Diagnosis and Management of Congenital Adrenal Hyperplasia: 20-Year Experience in Songklanagarind Hospital

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Abstract

Objective: To describe the types of congenital adrenal hyperplasia (CAH) in Thai children, and the clinical and laboratory findings associated with each type.

Material and Method: The medical records of CAH patients, aged 0 to 15 years, were retrospectively reviewed.

Results: During the 20-year period (1991-2011), there were 66 CAH patients. The most common type was salt-wasting 21-hydroxylase deficiency (21-OHDSW) (59.1%), followed by simple virilizing 21-hydroxylase deficiency (21-OHDSV) (30.3%), 20, 22-desmolase/steroidogenic acute regulatory protein (20,22D/StAR) deficiency (6.1%), and 3β-hydroxysteroid dehydrogenase (3β-HSD) deficiency (4.5%). The stimulated cortisol level was <18 µg/dL in most patients, with 20 cases of 21-OHDSV having a median level of 19.49 µg/dL. The median basal 17-hydroxyprogesterone (17-OHP) levels were markedly elevated in 21-OHDSW and 21-OHDSV patients (20,264 and 5,985 ng/dL, respectively), but was very low in 20, 22D/StAR deficiency patients (260 ng/dL). Bilateral adrenal enlargement, demonstrated by radioimaging, was helpful for diagnosis of 20,22D/StAR and 3β-HSD deficiency.

Conclusion: The most common type of CAH in our population was 21-hydroxylase deficiency. The steroid profile of cortisol, 17-OHP, and testosterone levels is helpful for clinical diagnosis of 21-OH deficiency. The low 17-hydroxyprogesterone and low cortisol levels in phenotypic female infant with salt-wasting crisis suggests the diagnosis of 20, 22D/StAR deficiency CAH.

Keywords: Adrenal crisis, Adrenal insufficiency, Ambiguous genitalia, Congenital adrenal hyperplasia, Precocious pseudopuberty

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