

Case Reports

Idiopathic Short Stature - A Case Report

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Abstract:

Short stature in childhood is the most common reason for referral to pediatric endocrinologists. Idiopathic short stature is defined as ≥ 2 SD below the corresponding mean height for a given age, sex, and population in a child with a normal birth size and normal body proportions and without evidence of any systemic, endocrine, nutritional, or chromosomal abnormalities. ISS children have normal GH responses to stimulation test and GH therapy has approved for the treatment of children with ISS.

A six-year-old girl presented with not growing well in comparison to other peers since 6 months of her age. Her WAZ was -4.5 SD, HAZ was -7.7 SD, and projected height is far below parental target height. Bone age was 2-3 year and growth hormone stimulation showed normal growth hormone secretion. Here we reported a six-year-old girl diagnosed as a case of non-familial ISS and treated with growth hormone. Diagnosis of Idiopathic short stature was done by exclusion of other causes of short stature and growth hormone therapy was indicated.

Keywords: Short stature, Idiopathic short stature (ISS), Growth hormone, Growth.

Background

Short stature is defined as a height more than two standard deviations below the mean for age, or less than the 3rd percentile. Most children with short stature have normal variants such as familial short stature, constitutional delay of growth and puberty, or idiopathic short stature (ISS). Approximately 5% of children referred for evaluation of short stature have an identifiable pathologic cause.¹ Idiopathic short stature is defined as the presence of a height >2 SD below the corresponding mean height for a given age, sex, and population in a child and without evidence of any systemic, endocrine, nutritional, or chromosomal abnormalities.² ISS children may be familial or non-familial. In familial ISS, height for age below the 3rd centile but within the parental target range and a family history of short stature maybe present. In non-familial ISS, height for age below the 3rd centile and also below the parental target range. A delayed skeletal maturation and pubertal delay maybe present.²

Recombinant human growth hormone therapy is approved in ISS to achieve normal adult height and higher doses than a replacement dose in growth hormone deficiency have been required as sensitivity to growth hormone is impaired in patients with ISS.³ In 2003, the United States Food and Drug Administration (FDA) approved rhGH for the treatment of ISS.⁴

Case presentation

A six-year-old girl presented with not growing well in comparison to other peers since 6 months of her age. Her birth weight was normal and growth during first six months was appropriate. On examination, she had frontal bossing, depressed nasal bridge, shallow orbit, micrognathia and small hands and feet (Fig1). She had no family history of short stature or delayed puberty. Her WAZ was -4.5 SD, HAZ was -7.7 SD, projected height was far below target height and upper segment and lower segment ratio was 1.25:1 (Proportionate short stature). Systemic examination showed no abnormality. Investigation revealed normal renal function, liver function, thyroid function and karyotype. Her bone age was 2-3 year (Fig 2). Growth hormone stimulation test by levodopa on day one showed growth hormone level at '0' min 3.78ng/ml, at '30' min 14.40 ng/ml and at '90' min 7.69ng/ml and by

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clonidine on day two showed growth hormone level at '0' min 2.99ng/ml, at '60' min 40.00ng/ml and at '90' min 16.60 ng/ml suggestive of normal growth hormone. We diagnosed the case as non-familial Idiopathic short stature and treated with recombinant human growth hormone 0.4mg/kg/week and the patient responded to treatment. Her height increment was 5.5 cm after 6 months of treatment. Growth hormone resistant was excluded as the patient responded to treatment with growth hormone.



Fig 1: The girl has frontal bossing, depressed nasal bridge, shallow orbit and micrognathia.



Fig 2: Bone age of the patient was 2-3 year

Discussion

Idiopathic short stature is known as normal-variant short stature or short stature of undefined cause.⁵ The patient was presented with short stature and her birth weight was normal and growth normally till six month of age. Her height for age was below 3rd centile and proportionate. Her target height was below parental target height. Other causes of short stature were excluded and we categorized the child as a non-familial idiopathic short stature. For diagnosis of ISS systemic diseases, growth hormone deficiency, intrauterine growth retardation, genetic or syndromic causes of short stature, psychosocial deprivation should be excluded.⁶

The patient was treated with recombinant growth hormone and responded to treatment with higher dose than dose required in growth hormone deficiency. A meta-analysis showed growth hormone therapy is effective in children with idiopathic short stature.⁷ Higher doses of growth hormone may confer a greater benefit.⁸

Conclusion

Idiopathic short stature is diagnosed when no cause of short stature was identified and responded to treatment with high dose of Recombinant human growth hormone.

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