CONGENITAL ADRENAL HYPERPLASIA: 20-YEAR EXPERIENCE IN SOUTHERN THAILAND

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Background: Congenital adrenal hyperplasia (CAH) is a group of inherited disorders resulting deficiency of enzymes in the synthesis of cortisol. The clinical symptoms vary depending upon the specific enzymatic deficiency.

Aim: To depict the clinical features, and laboratory and hormonal studies of CAH patients.

Subjects and method: The medical records of pediatric patients diagnosed with CAH at Songklanagarind Hospital during 1991-2010 were reviewed.

Results: There were 61 CAH patients. The most common type was 21-hydroxylase deficiency, salt-wasting type (21-OH, SW) (57.4%), followed by 21-hydroxylase deficiency, simple virilizing type (21-OH, SV) (31.1%). All patients with 20, 22-desmolase deficiency (n=4), 21-OH, SW (n=35) and 3 β -hydroxysteroid dehydrogenase deficiency (n=3) presented with dehydration at the median age of 1.8 months. Genital ambiguity was found in 53% of girls with 21-OH, SW. Nineteen patients with 21-OH, SV presented with enlarged clitoris/penis at the median age of 5 years. The median cortisol level after ACTH stimulation was < 18 μ g/dL in all types, except 19 patients with 21-OH, SV, in which the median cortisol was 19.49 μ g/dL. The basal 17-OHP level of these 19 patients was markedly elevated (5,985 ng/dL). All patients, except one, were assigned correctly as a boy or girl at the initial presentation.

Conclusions: The most common type of CAH in our institute was 21-hydroxylase deficiency. The most common clinical manifestation in the newborn period is dehydration, and in the childhood period is the enlarged clitoris/penis. The most useful investigation for definite diagnosis of 21-hydroxylase deficiency is basal 17-OHP level.