Dorsal resection of a thoracic hemivertebra in a 4-year-old boy with endochondral gigantism. A case report

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The authors present what appears to be the first case of congenital kyphosis due to a T12 hemivertebra in a four-year-old boy with endochondral gigantism syndrome of unknown origin. Because of his overgrowth, the patient had severe medical and orthopaedic problems and was almost immobile. Prior to surgery, he experienced a rapidly progressive thoracolumbar kyphosis to 60° (T10-L2). MRI of the brain and spine showed critical protraction of the spinal cord and myelopathy from compression at T12. Single-stage posterior resection of the hemivertebra with spinal shortening and dorsal transpedicular instrumentation of T10-L2 was performed. Although the bone tissue was cartilaginous and dysplastic, 42° (30%) correction was achieved along with decompression of the spinal canal. The patient experienced no neurological impairment post-operatively. At follow-up examination 1.5 year after surgery, the patient's movement disorder had improved markedly and he was able to stand and walk.

This very rare case demonstrates that single-stage posterior hemivertebra resection and transpedicular instrumentation for correction of congenital kyphosis can be a safe and effective procedure even in a very challenging case.

Keywords : congenital kyphosis ; endochondral gigantism ; myelopathy ; hemivertebra resection ; transpedicular instrumentation ; children.

INTRODUCTION

Congenital deformities of the spine caused by anomalous vertebral development are less common than idiopathic deformities [8]. Such deformities are present at birth, but may not become clinically apparent until later in childhood. Congenital kyphosis is a spinal deformity caused either by dorsal hemivertebrae or by defects of segmentation. Dorsal hemivertebrae occur after a failure in formation of the anterior part of the vertebral body, resulting in impairment of longitudinal growth

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anteriorly (20). Since progression of the kyphosis occurs as a consequence of biomechanics, there is a propensity for neurologic deficits when spinal cord compromise occurs (21).

To the best of our knowledge, this is the first case of congenital thoracic kyphosis caused by a wedge-shaped T12 hemivertebra from a suspected endochondral ossification defect in a 3-year-old male with gigantism (13). Despite his severe physical disability, the patient underwent resection of the dorsal hemivertebra and transpedicular instrumentation by posterior approach at a relatively young age because of rapid progression of the kyphosis and signs of myelopathy. His symptoms improved markedly after surgery.

CASE REPORT

History

A male infant was delivered by caesarian section to a healthy 30-year-old woman at 34 weeks of gestation. The patient was the family's third child. Pregnancy had been complicated by polyhydramnios and gestational diabetes treated with subcutaneous insulin from the 26th week. The parents were of German, Caucasian descent, healthy, and not consanguineous. Paternal height was 169 cm and maternal 170 cm. The oldest brother had a birth weight of 4260 g, and the middle brother 4500 g. At birth, the patient's weight was 6600g (+ 6.8 SDS), length was 61 cm (+ 6 SDS), and head circumference (OFC) was 38.5 cm (+ 3.2 SDS).

A striking phenotype was noted at the time of birth and became clearer during the follow-up period. There was macro-brachycephaly with a dysmorphic face, a small thoracic cage, a long trunk, deformity of the spine, rhizomelia, large hands and feet, absence of subcutaneous fat, a small umbilical hernia, inguinal hernias, and large joints with mild contractures. Immediate mechanical ventilation was necessary for obstructive apnea. Tracheal cannulation was performed at the age of 14 days for persistent obstructive apnea caused by complete mesopharyngeal and laryngeal collapse as well as tracheo-broncheal instability. Because of recurrent hypoglycaemia, continuous overnight feeding via

gastrostomy was continued until the age of 18 weeks. Neonatal screening, complete blood counts, blood chemistry, urine, and serum amino acid values were normal. Karyotype was 46XY, and multicolor- fluorescence *in situ* hybridization (FISH) and FISH-subtelomere region were normal. Echocardiogram revealed an atrial septal defect (ASD) type II and a patent ductus artery (PDA). During the first two months of life, intermittent primary hypothyroidism and central precocious puberty occurred (testicular volume increased to 11 ml). These conditions were treated for 12 months respectively with thyroxin and the gonadotropin-releasing hormone (GnRH) analog Leuprorelin. Skeletal survey as well as computed tomography of the head and chest showed dysplasia of bones with endochondral ossification centers, i.e. the sphenoid body, pars basilaris/lateralis of the occipital bones, the zygomatic arch, the mandibular head, vertebral bodies, the pelvis, long bone epiphyses, etc., from accelerated or uncontrolled growth, in contrast to the almost normal appearance of bones undergoing membranous ossification. Bone density was decreased. At the age of 18 months, left hand radiographs showed advanced and dissociated bone ages : telophalangeal 4 years, mesophalangeal 6 years, basophalangeal 8 years, metacarpal 5 years, and carpal 5 years. Motor and mental developments were retarded.

