Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY: ADRENAL, THYROID, AND GENETIC DISORDERS

Growth Hormone Therapy for Children With Duchenne Muscular Dystrophy and Glucocorticoid Induced Short Stature

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Background: Duchenne Muscular Dystrophy (DMD) is the most common form of inherited muscle disease in childhood. DMD patients have severe growth retardation due to several reasons including chronic treatment with glucocorticoid (GC). Data regarding the efficacy and safety of Recombinant Growth Hormone (rGH) treatment in DMD patients is very limited. The aim of this study is to evaluate efficacy and safety of growth hormone treatment in 4 DMD boys with glucocorticoid induced growth failure. **Methods and Results:** 4 prepubertal patients with DMD on high dose of GC (Deflazecort or Prednisone) at an age range between 12-13.6 years and significantly delayed bone age (8.5-11 years) were studied. rhGH was subcutaneously administered at a dose of 0.033 mg/kg/d for a duration of 12-18 months. Pretreatment annual growth rate of the 4 patients (0, 3,2.5, 1.5 cm/yr) improved during treatment (3.8, 7.8, 7.2, 3.3 cm/yr respectively). While height SD improved in 2 patients: -2.74 and -2.96 height SD to -2.52 and -2.64 height SD on rGH therapy, In the other two patients pretreatment continuous decline in height SD was arrested. Motor function decline was similar pre-growth hormone and during treatment. Cardiopulmonary function measured by ejection fraction and forced vital capacity was unchanged during the treatment period. **Conclusions:** This report of growth hormone therapy in DMD patients revealed an improved prepubertal growth velocity without detrimental effects observed on neuromuscular and cardiopulmonary function. Larger randomized control studies are required to prove safety and efficacy of this treatment in DMD patients.

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Growth Patterns in Children With Multiple Endocrine Neoplasia Type 2B: Small Stature in Childhood

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Background: Multiple Endocrine Neoplasia 2B (MEN2B) is characterized by medullary thyroid carcinoma (MTC) before the age of one, pheochromocytoma and several non-endocrine manifestations. Marfanoid habitus is considered to be an important related feature, leading to the assumption that children with MEN2B have a tall stature. However, very little is known about actual growth patterns in children with MEN2B and its implication for final height.
Aim: To describe the growth patterns during childhood and adolescence and relate final height to target height (TH) in MEN2B patients.
Methods: Growth during childhood was investigated in eight patients with MEN2B under care in a Dutch MEN expertise center. Growth charts were assessed in relation to parental height, age at diagnosis and at thyroidectomy, body mass index (BMI), pubertal development and extensiveness of disease manifestations.
Results: Four out of eight patients showed longitudinal growth below their TH range. Three others showed prepubertal growth in the lowest margin of their TH range. Small stature was accompanied by delayed bone maturation. Arm span to height ratio was not increased in any of the (six) patients studied. All four patients who reached adulthood attained final height within their TH range, despite small stature during childhood. Small stature in childhood was not associated with age at diagnosis, age at thyroidectomy, extensiveness of MTC, BMI or endocrine deficiencies.
Conclusions: Children with MEN2B did not present with marfanoid features regarding height or arm span. In contrary, short stature may be prevalent, with longitudinal growth beneath the individual TH range. Nevertheless, a normal final height within the TH range may well be reached. Growth patterns seem to be independent of both age at diagnosis and thyroidectomy as well as disease severity.

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Growth-Related Characteristics of Patients <18 Years of Age with Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency (21OHD): Real World Evidence from the I-CAH Registry

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Background: Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency (21OHD) is a rare, autosomal recessive disease of the adrenal cortex leading to a lack of cortisol production and compensatory ACTH secretion, which drives excess androgen production. The chronic exposure to excess androgen, coupled with supraphysiologic glucocorticoid doses, can lead to advanced skeletal maturation with reduced growth in puberty, premature epiphyseal