

## **PROFILE OF TURKISH CHILDREN WITH CONGENITAL ADRENAL HYPERPLASIA: A HOSPITAL STUDY**

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Congenital adrenal hyperplasia (CAH) is the most common cause of genital ambiguity in the newborn. In this study the clinical, laboratory and epidemiological characteristics of CAH were evaluated from the records of 46 patients managed in our clinic over a 10-year period. 21-hydroxylase deficiency (21OHD) was presented in 30 (91.3%), salt wasting form in 17 (13F, 4 M), simple virilizing form in 13 (10F, 3M), and late onset form in 12 girls. 11b-hydroxylase deficiency was diagnosed in 3 patients (6.5%, 3M). One male had 3b-hydroxysteroid dehydrogenase deficiency (2.2%). Age of diagnosis was 2.1±3.7 years (1 day-13 years) in patients with classical CAH. Of 34 patients with classic CAH, only 14 (41%) were diagnosed during the neonatal period and 11 were diagnosed after two years old. The karyotype was 46, XX in a total of 23 (68%) patients (7 of the 46,XX patients reared as boys) and 46,XY in 11 with classical CAH. The most prevalent presenting complaint was ambiguous genitalia (n:20). Family history of CAH was present in 8 families (11 patients) and consanguinity was present in 17 patients (37%). Feminizing genitoplasty was performed in 14 females and virilizing genitoplasty in 3 patients. Our short-term results of surgery to construct either female or male genitalia are good, but, long-term surgical and psychosexual functioning of these patients remain to be determined.

Parental consanguinity rate among families of the patients was higher than the general population in our region. A significant number of the patients with CAH still diagnose after neonatal period. Prompt diagnosis and treatment could minimize the detrimental effects of CAH.

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