Case Report

**A CASE OF GOLDENHAR SYNDROME ASSOCIATED
WITH GROWTH HORMONE DEFICIENCY**

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Summary We have experienced the case of a 10-year-old boy who had Goldenhar syndrome accompanied by growth hormone (GH) deficiency. His height increased after treatment with growth hormone was administered. We found no untoward effects of the hormone and we consider that treatment with GH is useful for patients who present with Goldenhar syndrome associated with growth hormone deficiency.

Key Words Goldenhar syndrome, growth hormone treatment

INTRODUCTION

Goldenhar syndrome is characterized by microtia, mandibular hypoplasia, vertebral anomalies, and epibulbar dermoids (Goldenhar, 1952). However, to our knowledge there have been no reports concerning endocrinological abnormalities in Goldenhar syndrome. Here, we report the case of a boy with Goldenhar syndrome who also presented GH deficiency. We treated him with GH.

CASE REPORT

A ten-year-old boy was admitted to our hospital because of short stature and convulsions. He was born to healthy unrelated parents after a 38-week-uncomplicated term pregnancy and normal delivery. His apgar score was 9 points at 1 min. His birth weight was 2,290 g and he measured 48 cm. He had been followed up at another hospital because of growth retardation and facial anomalies. He often showed generalized symmetrical tonic seizure from the age of 5 and had been treated with an anticonvulsant. He was referred to our hospital for the treatment of short stature and convulsions.

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Physical examination on admission showed left accessory auricle, microtia, left mandibular hypoplasia, left epibulbar dermoids and no mental retardation. Based on these findings he was having Goldenhar syndrome. His height was 100.8 cm (-6.5 S.D.) and his body weight was 16 kg (-2.5 S.D.). His father's height was 165 cm and his mother's height was 155 cm. His only older brother was not short. None of the members of his family presented any of the abnormalities of the Goldenhar syndrome. His bone age was 4 years and 7 months. His thyroid and adrenal function were normal. His GH response to L-dopa and glucagon with propranolol was blunted (the highest level was 1.2 and 2.7 ng/ml, respectively: normal level >7 ng/ml). His thyroid-stimulating hormone response to thyrotropin-releasing hormone as well as luteinizing hormone and follicle-stimulating hormone responses to luteinizing hormone-releasing hormone were all within normal range. Thus, based on the results of the endocrinologic examinations he was also diagnosed as having idiopathic GH deficiency.

Fig. 1. Growth of a boy with Goldenhar syndrome (●) in comparison with normal standards (gray shading). The arrow indicates the start of GH treatment.

We began to treat him with sodium valproate (400 mg/day) for the convulsions and anabolic steroids (1 mg/day) for short stature, because we could not immediately use GH. We began to treat him with GH (0.1 u/kg/wk) in addition to anabolic steroids when he was 12 years and 3 months old and continued the therapy for convulsions. His growth increased after GH treatment (Fig. 1). Signs of puberty were noted around the time he became 16 years old.

Now he is 23 years old but his bone age is delayed (16 years old). He is still under GH therapy and his height continues increasing. No untoward effects have been noted.

DISCUSSION

Goldenhar syndrome is an association of facial and auricular anomalies resulting from errors in the morphogenesis of the first and second branchial arches. This syndrome is also associated with pulmonary and airway-related malformations (Gorlin *et al.*, 1963; Booth and Berry, 1975; Kenawi and Dickson, 1976; Bowen and Parry, 1980; Mendelberg *et al.*, 1985; Scholtes *et al.*, 1987; Smith, 1988; Downing and Kilbridge, 1991) but, in general it is not associated with growth retardation. To our knowledge there are no reports in the literature regarding its association with endocrinological abnormalities.

Turner syndrome and septo-optic dysplasia are characterized by short stature and are often associated with GH deficiency. Such patients are effectively treated with GH (Arslanian *et al.*, 1984; Price *et al.*, 1990; Rongen *et al.*, 1990; Massa *et al.*, 1991; Job and Landier, 1991). But in other syndromes associated with growth retardation, it is not known whether GH therapy is effective or not. Recently, Horton *et al.* (1992) reported that growth-promoting therapy with recombinant human GH for achondroplasia is safe and effective.

Our patient was short due to GH deficiency; thus we treated him with GH. He used to grow in height at the rate of 3 cm/year before GH being administered and after treatment with GH his height increased at a rate of 9 cm/year. His pubertal stage did not change after the GH treatment; therefore we did not change to other modes of treatments. Between the age of 18 and 20 years, his weight increased at a high rate due to unknown factors, but this increase in body weight was not considered as an adverse effect of GH because the other physical parameters did not change and laboratory data were normal.

In conclusion we consider that GH therapy was effective in this boy with Goldenhar syndrome associated with GH deficiency. To our knowledge, this is the first case in the world of Goldenhar syndrome associated with GH deficiency in whom GH therapy was attempted and it proved effective.

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