On presentation to our orthopaedic department, at the age of 17 months, clinical signs of thoracic kyphosis were evident. Magnetic resonance imaging (MRI) of the brain and spine showed a dorsal hemivertebra at T12, pronounced osteodysplasia, and hypertrophy of the sphenoid and occipital bones as well as the craniocervical transition. These findings resulted in elongation of the brainstem and protraction of the spinal cord with thinning and signal alterations of the upper cervical portion. To minimize the risks of surgery and iatrogenic neurologic impairment, the initial plan was to follow the patient on an annual basis until the age of 4 to 5 years, when surgical correction of the spinal deformity would be attempted.

At the age of 29 months, MRI showed no relevant differences concerning the brainstem and the spinal cord. Neurologic status was not impaired by

the abnormal anatomy and somatosensory evoked potentials (SEPs) were normal.

In his first 3 years of life, the patient sustained three long bone fractures at varying sites without adequate trauma. Rapid and exaggerated callus production was noted following each fracture. Endocrine studies performed at various points in time revealed very low or suppressed growth factors and high insulin sensitivity.

Examination

At 3.5 years of age, the patient's height was 143 cm (fig 1a). Progressive thoracolumbar kyphosis was clinically evident (fig 1b). He was able to sit independently, move on the floor by crawling, and pull himself to his knees, but he was unable to pull himself to standing because of the limited extension of his lower limbs and sagittal imbalance of the spine, resulting in immobility (fig 1a). Excessive growth and weight gain were apparent. At 19 months of age, weight was 27.5 kg, height + 8.4 SD, body mass index (BMI) + 3.7 SD, ratio of sitting/standing height was 0.71 (97%), and body fat (scapular/triceps) 4%. At 3.5 years, weight was 36.5 kg, height + 9.6 SD, BMI + 2.8SD, and OFC 62 cm. The bone density scan (DXA) revealed exceptional osteoporosis (BMD 0.791 g/cm^2).

Kyphosis had increased dramatically. Antero-posterior and lateral radiographs of the spine showed 60° of kyphosis (fig 2). MRI of the spine and brain showed marked spinal stenosis at T12 causing both spinal cord compression and protraction with signs of myelopathy and signal alteration in the cervical spine. The brainstem was critically stretched (fig 3a). The neurological status of the boy was normal ; however, SEPs of the lower extremities were not measurable, most likely due to posterior cord involvement.

Fig. 2. — a. Radiographs (lateral and anterior-posterior views) of the spine at the age of 3.5 years in upright position, prior to surgery. The hemivertebra is located at T12. The kyphotic curve is 60° (T10-L2).

Fig. 2. — b. Radiographs of the thoracolumbar region (lateral and anterior-posterior views), prior to surgery.
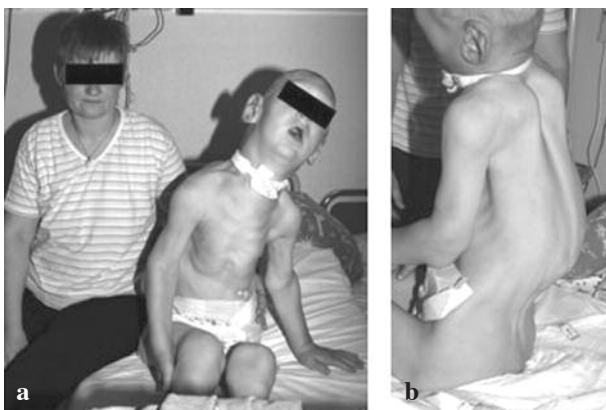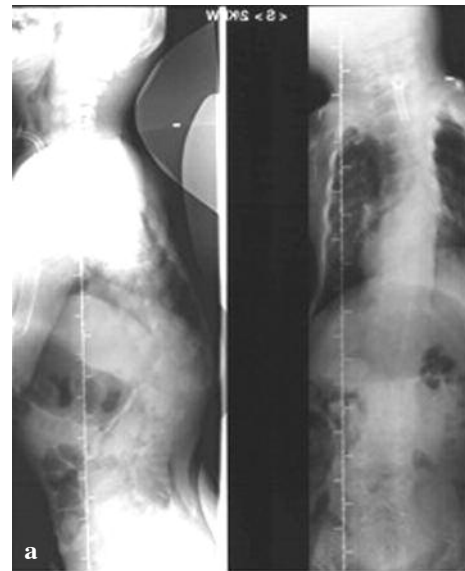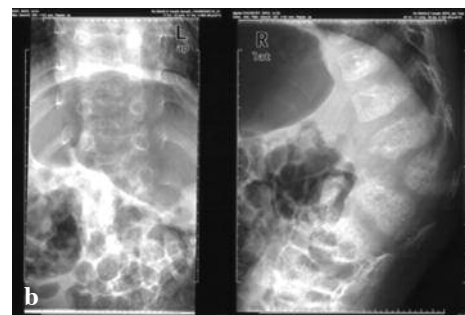

