Cytochrome P450 family 17 subfamily A member 1; 17α-Hydroxylase deficiency; Pseudohermaphroditism; Case report

Core Tip: 17α-Hydroxylase deficiency (17-OHD) is a rare form of congenital adrenal hyperplasia, characterized by hypertension, hypokalemia, and gonadal dysplasia. We report a case of 17-OHD admitted to our hospital due to limb weakness. The patient's blood pressure was difficult to control with various antihypertensive drugs. Her gonadal development was found to be abnormal, and chromosome analysis demonstrated karyotype 46,XY. The diagnosis was confirmed by the cytochrome P450 family 17 subfamily A member 1 (*CYP17A1*) test. The clinical manifestations of 17-OHD are complex. Hormone determination, imaging examination, chromosome determination and *CYP17A1* gene detection are helpful for early diagnosis.

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INTRODUCTION

17α-Hydroxylase deficiency (17-OHD) is a rare type of congenital adrenal hyperplasia (CAH), which is caused by mutations in the cytochrome P450 family 17 subfamily A member 1 (*CYP17A1*) gene, and the incidence of this disorder is approximately 1 in 50000[1]. In 1966, Biglieri *et al*[2] reported the first case of 17-OHD. To date, about 200 cases have been reported at worldwide[3-5]. At present, there is no unified standard for the diagnosis of 17-OHD, which is mainly based on the clinical manifestations, laboratory and imaging examinations, *etc*., and the diagnosis depends on the detection of gene *CYP17A1*[6]. We here report a patient with 17-OHD admitted to our hospital, and review the literature on the pathogenesis, clinical characteristics, diagnosis and treatment of the disease.

CASE PRESENTATION

Chief complaints

A 29-year-old female was admitted to the Department of Neurology in our hospital due to limb weakness for 1 d.

History of present illness

The patient had a history of syncope on several occasions, which lasted approximately 1 min and could be relieved without treatment.

History of past illness

She denied other medical history such as hypertension or coronary heart disease and had no history of smoking or alcohol consumption.

Personal and family history

Upon further investigation, the patient had primary amenorrhea and was unmarried and childless. Her parents were first cousins and her older brother was healthy.

Physical examination

At admission, her temperature was 36.5 °C, respiration rate was 23 breaths/min, pulse rate was 114 bpm, blood pressure was 184/127 mmHg, height was 180 cm, weight was 69 kg, and body mass index was 21.3 kg/m^2. The patient's breast development had only progressed to Tanner stage 1, and her vulva was similar to that of a female infant. The patient's pubic and axillary hair was undeveloped, and her Adam's apple was small. Pathological reflexes were not elicited (Figure 1).

Laboratory examinations

After admission, low serum potassium levels (2.26 mmol/L) were observed and appeared to be uncorrected after potassium supplementation. In addition, the patient had constant high blood pressure, with a maximum reading of 184/127 mmHg.

Imaging examinations

The results of the patient's biochemical and imaging examinations are shown in Figure 2 and Tables 1 and 2. Genetic analysis (Figure 3) showed homozygous mutations in the CYP17A1 gene (NM_000102.3:

Table 1 The biochemical examinations of the patient

Projects	Results	Reference range
Potassium (mmol/L)	2.26	3.5-5.5
Renin (mIU/L)	< 0.5	2.8-39.9
Aldosterone (ng/dL)	3.69	0-23.6
Angiotension II (pg/ml)	39.1	25-129
FSH (mIU/mL)	38.97	Follicular phase: 3.03-8.08; Ovulatory phase: 2.55-16.69; Luteal phase: 0.9-16.69; Postmenopausal: 26.7-133.4
LH (mIU/mL)	14.49	Follicular phase: 1.8-11.78; Ovulatory phase: 7.59-89.08; Luteal phase: 0.56-14; Postmenopausal: 5.16-61.99
Progesterone (ng/mL)	5.5	Follicular phase: < 0.1-0.3; Luteal phase: 1.20-15.9; Postmenopausal: < 0.1-0.2
Estradiol (pg/mL)	< 10	Follicular phase: 21-251; Luteal phase: 38-649; Postmenopausal: 21-312
Testosterone (ng/dL)	0.18	Male (21-49 yr): 2.4-8.71; Female (21-49 yr): 0.14-0.53
Cortisol (nmol/L)		
0 a.m.	83.46	45-135
8 a.m.	170.11	120-660
4 p.m.	106.01	55-200
ACTH (pmol/L)		
0 a.m.	8.54	0.4-4.0
8 a.m.	112.85	1.5-14.1
4 p.m.	27.15	0.95-9.5
Dehydroepiandrosterone-S (μg/dL)	17.80	95-510
GH (ng/mL)	0.648	< 8
Urine cortisol for 24 h (μg/24 h)	17.33	19.30-317.50
Urinary potassium 24 h (mmol/24 h)	204.5	25-100
Karyotype	46,*XY*	

