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17-ALPHA-HYDROXYLASE DEFICIENCY AND MITOCHONDRIAL VARIANT A8343G

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Purpose 17 α -Hydroxylase deficiency (17OHD) is a rare type of secondary hypertension and well-known as a group of autosomal recessive disorders of adrenal steroidogenesis caused by a genetic disorder in one of the steroidogenic enzymes. At the same time, the knowledge of mitochondrial genetic basis for 17OHD is limited.

Methods Clinical information, endocrine examination, computerised tomography (CT) scanning and mitochondrial genetic analysis were performed in one three-generation Han Chinese family with maternally transmitted hypertension.

Results A 13-year-old female proband with genotypic 46, XY suffered from 17OHD (male pseudohermaphroditism) was detected. She presented with hypertension, primary amenorrhea, and lack of secondary sexual characteristics. Laboratory tests showed hypokalaemia, low levels of androgens (testosterone and dehydroepiandrosterone), corticosteroid, high levels of adrenocorticotrophic hormone and progesterone. CT scanning found bilateral adrenal hyperplasia, womb and ovaries deficiency. The other matrilineal relative in this family exhibited the variable degree of hypertension and hypokalaemia. Full sequence analysis found mitochondrial variant A8343G (belong to haplogroup H) located at the position of A54 at the T Ψ C arm of tRNA^{Lys} gene in all matrilineal pedigrees, not in 270 controls. The variant A8343G may modulate mitochondrial K⁺ transport, lead to decrease of tRNA metabolism, increase of angiotension II level, enhance of mtROS generation and influence the transcription of tRNA herein affect the steady-level of protein synthesis.

Conclusions The findings suggested that A8343G in mitochondrial tRNA^{Lys} gene may have biologic plausibility to implicate in the pathogenesis of 17OD in the three-generation Han Chinese family.