

# Severe scoliosis associated with Costello syndrome: a case report

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## ABSTRACT

Costello syndrome is characterised by dwarfism, unique cutaneous lesions, a distinct facial gestalt, and mental retardation. There have been no detailed reports of severe spinal deformities requiring surgical treatment as a complication of Costello syndrome. We report a case of a 10-year-old girl with progressive scoliosis associated with Costello syndrome. She underwent anterior release and posterior surgical correction and fusion from T5 to L2 using a third generation hook and rod system plus spinous process wiring. Congenital portal vein deficiency and coagulopathy were other major complications. At 15-month follow-up, the patient had good balance and no evidence of instrumentation failure.

*Key words:* scoliosis; spinal fusion

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## INTRODUCTION

In 1977, Costello<sup>1</sup> reported 2 patients with a new syndrome characterised by short stature, redundant skin on the neck, palms, soles and fingers, curly hair, relative macrocephaly, a depressed nasal bridge, papillomata around the mouth and nares, a distinct facial gestalt, hyperextensible joints, and mental retardation. Several additional cases have been reported since then. Nonetheless, severe spinal deformity requiring surgical treatment has not been reported in people with Costello syndrome. We present the first detailed case of a girl with Costello syndrome who underwent anterior release, posterior surgical correction and fusion with the segmental spinal fixation for severe scoliosis.

## CASE REPORT

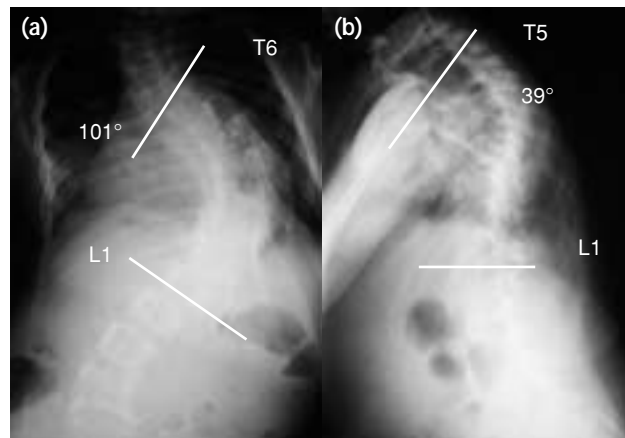
An 8-year-old girl with a scoliotic spinal deformity was assessed as an out-patient and recommended



**Figure 1** The patient presents with generalised macrocephaly, genua valga, and dark skin. The deformity of the thoracic cage is severe. Other features include curly hair, coarse face, a hypoplastic nasal bridge with a bulbous nasal tip, low set and posteriorly angled ears, thick lips, macroglossia, macrostomia, and short neck.

for an orthosis. She was unable to wear the orthosis appropriately because of severe mental retardation and her scoliosis continued to progress. In July 2003 she was referred, at age 9 years and 11 months, to the National Defense Medical College, Japan, for surgical treatment of her spinal deformity.

This patient was born by Caesarean section to healthy 33-year-old parents at 39 weeks of gestation. Her birth weight was 4465 g, body length 53 cm, and head circumference 40 cm. At 6 months, she was diagnosed with hydrocephalus and a ventriculoperitoneal shunt was inserted. At 8 months, she was diagnosed with Costello syndrome and followed up as an



**Figure 2** Radiographs of the spine showing (a) right thoracic Cobb angle from T6 to L1 being 101° and (b) kyphosis angle from T5 to L1 being 39°.

out-patient. Her karyotype was normal (46 XX). A computed tomographic scan of the abdomen performed at age 4 years due to hyperammonaemia showed congenital portal vein deficiency and a tumour about 2 cm in diameter in the liver. It was suspected that the tumour was a focal nodular hyperplasia. Deafness was noted at the age of 6 years and found to be due to a developmental anomaly of the auditory ossicles.

On admission, the patient was 10 years and 4 months old, 130 cm tall, and weighed 36.8 kg. Physical examination showed macrocephaly, curly hair, a coarse face, hypoplastic nasal bridge with a bulbous nasal tip, low set and posteriorly angled ears, thick lips, macroglossia, macrostomia, short neck, genua valga, hallux valgus, syndactyly of the left second and third toes, and dark skin (Fig. 1). Joint hyperextensibility was not seen. An examination of her back revealed thoracic scoliosis, with a right thoracic rib hump of 4.5 cm and severe deformity of the thoracic cage. Neurological examination revealed left ankle clonus and bilateral Babinski reflexes. Severe mental retardation was observed.