FSH: Follicle-stimulating hormone; LH: Luteinizing hormone; ACTH: Aadreno-cortico-tropic-hormone; GH: Growth hormone.

Table 2 The imaging examinations of the patient

Projects	Results
Plain CT scan of adrenal	Bilateral multiple adrenal lesions were considered to be multiple myeloid lipomas or diffuse adrenal hyperplasia
Enhanced CT scan of adrenal	Bilateral multiple adrenal lesions were considered to be multiple myeloid lipomas
Plain MRI scan of pituitary gland	Normal
X-ray examination of both hands	The epiphyses of the fingers, metacarpal and distal ulna and radius of both hands were not healed
Ultrasonography of the pelvis	No obvious uterine echo was observed
Plain MRI scan of the pelvis	No obvious cryptorchidism and uterine accessory tissues were observed

CT: Computed tomography; MRI: Magnetic resonance imaging.

c.81C>A (p. Tyr27*)). These genetic variations have been reported by Müssig *et al*[7] and Keskin *et al*[8]. Genetic analysis of the patient showed a heterozygous mutation (c.81C>A). Unfortunately, we were unable to perform a genetic analysis of the patient's older brother, as he was unavailable at the time of testing.

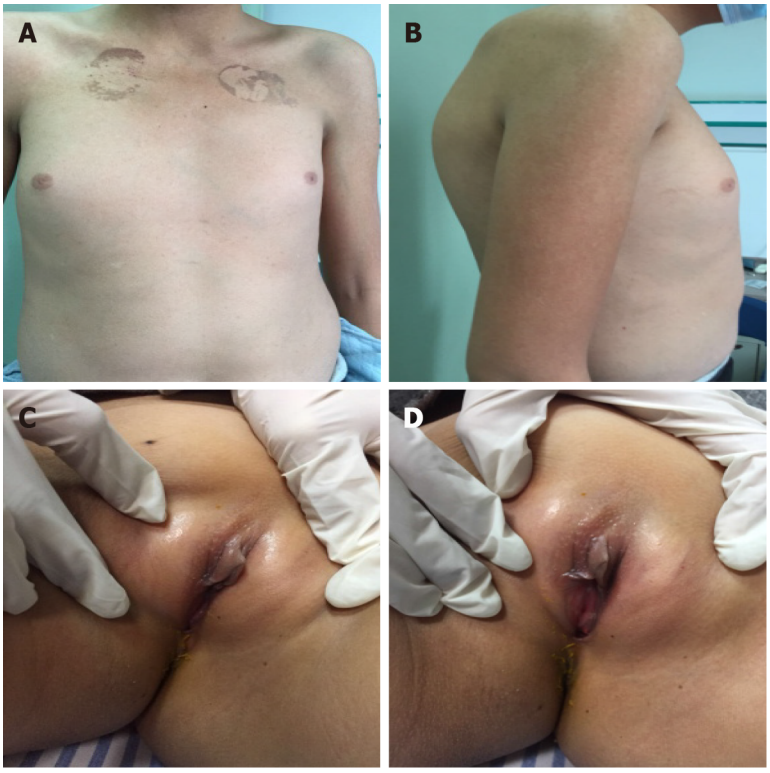

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Figure 1 Physical examinations. A and B: The patient showed absence of breast development and axillary hair; C and D: The patient's vulva was similar to that of a female infant and had no pubic hair.

