TITLE

Favourable growth hormone treatment response in a young boy with achondroplasia

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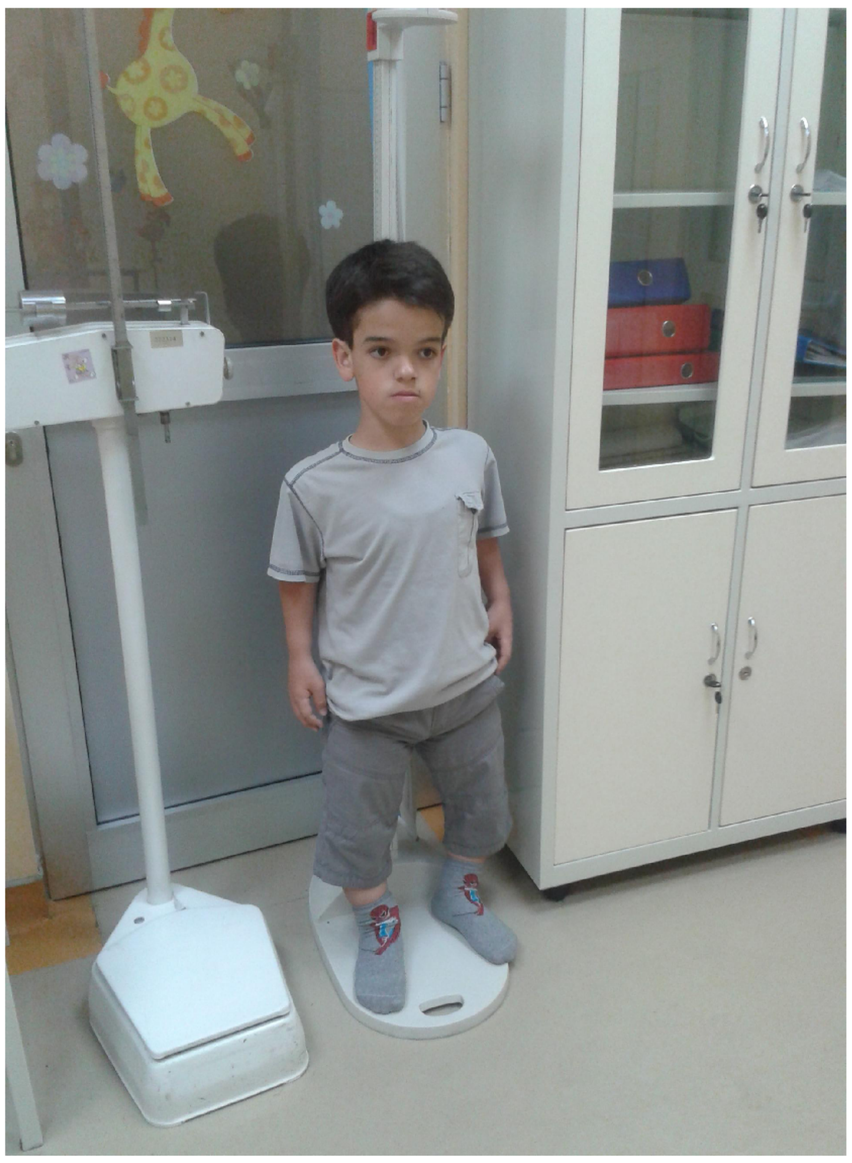
OBJECTIVES

Achondroplasia is a skeletal dysplasia, being the most common cause of rhizomelic dwarfism.



METHODS

We present a ten year old boy who was first diagnosed prenatally. He had a mutation c1138G>A in the gene FGFR3 in a heterozygotic constellation. His IgF1 levels and IgFBP3 were normal. Two stimulation tests for growth hormone were performed with normal levels of the hormone. His psychomotor development was adequate for his age except for speech difficulty. He started with recombinant hGH (r-hGH) at the age of 3.4 years in a dose of 0.06 mg/kg. His mean Height SDS (HtSDS) was -2.2. The growth increased to 10 cm/year in the first year of therapy (HtSDS -1.1). It decreased during the second year to 4 cm (HtSDS -1.7) and again increased during the third year to 8 cm/year (HtSDS - 1.3). In the next years the growth was constant (6.5, 2.3, 3.5 cm / year). He is still growing in the 3rd percentile of the growth curve (HtSDS – 1.2). The body disproportion did not aggravate during treatment.

CONCLUSIONS

The growth was satisfactory in the first 4 years of treatment, although he still continued to grow. The young age at the start of treatment was also of importance. Our other patients with achondroplasia who started treatment older had a poor response to growth hormone.

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