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Titel des Beitrags: Growth Pattern of Untreated Boys with Simple Virilizing Congenital Adrenal Hyperplasia Indicates Relative Androgen Insensitivity during the First Six Months of Life.

Abstract: Context: Mild forms of simple virilizing congenital adrenal hyperplasia (CAH) may be missed in newborn screening. In the pre-newborn-screening era, missed diagnosis of simple virilizing CAH was not infrequent in boys. Elevated adrenal androgens lead to accelerated growth and bone maturation. Traditional treatment of CAH consists of the suppression of ACTH through glucocorticoid replacement, in an attempt to reduce excessive androgen production. Objective: To retrospectively analyze early growth pattern and bone maturation in untreated boys with simple virilizing CAH. Patients: In the pre-newborn screening era, 13 boys had a late diagnosis of simple virilizing classical CAH. Diagnosis of 21-hydroxylase deficiency was confirmed by mutation analysis of the CYP21A2 gene in all patients. Growth data were retrospectively collected from standardized preventive medical checkups at the regular pediatrician until the time of diagnosis of CAH. Results: Length was 0.1 ± 0.8 SDS (mean ± SD) at birth, 0.2 ± 1 SDS at 3 months, 0.2 ± 0.9 SDS at 6 months, 0.7 ± 1 SDS at 1 year, +1.1 ± 0.9 SDS at 2 years and +1.8 ± 1.2 SDS at 4 years. At diagnosis, mean chronological age was 4.4 ± 1.6 years and height SDS was 2 ± 1.7. Bone age was accelerated (9.4 ± 4 years) at diagnosis. Signs that had led to diagnosis were pubic hair (n = 11), accelerated growth rate (n = 6) and birth of an affected sister (n = 3).
Despite late start of hydrocortisone treatment, mean final height was -1 ± 0.9 SDS. Seven of 18 patients had a final height within 1 SD of target height. Conclusion: Height velocity is not markedly increased in untreated boys with simple virilizing CAH in the first 6 months of life, indicating that infants are relatively androgen insensitive during that period. After the first 6 months of life, growth velocity increases significantly and elevated androgens lead to advanced skeletal maturation. This observation has implications for lower hydrocortisone doses to be used in CAH children during the first 6 months of life. In addition, staying alert for clinical symptoms and signs of simple virilizing CAH is still warranted, since mild forms may be missed in newborn screening.

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