FINAL DIAGNOSIS

The clinical manifestations of this patient combined with the results of various auxiliary examinations, resulted in a final diagnosis of 17-OHD associated with multiple myeloid lipomas of the adrenal gland.

TREATMENT

Following the diagnosis of 17-OHD, the patient started on oral dexamethasone (0.75 mg/d), which will be a lifelong medication. When her blood pressure and potassium level had returned to normal, she was discharged from the hospital.

OUTCOME AND FOLLOW-UP

One year later, the patient's electrolytes (serum potassium level 4.6 mmol/L) and blood pressure (130/75 mmHg) were normal on re-examination.

DISCUSSION

CAH is an autosomal recessive disorder caused by mutations in the genes encoding essential enzymes for the synthesis of corticosteroids. As a result of the imbalance between glucocorticoids and mineralocorticoids, this leads to metabolic disorders, and thus morbidity and mortality in these patients are very high[9]. The enzymes involved include 21-hydroxylase, 11β-hydroxylase, 17α-hydroxylase, 3β-hydroxysteroid dehydrogenase/isomerase *etc*. These enzyme defects (reduced or absent activity) can lead to CAH, but with different clinical manifestations[10]. Of these enzymes, 21-hydroxylase deficiency is the most common, accounting for more than 95% of cases[11], followed by 11β-hydroxylase deficiency. 17-OHD accounts for about 1% of all CAH cases, with an estimated incidence of 1 in 50000 to 100000[12,13].

