LONG TERM OUTCOME OF PATIENTS WITH CONGENITAL ADRENAL HYPERPLASIA, 21 HYDROXYLASE DEFICIENCY

Introduction

Congenital adrenal hyperplasia (CAH) comprises of a group of inherited autosomal disorders affecting the steroid synthesis. The most common type is 21 hydroxylase deficiency. Treatment of CAH mainly aims at maintaining the normal growth velocity and attaining normal puberty. Accumulation of steroid precursors, occasional occurrence of precocious puberty can alter the normal growth and pubertal patterns. The control of the disease is often difficult and they may need supraphysiological doses of steroids which may lead to complications.

Aims & Objectives

The aim of this study is to determine the prevalence of short stature and other growth disorders, the prevalence of obesity, hypertension and metabolic complications in adolescents with CAH, 21 hydroxylase deficiency and also to assess the effect of glucocorticoid dosing on growth, obesity, blood pressure and metabolic profile.

Materials and Methods

This study was conducted in the outpatient department of Child health I, Christian Medical College (CMC), Vellore. All adolescent children between 10 – 22 years of age diagnosed with CAH 21 hydroxylase deficiency in CMC and on regular follow up till adolescence attending the outpatient department from February 2016- September 2016 were included in the study. Data regarding demography, treatment details, height and body mass index were collected. All patients underwent biochemical testing for metabolic profile like lipid profile, alanine aminotransferase, fasting glucose and fasting insulin. 17 hydroxy progesterone (17 OHP) levels done at regular intervals were collected from previous records, for analyzing the adequacy of the treatment. The dosage of hydrocortisone per body surface area was collected from the patient’s chart since the time of diagnosis.

Results

Twenty six patients were included in the study of which 16 were females (62%) and 10 were males (38%). Salt wasting was the most common type seen among the patients with 65% (n=17) of the total study population. Around 23% (n=6) of patients had Short stature as defined by height SDs <-2. Obesity and overweight was found in 39% (n=10) and 15% (n=4) respectively. Abnormal lipid profiles were seen in majority of the children with CAH (Hypercholesterolemia in 46%, Abnormal HDL in 35%, abnormal LDL in 31%) and insulin resistance was seen in two patients. Short stature and obesity was more prevalent in children with CAH compared to the general population.

The risk of short stature was more when the disease control is not adequate in adolescent age group. During the adolescent period, 28% (n=5) had short stature in the uncontrolled disease activity group compared to 20% (n=1) in the group with suppressed 17 OHP level. The risk of obesity is more when the disease control is not adequate both during childhood and adolescence. 61% of patients with elevated 17 OHP levels during childhood and 62% of patients with elevated 17 OHP levels during adolescence had overweight and obesity indicating that obesity are more due to poor disease control.

Metabolic abnormalities like dyslipidemia were more prevalent when the 17 OHP values were unsuppressed during both the age periods but more during the adolescent period. Higher Steroid dosing as defined by dose >15mg/m2/day demonstrated no significant effect on short stature and body mass index. Transient hypertension was seen in two patients receiving fludrocortisone. Insulin resistance was seen in two patients in our study population and both the patients had received higher dose of glucocorticoids.

Conclusion

We conclude that adequate suppression of adrenal androgens as measured by 17 OHP levels is important during both the childhood and adolescent periods to attain normal adult height and normal BMI. Unsuppressed 17 OHP values during both the childhood and adolescent age group was also a major risk for obesity and overweight. Metabolic

abnormalities like dyslipidemia were more deranged when the 17 OHP values were unsuppressed, more during the adolescent period. Higher Steroid dosing demonstrated no significant effect on stature, body mass index and metabolic abnormalities. We conclude that dose of 10-15mg/m2/day of hydrocortisone seemed to be safer.

Keywords: Congenital adrenal hyperplasia, final height, obesity, metabolic profile, steroid dosing.