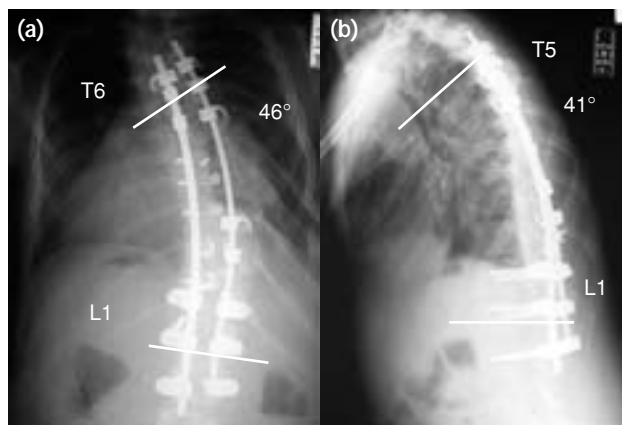
Results of blood examinations showed that the serum aspartate aminotransferase (AST) level was 49 IU/l (normal range, 6–43 IU/l), prothrombin time activity 62.4% (normal range, 80–120%), and activated partial thromboplastin time 48.4 seconds (control, 30.3 seconds). A Doppler echocardiographic study was normal. Serial plain radiographs of the spine showed that the scoliosis was progressive. The right thoracic Cobb angle from T6 to L1 had advanced from 32° to 101° over the preceding 22 months and the ky-



**Figure 3** Magnetic resonance imaging showing syringomyelia at the middle thoracic level.

phosis angle from T5 to L1 was  $39^\circ$  (Fig. 2). Magnetic resonance imaging (MRI) showed syringomyelia at the middle thoracic level but not the cervical level (Fig. 3).

At age 9 years and 11 months, anterior release, posterior surgical correction and fusion was performed, followed by costectomy of the right seventh rib and anterior release from T7/8 to T10/11 disc levels. The vertebrae were covered with thick soft tissue that bled heavily. The rib was split into bone chips that were grafted to each disc level. The patient was then placed in a prone position, and posterior correction and fusion was performed from the T5 to L2 levels using the third generation hook and rod system plus spinous process wiring. Spinous process wiring was applied because syringomyelia was observed at the middle thoracic level on MRI. The paravertebral muscle also bled heavily. During the corrective procedure, the spinal evoked potential (SEP) decreased 30% in amplitude, so correction was reduced to allow the SEP amplitude to recover. The operation time was 12 hours, and the intra-operative blood loss was 6313 ml. Proliferation of capillaries, small vessels, and lym-



**Figure 4** Postoperative radiographs showing (a) the scoliosis angle improved to  $46^\circ$  and (b) the kyphosis angle to  $41^\circ$ .

phoid ducts was observed in histological sections of the thick soft tissue around the vertebrae. Postoperatively, the scoliosis angle improved to  $46^\circ$  (correction rate, 55%) and the kyphosis angle was  $41^\circ$  (Fig. 4).

Postoperatively, the patient developed symptoms of disseminated intravascular coagulation (DIC) and continuous haemodiafiltration was performed. After removal of the chest drain, a right chylothorax was observed and the drain was replaced, and left in place for 32 days. The patient began to learn to stand with a hard orthosis at day 33, and at day 42 she attempted to ambulate with a walker. At 15-month follow-up, her balance had improved remarkably. Plain radiographs showed a  $7^\circ$  loss of correction but no instrumentation failure.

## DISCUSSION

In 1977, Costello<sup>1</sup> reported 2 children with nasal papillomata, coarse faces, mental retardation, and growth retardation. Additional notable features of this syndrome include high birth weight, curly sparse hair, joint hyperextensibility, and tight Achilles tendons.<sup>2</sup> The aetiology of Costello syndrome remains unknown.<sup>3</sup> Disruption of elastin fibres has been shown in 2 patients: one died of rhabdomyolysis at 6 months.<sup>4</sup> Normal elastin fibres have been reported in other cases.<sup>2</sup> To make the diagnosis, it is important to combine the clinical features with a history of early failure to thrive and cardiac and/or skeletal abnormalities.<sup>5</sup> As hydrocephalus and syringomyelia are often seen in this syndrome, MRI should be

performed in such patients. In the present case, congenital portal vein deficiency and coagulopathy were observed, both of which have not been described in previous reports and are extremely rare.<sup>6</sup>

Spinal deformities associated with Costello syndrome have been reported: 17% of which are scoliosis and another 17% are kyphosis.<sup>7</sup> Severe deformity requiring surgical treatment is extremely rare and only one child required surgery.<sup>7</sup>

In our patient, severe mental retardation precluded the use of a brace and her severe spinal deformity necessitated surgical intervention. This decision was carefully made in view of the possibility of coagulopathy stemming from her congenital portal

vein deficiency. We reviewed the case thoroughly with anaesthesiologists and a paediatrician before finally opting for a surgical procedure. During the surgery, both anterior and posterior exposures were bleeding heavily, leading to a total blood loss of over 6000 ml. Her postoperative course was complicated by DIC and chylothorax, but with intensive, lengthy therapy, the patient recovered and was able to ambulate with good balance at discharge. Instrumentation failure was not seen at the final follow-up visit, although it was possible that a spinal hook or rod was displaced. Careful long-term follow-up is required because of the possibility of disorders at adjacent disc levels.

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