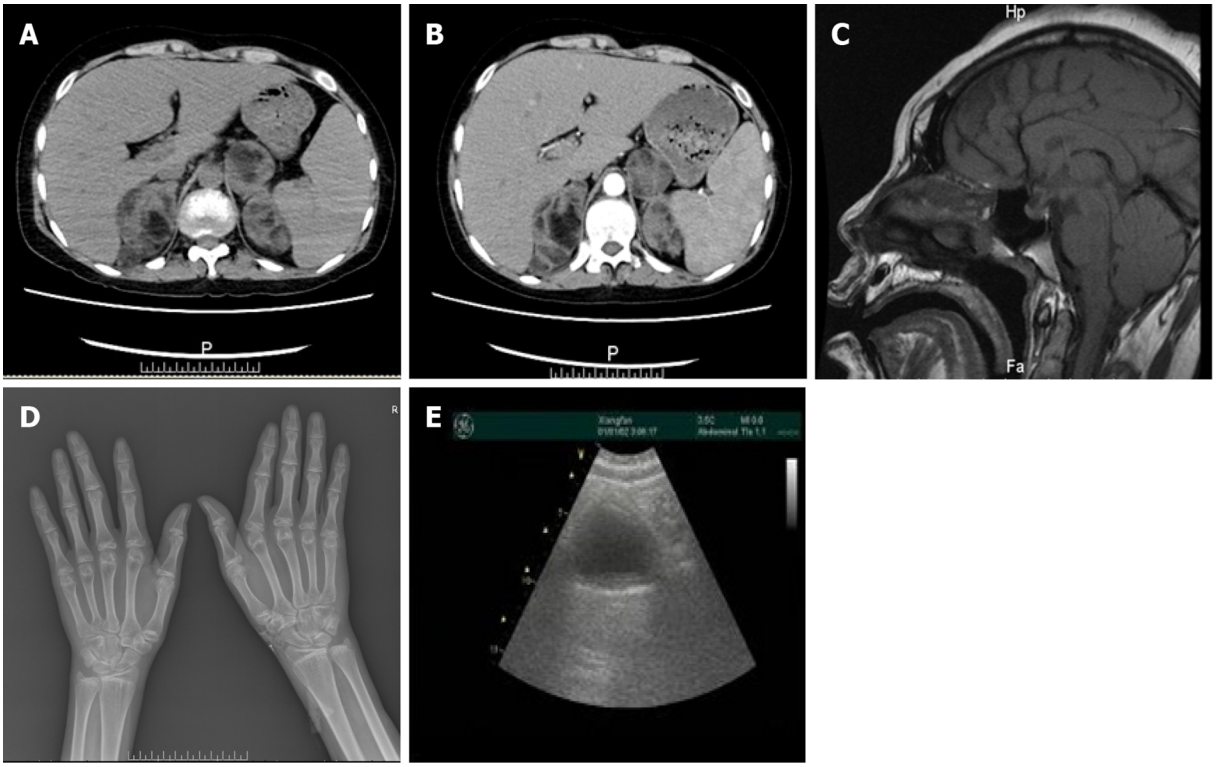

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Figure 2 Imaging examinations. A: Plain computed tomography (CT) scan of adrenals; B: Enhanced CT scan of adrenals; C: Plain magnetic resonance imaging scan of the pituitary gland; D: X-ray examination of both hands; E: Ultrasonography of the pelvis.

17-OHD mainly manifests as hypertension, hypokalemia and abnormal sexual development. *CYP17A1* encodes an enzyme with 17a-hydroxylase and 17,20-lyase activities, which is essential for the normal production of adrenal and gonadal glands[14]. When it is deficient, pregnenolone cannot translate into 17-hydroxyprogesterone and 17-hydroxylpregnenolone, resulting in the impairment of cortisol and gonadal hormones (including testosterone and estrogen)[13]. Cortisol synthesis disorders lead to an increase in adrenocorticotropic hormone (ACTH) feedback, which further activates the 17-deoxy pathway of the zona fasciculata, producing overstimulation of this pathway and increasing progesterone, corticosterone and deoxycorticosterone (DOC) synthesis. The excessive levels of these hormones then lead to hypertension, and hypokalemia. Deficiency of gonadal hormones causes primary amenorrhea in women[15] and feminization of external genitalia in men[16]. Sexual dysplasia[6] in male patients mostly manifests as pseudohermaphroditism, with infantile female genitalia and a blind end vagina, while the internal genitalia is of the male type with small testicles and dysplasia, and external genitals are difficult to distinguish, such as small penis or mammary gland development. Female patients can be normal at birth, but do not develop secondary sexual signs with primary amenorrhea. There is no pubic or axillary hair growth in both men and women. After puberty, both follicle-stimulating hormone and luteinizing hormone are significantly increased. Due to the lag in bone age, the patient's height continues to increase slowly after reaching adulthood. The bone age of our patient was below the actual age, but she was tall (180.0 cm). In addition, some patients are prone to fatigue, infection, different degrees of skin pigmentation[17] and osteoporosis. The diversity of clinical manifestations in 17-OHD patients is due to the different mutation sites on the gene encoding the enzyme and different effects on the enzyme function. Therefore, in the clinic, the existence of hypertension, and hypokalemia accompanied by sexual dysplasia, should be considered as possible 17-OHD. This deficiency should be distinguished from several other diseases, such as 5α-reductase deficiency, androgen insensitivity syndrome and 3β-hydroxysteroid dehydrogenase deficiency. However, both 5α-reductase deficiency and androgen insensitivity syndrome are generally not accompanied by hypertension and hypokalemia[18,19], and the clinical manifestations in this case did not meet the criteria for 3 β-hydroxysteroid dehydrogenase deficiency[20]. Hence, we were able to rule out these diseases.

There is no complete cure for 17-OHD, and treatment mainly consists of appropriate glucocorticoid and sex steroid hormone supplementation, social sex selection and psychological interventions. Low-dose glucocorticoid (dexamethasone or prednisolone) replacement therapy is administered in order to decrease and normalize the blood levels of 11-DOC and ACTH, which can normalize blood pressure and electrolyte imbalances[21]. However, due to the need to avoid high-dose glucocorticoid therapy and complete inhibition of the hypothalamic–pituitary–adrenal axis, DOC cannot be completely suppressed,