Fig. 1. — (a) Frontal view of the patient (age : 3.5 years ; 143 cm, 39 kg) and his mother (164 cm). The patient is able to sit, has a tracheostomy for tracheomalacia, is fed by percutaneous gastrostomy tube, and is incontinent. (b) Lateral view of the patient (age : 3.5 years). Congenital kyphosis of the thoracolumbar region is evident.
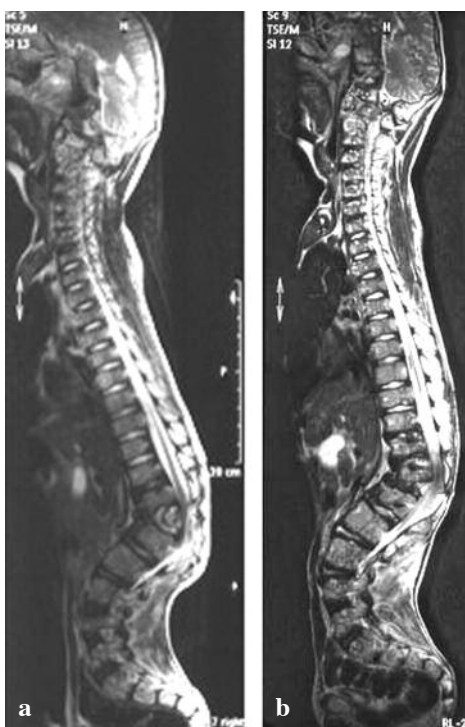
Fig. 3. — MRI of the spine and brain. Before surgery (a) relevant elongation of the brain stem with signal alterations in the cervical spine and myelopathy of the thoracolumbar region are evident. Follow up 18 months after surgery (b).
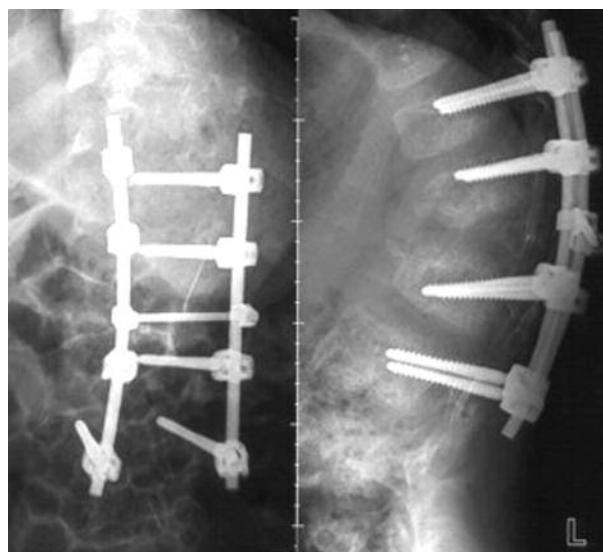

Fig. 4. — Radiographs of the thoracolumbar region (lateral and anterior-posterior views) after single-stage posterior resection of the hemivertebra T12 with dorsal instrumentation (T10-L2). The Cobb angle of the segmental kyphosis (T10-L2) measures 42° after surgery (30% correction).

We considered these clinical and radiologic findings as indications for surgery. Resection of T12 and dorsal instrumentation of T10–L2 was planned to correct the spinal deformity, shorten the spine, and decompress the spinal cord.

