Homozygous *CYP17A1* Mutation Identified in a Chinese Family with 46, XX and 46, XY 17α-Hydroxylase Deficiency

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OBJECTIVES

Background: Congenital adrenal hyperplasia due to 17α-hydroxylase deficiency is a rare autosomal recessive disorder, characterized by sexual infantilism, amenorrhoea, hypertension and hypokalemia, which is caused by CYP17A1 gene mutations.

Objective: To provide a descriptive analysis of 17α-hydroxylase deficiency in two female siblings with different karyotype of 46, XX and 46, XY.

METHODS

The clinical features and biochemical data of a pair of 46, XX and 46, XY Chinese siblings with 17α-hydroxylase deficiency from China were studied. Direct DNA sequence analysis of the *CYP17A1* gene was performed.

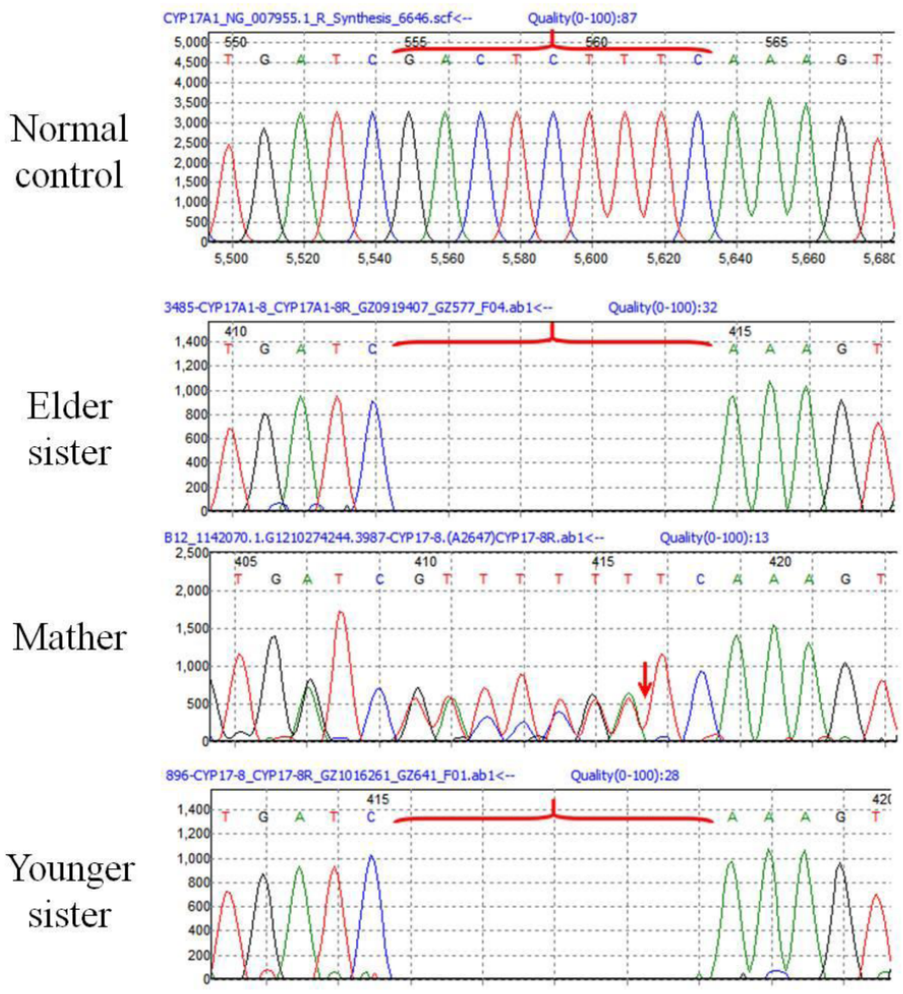

Fig. 1 Sequencing result of *CYP17A1* gene exon 8
The same homozygous mutations (c.1459_1467delGACTCTTTC (p.Asp487LysfsX20)) were identified in both patients, while their mother was found to carry heterozygous mutation.

RESULTS

The two female siblings were evaluated for the same complaints of complete lack of female secondary sex characteristics at age of 15 years 8 months and 14 years respectively. Both of the elder (46, XX) and younger (46, XY) had markedly reduced serum levels of cortisol, E_2 and T, accompanied with increased serum levels of LH, FSH, P and ACTH. The elder had normal blood pressure with normal serum K^+ level and PRA, while the younger had slight hypertension with serum K^+ and PRA in the low-normal range (3.48 mmol/L, 50 ng/L per h respectively). Pelvic ultrasonography revealed a pre-pubertal uterus in the older, and absence of ovaries and uterus in addition to a blindending vaginal tract in the younger. Cosyntropin administration did not cause a rise in serum cortisol and 17OHP levels but a rise in serum P (0.6-4.9 ng/ml, 1.8-4.9 ng/mL respectively) in the two sibings. The younger underwent bilateral orchidectomy, and the histology showed normal testicular tissues. The same homozygous mutations (c.1459_1467delGACTCTTTC(p.Asp487LysfsX20)) in *CYP17A1* gene were identified in both patients(Fig. 1).

CONCLUSIONS

We confirmed the diagnosis of 17α-hydroxylase deficiency in these two siblings.

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Gonads

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