Acta Orthop Scand 1988;59(1):66–70

Cervical instability in skeletal dysplasia

Report of 6 surgically fused cases

Olle Svensson and Stig Aaro

Six patients with skeletal dysplasias complicated by deformity or instability of the cervical spine were reviewed. Three patients had congenital spondyloepiphyseal dysplasia and 1 each suffered from Morquio's syndrome, pseudoachondroplasia, and chondrodystrophia calcificans congenita. Four patients had myelopathy of varying severity from slight weakness of one leg to tetraplegia. At a mean age of 6 (3–11) years, they underwent posterior fusion and immobilization in a halo brace. In all the cases the extent of postoperative neurologic recovery was, however, limited. Our cases illustrate the need of early diagnosis and fusion of cervical instability.

Well over a hundred different kinds of skeletal dysplasias have been described (Rimoin 1978). Partly because of our lack of understanding of the underlying pathomechanisms (Stanescu et al. 1984), no casual therapy can be offered. However, the growth disturbances may lead to a number of treatable complications, the most serious being compression of the spinal cord. It is commonly caused by atlantoaxial instability, but compression may also occur in congenital kyphosis (McKusick 1972). Although medullary compression may cause tetraplegia or sudden death, its presentation is usually more insidious, involving a slowly progressive loss of strength (Kopits 1976), which may be hard to evaluate in multihandicapped children. Neurologic recovery in these cases generally requires early diagnosis and rational treatment.

We report our experience with surgical treatment of cervical deformity or instability in 6 cases of skeletal dysplasia.

Patients and methods

The series comprised 6 boys – suffering from skeletal dysplasias complicated by atlantoaxial instability or cervical deformity – who were subjected to cervical spinal fusion during the period 1976–1985 inclusive (Table 1).

At a mean age of 6 (3–11) years, they underwent posterior cervical fusion and fixation in a halo brace. The operations were performed with the patient in halo traction, and recently also under spinal cord monitoring. Through a posterior midline incision, the arches were freed subperiosteally. In Cases 1–4, the atlas was more or less displaced into the foramen magnum, and because of this the fusion was made with the occiput. Cancellous bone grafts were obtained from the iliac crests. In Case 5, heterologous bone was used. In Case 1, we used internal fixation with wires according to Gallie (1939). External fixation was planned for at least 3 months, but in 3 patients the halo loosened prematurely after 2 months and was replaced by a brace until consolidation.

Case reports

Case 1. During his second year, this boy developed a progressive growth retardation with a disproportionately large head and a short trunk with a pronounced lumbar lordosis. Also, a slight weakness of one leg was noted. Radiographs showed delayed ossification of vertebrae and epiphyses. At the age of 2, an increased distance between C1 and C2 (12 mm) was found. At first, the instability had not been diagnosed because radiographs were not taken with the neck in full

Department of Orthopedics, Karolinska Institute University Hospital, S-141 86 Huddinge, Sweden

Table 1. Data for 6 patients with skeletal dysplasia and cervical spine instability

Patient	Diagnosis	Birth weight (kg)	Birth length (cm)	Neurologic deficit	Cervical lesion	Age at operation (yr)	Complication	Follow-up (yr)
1	Spondyloepiphyseal dysplasia	3.8	44	Weakness of one leg	Instability C1–C2	3		2
2	Spondyloepiphyseal dysplasia	3.7	47	–	Instability C1–C2	6	Loosening of pins	7
3	Spondyloepiphyseal dysplasia	3.2	45	–	Instability C1–C2	8	Superficial pin infection; loosening of pins	2.5
4	Morquio's disease	4.3	56	Tetraparesis	Instability C1–C2	5	Slight pressure sore	8
5	Chondrodystrophia calcificans congenita	1.7	44	Tetraparesis	Congenital kyphosis	6	Slight pressure sore	8
6	Pseudochondroplasia	3.5	48	Tetraparesis	Congenital kyphosis	11	Transient neurologic deterioration; loosening of pins	1

flexion. The clinical and the radiographic pictures were suggestive of spondyloepiphyseal dysplasia (SED). Because of the cervical instability and the neurologic symptoms, posterior fusion, internal fixation with wires, and autologous bone transplantation were done. The postoperative course was uneventful, and after 3 months his parents stated that he had become stronger and more physically active.

