中文題目：先天性腎上腺增生症合併類固醇 17-羥酵素缺乏以不典型高醛固酮血症為表現

英文題目：Atypical Presentation of Hyperaldosteronism in Congenital Adrenal Hyperplasia with 17 Alpha-Hydroxylase Deficiency

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Abstract: Congenital adrenal hyperplasia (CAH) is a rare and a group of disorders, characterized by genetic defect of autosomal recessive inheritance and impaired cortisol production. Compared with 21-hydroxylase deficiency, 17-alpha hydroxylase deficiency (17OHD) is even rare, about 1% of all CAH cases. Hypoaldosteronism, due to low renin suppressed by deoxycortisosterone(DOC) and corticosterone, is one of the hallmarks of 17OHD. Here we presented a case of 17OHD, confirmed by endocrine profiles and genetic testing, initially misdiagnosed as adrenal tumor with hyperaldosteronism. The serum potassium and blood pressure were controlled by cortisone 25mg daily. 17OHD could relate to hypo-, normo- or hyperaldosteronism. This case demonstrates that the presence of hyperaldosteronism does not exclude the differential diagnosis of 17OHD.