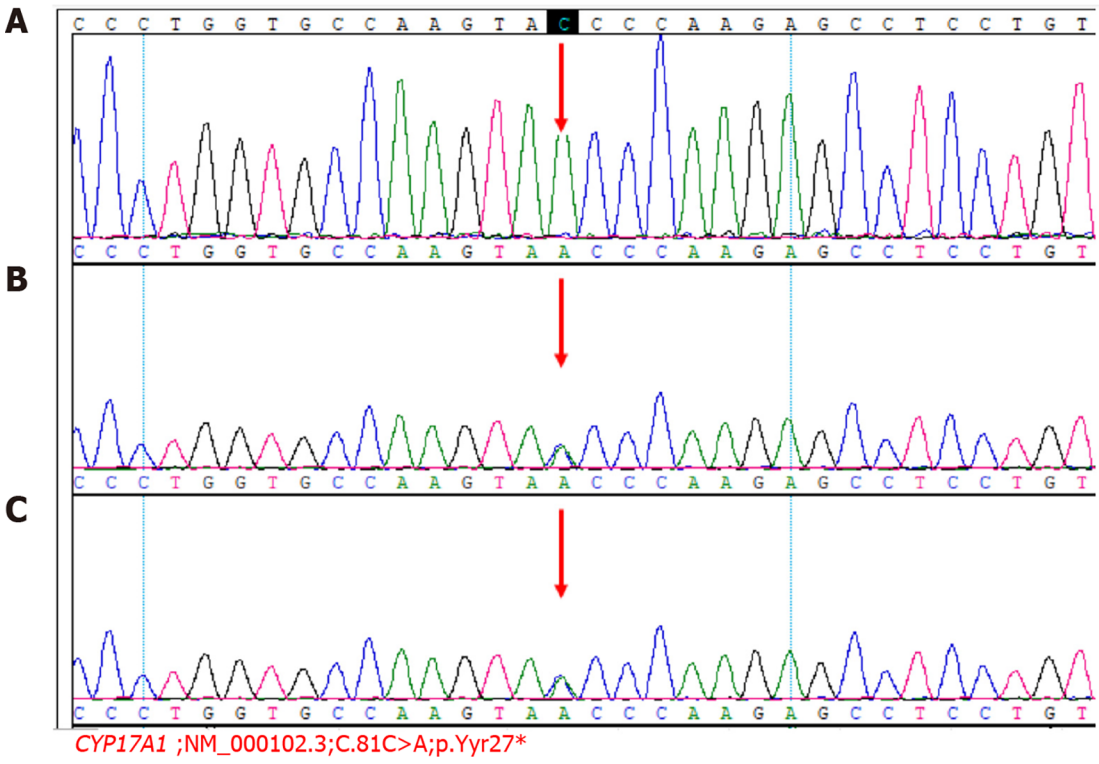

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Figure 3 Cytochrome P450 family 17 subfamily A member 1 gene analysis. A: The patient; B: Her father; C: Her mother.

and many patients will eventually become hypertensive, and corticosteroid receptor antagonists or calcium channel blockers can be used to control blood pressure[11]. Therefore, during treatment, spironolactone and nifedipine sustained release tablets are given to control blood pressure. In addition, sex hormone replacement is required for breast and uterus development and to maintain female sexual characteristics. Patients require estrogen and progestin circulation therapy to induce circulatory arrest bleeding and prevent endometrial hyperplasia. If the patient decides to be considered male, androgen replacement therapy may be given, and extensive genital reconstructive surgery may be performed, such as gonadectomy, to avoid malignant degeneration of the testes within the abdomen[13]. In this case, after full communication with the patient and her family, she decided to temporarily discontinue sex hormones and surgical treatment.

CONCLUSION

In summary, 17-OHD is extremely rare in clinical practice, and is prone to misdiagnosis and missed diagnosis. For patients with abnormal development of hypertension and hypokalemia, attention should be paid to the differentiation of this disease. Chromosome karyotype analysis and gene sequencing can help in diagnosis. Hormone replacement and antihypertensive treatment should be given as soon as possible after diagnosis. In addition, the psychological state of patients should be closely monitored.

FOOTNOTES

Author contributions: Gong Y and Qin F treated the patient; Zhou XJ drafted the manuscript; Li WJ and Li LY participated in the analysis and the interpretation of the data; He P critically revised the manuscript; all authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

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