Case 2. This boy was apparently normal at birth. During his first years of life, his length decelerated from −2 standard deviations (SD) to −5 SD. Clinically and radiographically, he had changes typical of SED. The patient was physically active and had no neurologic deficits. At 6 years of age, an atlantoaxial separation of more than 10 mm was demonstrated. A spinal fusion from the occiput to the second cervical vertebra was performed. The postoperative course was uneventful.

Case 3. The clinical and radiographic findings, as well as the case history of this patient with SED are very similar to those of Case 2. At the age of 8, hypoplasia of the odontoid (Figure 1) and a mobility between C1 and C2 exceeding 10 mm was demonstrated. A posterior fusion of the occiput

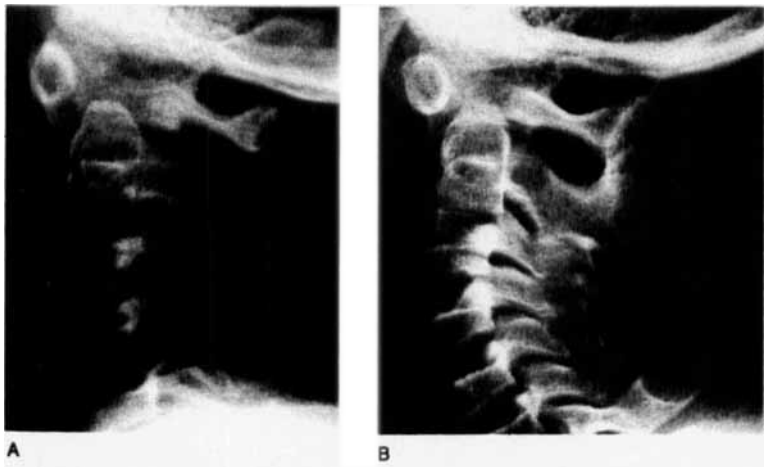

Figure 1. Case 3. Spondyloepiphyseal dysplasia congenita with hypoplasia of the dens axis.
A. Preoperatively.
B. One year after posterior fusion.

to C2 was done. The postoperative course was uneventful apart from a superficial infection around one of the pins.

Case 4. During his second year, this patient's length decelerated from +2 SD to −1 SD. At 4 years of age a progressive muscular weakness was noted, especially of the upper extremities. He had typical stigmata of Morquio's disease, e.g., a disproportionately large head with characteristic facial features, a short neck and trunk with kyphosis and pectus carinatum. Radiographs showed multiple deformities of the extremities and spine, including aplasia of the dens axis. Apart from an increased urinary excretion of keratan sulfate, the biochemical profile was normal. At aged 5, he was tetraparetic, atlantoaxial instability of 11 mm was demonstrated, and a posterior fusion was performed. Now, 8 years later, his tetraparesis is essentially unchanged: he cannot walk more than a few steps, and he has great difficulty in managing the activities of daily living.

Case 5. This boy was born prematurely after 35 weeks' gestation. At birth, short extremities, hyperthelorism, epicanthus, and a saddle nose were noted. Radiographs showed spotty calcifications in several epiphyses suggestive of chondrodystrophia calcificans congenita. At about 2 years of age a cervicothoracic kyphosis was observed, and during the following years he developed a slowly progressive tetraparesis with spasticity of the legs, weakness of the upper limbs, and difficulties to urinate. At the age of 5, he had a cervicothoracic kyphosis of 90°. Posterior spinal fusion from C5 to T3 was performed; the correction of the deformity did not exceed 20°, but the fusion healed in this position. Today, at aged 14, he has no urinary problems, but he still has weakness in one arm and of both legs, as well as a slight spasticity.

