A Boy with Nutritional Growth Retardation due to Achalasia

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Abstract. A 6-yr-old boy was seen at our hospital due to short stature (–2.0 SD) and frequent vomiting. Peak GH response to arginine and insulin was >10 ng/ml at 10.5 yr of age. Growth retardation was progressive. At 12 yr of age, he was still in prepubertal status with a bone age of 8. His height and body weight were 121.6 cm (–3.57 SD) and 19.0 kg (–2.05 SD). Serum level of IGF-1 was low (35 ng/ml), with marginal hypothyroidism due to non-thyroidal illness (T3 0.93 ng/ml, T4 6.7 µg/dl and TSH 0.08 µU/ml). Serum levels of prealbumin and retinol binding protein were also decreased. Achalasia was diagnosed by esophagography. At the age of 12 yr and 6 mo he underwent laparoscopic extramucosal myectomy with fundoplication, and subsequently his nutritional status improved dramatically. At 15.5 yr old, he had gained height and body weight, 153.2 cm (–2.61 SD) and 38.9 kg (–1.95 SD), respectively, and sexual development had advanced too. The clinical course of this boy revealed that achalasia should be added to the list of causes of nutritional growth retardation in children.

Key words: growth retardation, achalasia, malnutrition, delayed adolescence

Introduction

Achalasia is characterized by a generalized derangement of motility with absence of propulsive peristalsis in the body of the esophagus, failure of the gastroesophageal sphincter to relax in response to swallowing and esophageal hypersensitivity to cholinergic drugs (1). The disease is caused by degeneration and reduction in the number of ganglion cells in Auerbach’s plexus with patchy fibrosis and scarring in the esophageal musculature (1). Achalasia is extremely rare in children. In such cases, the disease is progressive and difficult to diagnose because of ambiguous symptoms.

Case Report

We report the case of a 15-yr-old boy. His records showed a choreoathetosis-like involuntary movement under 1 yr of age, and frequent vomiting since 3 yr of age. He was significantly short, SD of height was –2.0 at 6 yr of age, and his height was 116.8 cm (–3.7 SD) and body weight was 17.5 kg (–2.2 SD) at 10 yr and 6 mo of age. GH stimulation tests were performed. The peak GH responses to arginine (10.9 ng/ml) and insulin (11.3 ng/ml) were normal at that time. Response to the LHRH loading test was from <0.5 mU/ml (normal range; NL, 0–0.4) to 6.1 mU/ml (NL, 0.4–6.0) for LH, and from 1.9 mU/ml (NL, 0.6–3.0) to
13.0 mU/ml (NL, 6.3–15.6) for FSH, and the boy was considered to be still in prepubertal status. TSH response to the TRH loading test was from 0.34 µU/ml to 4.3 µU/ml, and ACTH response to insulin-induced hypoglycemia was from 11.1 pg/ml to 58.9 pg/ml. Mean GH concentration during sleep was 8.6 ng/ml (NL, >5 ng/ml) and urinary GH excretion was 25 ng/g.cre (NL, 5.6–24.5), whereas the serum level of IGF-1 was very low for his age, 44.0 ng/ml (NL, 127–424 ng/ml).

Dysphagia, vomiting and involuntary movement persisted, and we also noted a reduction in tears and sweating, as judged from the results of the Schirmer and pilocarpine tests. Salivary secretion was assessed by the Gum test and was normal. His height was 121.6 cm (−3.6 SD) and his body weight was 19.0 kg (−2.4 SD) at 12 yr of age with normal appearance. Penis and pubic hair were still in Tanner 1° and testis volume was 3 ml. Neurological findings were grossly normal including muscle power and intellectual function. Magnetic resonance imaging of the brain was normal but EEG revealed generalized spike and wave complexes with increased fast activity. Biochemical indices of nutrition such as albumin, 3.8 g/dl (NL, 4.0–5.0), total cholesterol, 127 mg/dl, prealbumin, 12.4 mg/dl (NL, 22–44), and retinol binding protein, 1.9 mg/dl (NL, 2.4–7.0) were all below the normal ranges. He showed marginal hypothyroidism due to non-thyroidal illness: TSH 0.08 µU/ml, T3 0.93 ng/ml and T4 6.7 µg/dl. LH (<0.5 mU/ml), FSH (1.2 mU/ml), testosterone (<5 ng/ml) and IGF-1 (35 ng/ml) were all very low.

Esophagography revealed a functional obstruction at the cardiac level, with almost complete stagnation of the contrast medium for 60 sec. Achalasia was definitively diagnosed with the findings of an esophagogram and a functional test of the esophagus. Endoscopic balloon dilatation
was performed 3 times in the following 6 mo, with only partial and transient remission of dysphagia. At 12 yr and 6 mo of age, the patient finally underwent laparoscopic extramucosal myectomy with fundoplication, and subsequently there was a marked amelioration of all symptoms.

Figure 1 summarizes the changes in the serum levels of testosterone and IGF-1 before and after the surgical manipulations. The figure depicts a marked increase in IGF-1 level soon after the balloon dilatation followed by a gradual increase in testosterone level. Figure 2 shows the growth chart of the patient. The height is plotted with both chronological and bone ages. Growth retardation was progressive by the time the fundoplication surgery was performed. The patient has shown catch-up growth since then together with the advent of pubertal development. At 15 yr and 6 mo of age, his height was 153.6 cm (–2.6 SD) and body weight was 38.9 kg (–1.9 SD), with a testis volume of 20 ml. His bone age was still two years younger than the chronological age, therefore, his final height is expected to reach the normal range.

Discussion

From the clinical features, achalasia in children is classified into neonatal, infantile and adult types, the latter occurring from school age and thereafter. Infantile achalasia tends to be progressive, and is more commonly accompanied by lung complications and malnutrition than the adult type. Conservative therapy is usually ineffective on the infantile type. Achalasia is very rare in children. Each year 0.4–1.1 per 10^5 population are affected, but only 3–4% of the patients are children (1). Diagnosis of achalasia in children is sometimes difficult, because the symptoms tend to be non-specific and the incidence of the disease is very rare. It is diagnosed within 3 yr after the onset in 80% of cases, but it takes longer than 5 yr for diagnosis in 9% of cases (2), as was the case with the present patient.

Nutritional growth retardation is characterized by proportional short stature without deformities in the extremities, and without emaciation in most cases (3). A careful check up of past growth records may reveal the onset of failure to thrive. Laboratory features of malnutrition are low levels of nutritional markers such as prealbumin, retinol binding protein, transferrin, albumin, choline esterase, branched chain amino acids and total cholesterol (4), as was the case with the present patient. Decrease in serum IGF-1 level is more prominent in patients with protein calorie malnutrition than in those with nutritional growth retardation. Delay in bone age is not always remarkable in patients with nutritional growth retardation. In the present case, the decrease in serum IGF-1 level was very severe, along with marked delay in bone age and pubertal development.

Achalasia is sometimes associated with involuntary movement due to myopathy or Parkinson’s disease. To date, its relationship with choreoathetosis has been unknown. Choreoathetosis preceded the onset of dysphagia, and this clinical course masked the diagnosis of achalasia in the present case. The involuntary movement disappeared after the surgery. In summary, this is a rare case with severe nutritional growth retardation caused by achalasia. The onset of symptoms dated back to <1 yr of age, and thus, the diagnosis was difficult. After laparoscopic extramucosal myectomy with fundoplication, catch-up growth has been observed, with pubertal development and a marked increase in serum IGF-1 level. Achalasia should be added to the list of causes of nutritional growth retardation in children.

References

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