



POSTER PRESENTATION

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Phenotype of patients with congenital adrenal hyperplasia due to 11 β -hydroxylase deficiency

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Congenital adrenal hyperplasia (CAH) is one of the most common metabolic diseases. It is caused by a severe or partial impairment of adrenal steroidogenesis affecting cortisol biosynthesis. Approximately 5–8% of all cases are due to steroid 11 β -hydroxylase deficiency (11OHD; OMIM +202010), which occurs in approximately 1:100,000 to 1:200,000 live births in non consanguineous populations. Mutations in the CYP11B1 gene, causing 11 β -hydroxylase deficiency in the zona fasciculata in the adrenal cortex, have been identified. Our aim is to describe clinical and biochemical features in patients with CAH due to 11 β -hydroxylase deficiency. The case series report included 9 patients (6 male and 3 female) from 7 unrelated families who was identified novel and/or reported homozygous or compound heterozygous mutations in CYP11B1 gene. Diagnosed age was from 2 to 11 years old. All three female cases presented with ambiguous genitalia at birth. Other clinical features were hypertension (6/7 cases); hyperpigmentation (5/7 cases); pseudo-precocious puberty (male) (5/5 cases). Hypokalemia was noted in 3/7 cases. Three cases need antihypertensive drug associated with hydrocortisone replacement therapy. In conclusions, the clinical hallmark of 11 β hydroxylase deficiency is variable and virilization and hypertension are the prominent clinical features of 11 β hydroxylase deficiency. Biochemical identification of elevated precursor metabolites is not usually available and mutation analysis of CYP11B1 will held confirmation of diagnosis.

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