Case 6. This boy with pseudoachondroplasia had mutiple deformities and a progressive muscular weakness. At the age of 10, he woke up tetraplegic after a femoral osteotomy. Radiographs revealed a failure of formation of the body of C3, which was present only as a rudiment, with a local gibbus and a dislocation between C3 and C4 (Figure 2). Myelography and CT demonstrated a marked

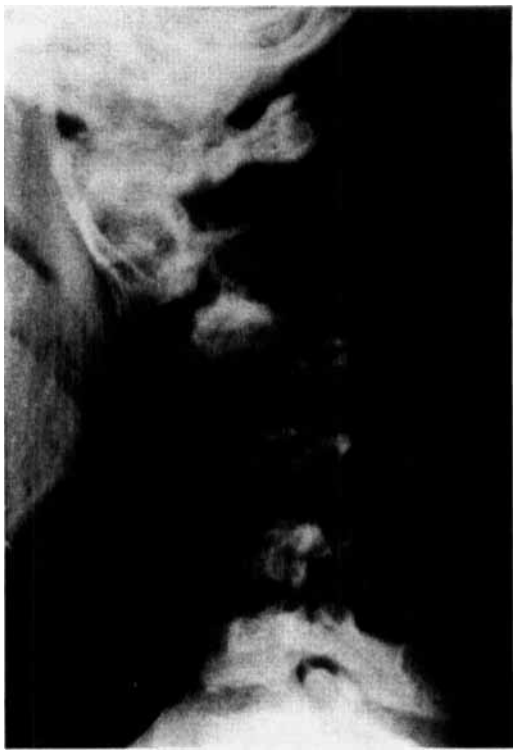

Figure 2. Case 6. Pseudoachondroplasia with cervical dislocation as a result of failure of anterior vertebral formation.

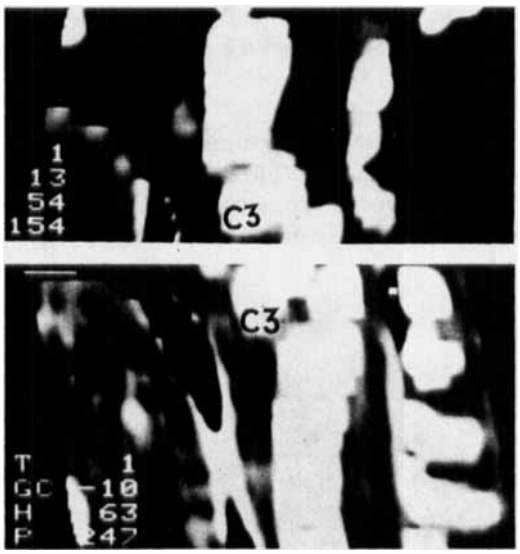

Figure 3. Case 6. Sagittal reconstructions of a computerized tomography of a cervical myelography showing that the medullary canal has an anteroposterior diameter of only a few millimeters. Because of the patient's deformity, the upper and the lower cervical spine were examined in different projections. Consequently, the angular deformity is not visualized on these two pictures.

local narrowing of the spinal canal (Figure 3). The tetraplegia regressed partially. Open-door laminoplasty (Itoh and Tsuji 1985) and posterior fusion were carried out. His neurologic symptoms temporarily increased, but now his condition is about the same as before spinal surgery.

Results and discussion

Compression of the spinal cord is a common complication in skeletal dysplasias. Kopits et al. (1976) reported clinically manifest myelopathy in about half of a series of patients suffering from various skeletal dysplasias. In Morquio's disease and SED most, if not all, patients develop dysplasia of the dens and atlantoaxial instability (McKusick 1972, Kopits 1976, Lipson 1977). In our series these diagnoses were responsible for the observed cases of atlantoaxial instability, and 2 of these patients had myelopathy. However, atlantoaxial instability also occurs in numerous other skeletal dysplasias (Kopits 1976). The other 2 patients included in this material both had congenital kyphosis, in which progressive myelopathy is also common (Winter 1983).

