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Case Report

ASPHYXIATING THORACIC DYSTROPHY: SURGICAL CORRECTION AND 2-YEAR FOLLOW-UP IN A GIRL

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Summary A 15-month-old girl under mechanical ventilation with asphyxiating thoracic dystrophy underwent surgical thoracic expansion according to the procedure of Todd *et al.* (1986). Now aged 4 years, she is free from respiratory distress, is of normal intelligence, and leads an active life.

Key Words asphyxiating thoracic dystrophy, Jeune syndrome, surgical treatment, Todd's thoracic expansion technique, long-term prognosis

INTRODUCTION

Asphyxiating thoracic dystrophy (Jeune syndrome) is an autosomal recessive disorder with constriction and narrowing of the thorax; broad, short, and horizontal ribs; and short-limbed dwarfism with abnormalities of the pelvis and limb bones. Cystic lesions occur in the kidney, liver, and pancreas. Renal complications may appear as early as 2 years of age, and are progressive. There is an increasing awareness that the disorder has a wide spectrum of severity, with neonatal death as a consequence of asphyxia on its one end, and short stature with a narrow thorax in an otherwise normal individual on the other end (Giorgi *et al.*, 1990). Among the patients with a chest circumference below 28 cm at birth, there has been no instance of survival beyond 2 years of age without surgical intervention (Ravitch, 1977).

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Various attempts at surgical correction of the thoracic constriction have been reported with mixed outcome (cf. Ravitch for references up to 1977). Todd *et al.* (1986) reported a procedure of sagittal sectioning between the sectioned sternal halves, successfully applied to a 9-month-old girl with a severe form of the disorder. We here describe application of the procedure to a 15-month-old girl with the disorder, together with a 2 year follow-up history.

CASE REPORT

The patient, a girl, was born at full-term to a 31-year-old, G3P2 mother and 32-year-old father, both healthy and unrelated. The first child, a boy, had a chest circumference of 26.4 cm (−3.5 SD), and died from respiratory failure at 16 days of age. The second child, a boy, was normal and healthy.

The patient at birth weighed 2,782 g (−1.1 SD), length 45 cm (−2.4 SD), head circumference 33 cm (−0.1 SD) and chest circumference 27.5 cm (−3.1 SD). She was referred to us at 3 months of age in view of developing cyanosis at crying. She had a narrow, constricted thoracic cage, short, horizontal ribs, and short limbs.

She was free from respiratory distress until 8 months of age, when she was admitted to us with fever, cough, and wheezing. She had respiratory arrest on the night of admission. She was resuscitated, intubated, and sustained on mechanical ventilation for 18 days. Similar episodes occurred 4 times during the ensuring 7-month-period with increasing duration of mechanical ventilation and decreasing intervals. She failed to thrive.

At age of 15 months, she weighed 6,983 g (−2.5 SD), length 67.4 cm (−3.4 SD), and chest circumference 40.0 cm (−3.1 SD). She had a bell-shaped thoracic cage, short ribs (Fig. 1, A and B), short limbs and hands, irregular epiphyses and metaphyses of the tubular bones, and hypoplastic iliac wings. Analysis of arterial blood gas revealed retention of CO_2 at 55 to 65 mmHg, and chronic respiratory acidosis of pH 7.26 to 7.34 and Base excess at 0 to +4. Lung function tests at age 14 months included: crying vital capacity 80 ml, peak inspiratory flow 291 ml/sec, forced vital capacity 75.1 ml, 1-sec forced expiratory volume 56.6 ml, and 1-sec forced expiratory rate 75.4%. Her parents did not approve of tracheostomy. She was constantly on intravenous bronchodilators, and nedeed sedation on several occasions.

Surgical thoracic expansion was performed at 15 months of age according to the procedure of Todd *et al.* (1986), when the patient was on mechanical ventilation for 4 weeks. Briefly, a sagittal section was made on the midline of the sternum, a 3.2 cm wide, methyl methacrylate prosthesis was placed between the sectioned halves, and fixed with wires (Fig. 1, C and D). She was free from mechanical ventilation 9 days after the operation, and was discharged at the 22nd postoperative day. Lung function tests a week after the operation were: crying vital capacity 99 ml, peak inspiratory flow 375 ml/sec, forced vital capacity 94.9 ml, 1-sec forced