Procedure and Postoperative course

Under general anaesthesia, the patient was placed in a prone position with the abdomen relieved of all pressure on rolls. The back was prepared and draped in the routine fashion. A longitudinal skin incision was made from two segments above to two segments below the hemivertebra. Paravertebral muscle was retracted laterally, and the laminae were exposed. The hemivertebra was identified radiographically. The lamina of the hemivertebra was identified and removed with the transverse process. Epidural bleeding was controlled with thrombin soaked gelfoam. The dural sac was found to be narrowed and deviated posteriorly. The body of the hemivertebra with its periosteum was then entirely removed. The adjacent intervertebral discs were completely excised, and the end plates of the adjacent vertebrae were curetted. Bone chips from the removed hemivertebra were inserted into the defect. Under fluoroscopy, Titanium polyaxial pedicle screws (6 × 50mm, ART-System®, Advanced Medical Technologies, Germany) were inserted into T10, T11, L1, and L2. Two pre-flexed rods were connected to the pedicle screws. Compression force was applied to shorten the spinal column, and final stabilization was achieved. One cross-connector was fixed between the rods. The Stagnara wake-up test was performed. Adequate bone graft was implanted posteriorly and the wound was closed in standard fashion. Overall operative time was 257 minutes, with an approximate blood loss of 400 mL. Postoperatively, the neurologic status was unchanged. Histology of the resected vertebra showed chondral dysplasia. Four days after surgery, a stiff thoracolumbosacral brace (polyethylene) was fitted and the patient was mobilized with full weight bearing.

Postoperatively, the kyphotic angle (T10-L2) of the patient was 42°, a correction of 30%. The boy
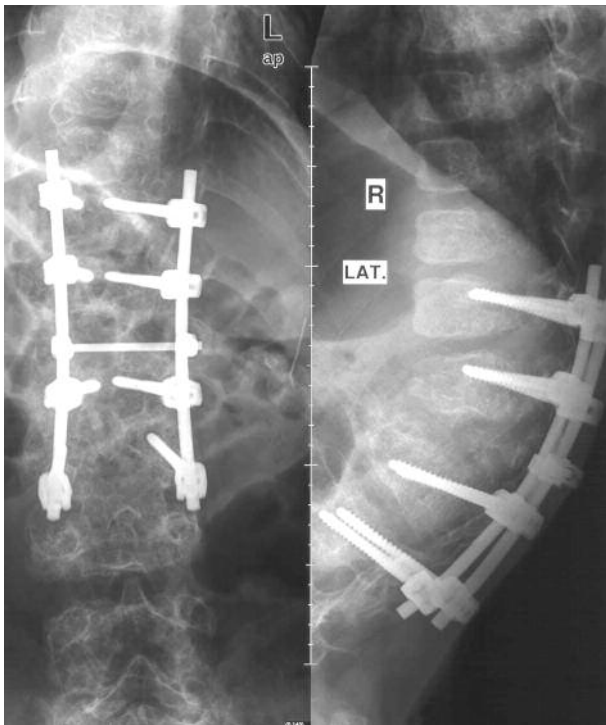
Fig. 5. — Radiographs of the thoracolumbar region (lateral and anterior-posterior views) 18 months after surgery. The Cobb angle of the segmental kyphosis (T10-L2) is 44° (2° loss of correction since surgery).

was discharged 16 days after surgery. No complications occurred during his admission.

Eighteen months after surgery, the boy's upper and lower extremity strength had clearly improved. He was able to stand and walk with the walking frame. Neurologic status was completely normal. The follow up MRI of the spine showed relevant decompression of the spinal cord (fig 3b). Compared to films performed immediately after surgery, radiographic evaluation showed 2° loss of correction (fig 5).

DISCUSSION

Congenital kyphosis is much less common than congenital scoliosis ; it results from failures of segmentation or of vertebral formation (*16*). A mixed defect can also occur (*20*). Our patient was diagnosed with a dorsal hemivertebra of T12. Formation of the anterior portion of the vertebra, which normally takes place between the 20th and 40th day of devel-

opment, failed to occur. This caused progressive kyphosis (*10*). Kyphosis develops prenatally during the late phase of chondrification and progresses throughout the period of ossification (*16*).

Because of its dorsal location and of the surrounding muscle tension, the residual hemivertebra becomes wedge-shaped. Kyphotic progression occurs as a mechanical consequence (*21*).

