Homozygous \textit{CYP17A1} Mutation Identified in a Chinese Family with 46, XX and 46, XY 17α-Hydroxylase Deficiency

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\textbf{OBJECTIVES}

Background: Congenital adrenal hyperplasia due to 17α-hydroxylase deficiency is a rare autosomal recessive disorder, characterized by sexual infantilism, amenorrhea, hypertension and hypokalemia, which is caused by \textit{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.

\textbf{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 \textit{CYP17A1} gene was performed.

\textbf{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/mL per h respectively). Pelvic ultrasonography revealed a pre-pubertal uterus in the older, and absence of ovaries and uterus in addition to a blind-ending 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 siblings. The younger underwent bilateral orchidectomy, and the histology showed normal testicular tissues. The same homozygous mutations (c.1459_1467delGACTCTTTTC (p.Asp487LysfsX20)) in \textit{CYP17A1} gene were identified in both patients (Fig. 1).

\textbf{CONCLUSIONS}

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

\textbf{References}