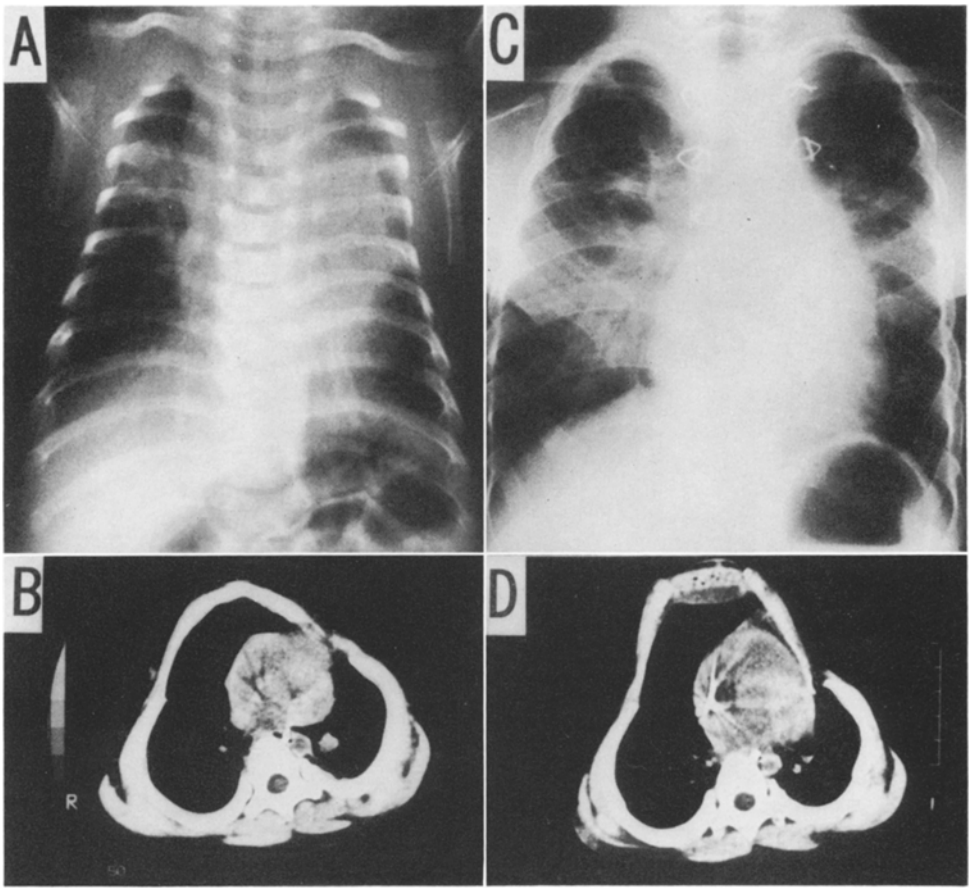

Fig. 1. A. Chest X-ray film at age 14 months. A narrow thoracic cage with short, horizontal ribs. B. Computed tomographic section with a bell-shaped thorax. C, D. Those 10 days after surgical correction.

expiratory volume 83.6 ml, and 1-sec forced expiratory rate 88.1%.

She has remained free from respiratory distress, and led normal life during the 2 9/12 year follow up period. When last seen at age 4 years, she weighed 14.2 kg (−0.6 SD), height 94.7 cm (−1.3 SD) and chest circumference 50.8 cm (−0.5 SD) (Fig. 2). She was of normal intelligence. Her liver, pancreas, and kidneys were both morphologically and functionally normal.

DISCUSSION

Past attempts at surgical expansion of the thoracic cage in asphyxiating thoracic dystrophy have included subperichondrial resection of 7 to 10 mm portions

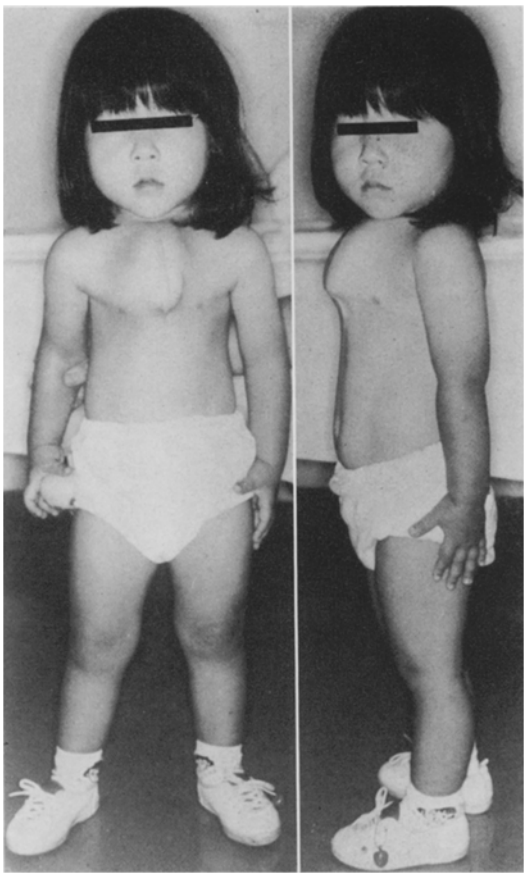

Fig. 2. The patient at age 4 years.

of rib cartilages; splitting the sternum and inserting an iliac graft; splitting the sternum and separating the halves with a rib graft; and splitting the sternum and holding the halves with a stainless sheet wire strut sutured to the ribs (cf. Ravitch, for references). These attempts were either unsuccessful with the patient dying soon after operation, or temporarily successful but cumbersome. On the other hand, the procedure of Todd *et al.* (1986), as applied to the girl we described, is simple and effective. Todd's patient, a 9-month-old girl, received a 2.5 cm-wide prosthesis, while our patient, a 15-month-old girl, received a 3.2 cm-wide prosthesis. Otherwise, the operative procedure used was essentially identical. The long-term follow-up data are not available of Todd's patient, while the girl we described was followed up for 2 9/12 years.

Our patient was doing well when last seen at age 4 years, although her respiratory capacity was still subnormal. Several factors seem to have worked in her favor. She survived up to 15 months of age when she underwent operation, where-

as her elder brother, apparently suffering from the same disorder, succumbed at 16 days of age. She was without renal, hepatic, or pancreatic insufficiency, complications that are likely to affect her long-term prognosis.

In conclusion, the procedure applied to our patient, splitting the sternum and inserting a prosthesis, is the method of choice in the surgical treatment of severe asphyxiating thoracic dystrophy.

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