CHAPTER 36

MEDICAL SCIENCES
PADIATRICS

Doctoral Theses

01. SINGH(Pratiksha)
Study of Biochemical Profile and Polymorphisms of the Growth Hormone Receptor Gene in Patients with Idiopathic Short Stature and Proven Growth Hormone Deficiency.
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Abstract
(Not Verified)

Objective: To evaluate the clinical and biochemical profile and Growth hormone receptor Polymorphism of children presenting with short stature to a tertiary care centre, and also to study the molecular players/polymorphism that might give us an insight into good response to a relatively expensive modality of treatment. This would thus be directing resources to patients with GH deficiency who would respond best to it. Result: A total of 473 children with a median age of 3.65 years (Range 2-18 yrs) were enrolled. 23% of the children each were diagnosed as Growth hormone deficient and Idiopathic short stature. Celiac disease also contributed significantly in 18% of cases. The other causes seen were skeletal dysplasia (7%), syndromic (12%) and malnutrition (2%). Amongst children with endocrine disordisorders, 40% children had hypothyroidism, panhypopituitarism was seen in 10% children and 50% had Laron's syndrome. In Children with chronic disorders, 72% were diagnosed with Thalassemia, 21% with chronic kidney disease and 1 child had renal tubular acidosis. Constitutional and familial short stature were seen in 6% and 2% children respectively. Amongst patients with GHD, 60.7% had wild type (GHRfl/fl), 19.2% were heterozygous (GHRfl/GHRd3) and 20.1% were homozygous (GHRd3/d3), whereas for idiopathic short stature they were 67.5%, 14.5% and 18% respectively. Conclusion: With high index of suspicion, availability of testing and following an algorithmic approach, diagnosis could be attained in 85% of cases. Our data indicates the changes in profile from those rereported earlier in our country. Growth hormone deficiency and celiac disease contribute sigsignificantly even though majority are normal variants. Also, genotyping done would help in prediction of response to recombinant GH therapy in a resource constraint resulting in appropriation of finances which could be utilized for a higher priority area.

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