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Title: CONGENITAL ADRENAL HYPERPLASIA: EXPERIENCE OF A PEDIATRIC ENDOCRINOLOGY UNIT
Background: Congenital adrenal hyperplasia (CAH) is caused by inborn errors of steroidogenesis in which cortisol is not sufficiently produced by the adrenal cortex. CAH due to 21-hydroxylase deficiency accounts for about 90% of diagnosed cases of CAH. In classic 21-hydroxylase deficiency prenatal exposure to excess androgens results in virilization of the female fetus. Seventy five per cent of classic 21-hydroxylase deficiency cases do not effectively synthesize aldosterone and are salt-wasting, a condition that is potentially fatal.
Nonclassic 21-hydroxylase deficiency, is associated with a milder enzymatic defect. This condition is rarely recognized in infants, but rather is a potential cause of premature adrenarche and pubarche in children, virilization in young women, and variable symptoms in young men. The treatment of 21-hydroxylase deficiency is lifelong hormonal replacement.
Aim: To describe and evaluate clinical presentation, growth and metabolic control in a group of patients with CAH.
Patients and methods: A retrospective study was performed by consulting clinical records of patients with CAH followed in the Pediatric Endocrinology Unit from 1999 to 2008. This resulted in a sample of 7 patients.
Results: Six patients had 21-hidroxylase deficiency and one patient had 11-hidroxylase deficiency. The age range was from 15 days to 13 years. There was only one case of classic CAH, a newborn girl with clitoris enlargement. None of the patients presented with salt-wasting crisis. The other six children presented with premature pubarche, precocious puberty and hirsutism. 17-OH-Progesterone was high in all the cases and in one patient there was a high level of 11-deoxycorticosterone. Advanced bone age was found in 71%. Six children were treated with hydrocortisone. Follow-up was based on growth, bone age, puberty and monitoring serum metabolites. Outcome was good, with normalization of bone age and suitable growth.
Conclusion: Non-classic 21-hydroxilase deficiency was the most common form of CAH in our cases review study, which was expected due to our mediterranean background. There was one case of 11-hydroxilase deficiency, a much more rare condition.
With carefully supervised medical treatment, congenital adrenal hyperplasia patients have the capacity for normal puberty and growth.