In a study of 130 patients, Winter *et al* reported on the natural history and clinical management of congenital kyphosis, and classified the deformity into three types based on the precipitating vertebral anomalies (*20*). Eighty-six patients (66 %) had vertebral body formation defects (Type I), 19 patients (15%) had vertebral body segmentation failures (Type II), 18 patients (14 %) had mixed-type defects (Type III), and 7 patients (5%) had non-classifiable kyphoses. Thirty patients were followed for more than one year, and showed a mean progression rate of 7° per year. Treatment of choice was early posterior fusion for patients under 3 years of age. In older children with severe deformity and critical angles of more than 50° of kyphosis, combined anterior and posterior fusion was preferred (*18-20*). Excision of hemivertebrae was performed as early as 1928 by Royle (*11*). Early case reports include neither operative details nor radiographic examinations (*3,17*). In 1965, Hodgson published a study discussing the correction of fixed spinal curves, including excision of hemivertebrae (*4*). Later, Leatherman and Dickson outlined a two-staged anterior and posterior corrective surgery for congenital spine deformities (*6*). The authors maintained that the use of posterior spinal fusion alone would not prevent further progression of the congenital deformity. Several articles published subsequently described a two-stage procedure to resect the hemivertebrae and correct the congenital deformity (*5,15*). In the past two decades, surgeons have increasingly performed a less invasive, single-stage resection of hemivertebrae by posterior approach alone in patients with hemivertebrae at the thoracolumbar level (*1,7*).

Braces and other nonoperative therapies have proven to be ineffective, therefore treatment of dorsal hemivertebrae should be surgical. Exact indications and selection of surgical technique remain controversial (*21*).

Congenital kyphosis has a propensity to cause neurologic deficits. For cases with paralysis, posterior decompression of the spinal cord and early fusion is the treatment of choice (*2, 18*).

The length of fusion need not be long if an appropriately sized screw-rod system of transpedicular instrumentation is used. In 2002, Ruf *et al* published a retrospective study of 21 patients undergoing hemivertebra resection and transpedicular instrumentation using a posterior approach (*12*). In this study, the mean follow-up time was two years. Mean patient age was 5.8 years. Of the resected hemivertebrae, nine were located in the thoracic spine (T1–T9), eight in the thoracolumbar region (T10–L2), and four in the lumbar spine (L2–L4). Segmental kyphosis averaged 22.6° (range : 2-42°) before surgery, 7° (range : -4-21°) after surgery, and 9.4° (range -3-25°) at follow-up.

Our patient was treated with a posterior resection of the hemivertebra at T12 and transpedicular fusion of T10–L2 in a single-stage procedure, which is the current standard of care for this type of deformity. No neurologic complications occurred during the operation. The postoperative course was normal. The kyphotic curve measured 60° preoperatively and 42° after surgery, thus yielding a 30% correction.

In the literature, mean correction rates between 45.4 and 77.6% have been reported after single-stage posterior resection and instrumentation. In these series, mean ages of the patients were 10 (*9*) and 16.5 (*14*).years. Our patient had a combination of problems. Because of the massive kyphotic progression as well as the spinal imbalance additionally associated with gigantism, surgery was indicated at a very young age. Because of the patient's overgrowth, the vertebral bone was not only immature and histologically dysplastic, but also exceptionally osteoporotic. Greater correction was thus not attempted, in the hope of avoiding pedicle screw avulsion. The pedicle screws used were selected from an adult instrumentation system, of the largest possible diameter.

At final follow-up 18 months after surgery, muscle strength of the patient's extremities had improved enough that he was able to stand up and walk using a walking frame. The instrumentation was intact, and loss of correction was 2°.

CONCLUSION

Congenital kyphosis from dorsal hemivertebrae is a rare spinal deformity that is usually progressive, so that early surgical correction is recommended to avoid secondary deformities and neurologic deficits. To minimize the trauma from surgery, single-stage posterior resection of the hemivertebra with transpedicular instrumentation of a short segment appears as the most suitable option. The instrumentation used should be stable enough to withstand early postoperative mobilization of the young patient. The challenge in this particular case was a rare combination of problems : the patient's very young age, the extreme overgrowth resulting in vertebral dysplasia, the morphologic changes to the bone structure, and the osteoporosis.

A satisfactory correction of the kyphotic curve (30%) was achieved. This resulted in clear improvement in both spinal balance and muscle strength in all extremities. Although the surgery was tolerated well by the patient and was not associated with complications, close clinical follow-up with radiographic controls will be needed until cessation of growth to detect possible secondary deformities at an early stage of development.

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