Typically, the myelopathy becomes manifest as a slowly progressive decrease of physical strength and stamina (McKusick 1972), whereas the sensory modalities usually are well preserved until the terminal phase (Kopits 1976). In our series the 4 patients with neurologic deficits all had clinical presentations involving a slow decrease of motor function, which is difficult to evaluate in multihandicapped infants; and the average doctor's delay, i.e., the time between documented symptoms and operation, was 3 years. It is worth noting that no patient complained of cervical pain. In Case 6 the symptoms had progressed slowly for several years when an injury was inflicted during endotracheal intubation. This illustrates the unpredictability of the neurologic affection (McKusick 1972).

Clearly, all patients with suspected skeletal dysplasias should have radiographs of the cervical spine not only as a part of the routine examination, but especially before endotracheal intubation. It is important that the examination includes projections taken in full voluntary flexion and extension; otherwise, instability may escape detection. Atlantoaxial instability is defined as an atlas-dens distance exceeding 3–4 mm, depending on the age of the patient (Fielding et al. 1976). In our cases, however, the recorded atlantoaxial gaps all exceeded 10 mm. Yet, evidence from myelographic studies indicates that the degree of medullary compression does not always correlate with the radiographically measurable instability (Lipson 1977), and computerized tomography is often valuable (Roach et al. 1984).

Because the benefit of surgery largely is prophylactic, absence of neurologic symptoms does not, according to our opinion, weaken indication for cervical fusion. Further, in Morquio's disease and SED, where the development of atlantoaxial instability is very common (McKusick 1972), it may be justified to perform cervical fusion as a prophylactic measure. Indeed, in cases of congenital kyphosis, e.g., with failure of anterior vertebral body formation, which often run a progressive course, a conservative attitude is dangerous (Dubousset 1983).

In all patients the posterior fusions healed. However, it is generally agreed that in the surgical treatment of advanced congenital kyphosis, an anterior fusion is necessary (Winter 1983). We believe that this does not always apply to the cervical spine. First, the load on the cervical spine is less than in the thoracic and lumbar spine; and secondly, the anatomy of the vertebral arches of the cervical spine places a posterior fusion in a mechanically favorable position. In Cases 5 and 6, we initially refrained from performing an anterior fusion, and it later proved to be unnecessary. In Case 1, we performed internal fixation; but we now think that this is unnecessary and even futile in cervical fusions in small children, because the arches are soft and stability can hardly be expected. External fixation gave sufficient stability. In this series, halo-bracing resulted in a number of complications (Table 1), most of which are avoidable. Thus, it is important to instruct the patient and the parents that local pain invariably means that the pins are coming loose and that any complaint from the patient merits rapid consultation.

Because the danger in these conditions lies in the instability, it seems obvious that the greater the instability, the stronger the indication for fusion. As the pathologic lesion is almost always a chronic myelopathy, we feel that laminectomy and extensive decompression are rarely, if ever, indicated, especially as extensive decompressive

operations entail a substantial risk of hematomyelia (Bunnell and MacEwen 1983). In Case 6 an anterior decompression was considered to be too dangerous, and the patient was instead subjected to an open-door laminoplasty (Itoh and Tsuji 1985).

Compression of the spinal cord is the most important orthopedic complication in skeletal dysplasias. Posterior cervical fusion is a simple and safe procedure, and we think that it should be performed on wide indications – preferably before the patient develops neurologic deficits.

References

Bunnell W P, MacEwen G D. Congenital deformities of the spine. In: Surgery of the musculoskeletal system (Ed. Evarts C M.). Churchill Livingstone, New York 1983:363–409.

Dubousset J. Cyphoses et cyphoscolioses angulaires. Rev Chir Orthop 1983;69(Suppl 2):53–9.

Fielding J W, Hawkins R J, Ratzan S A. Spine fusion for atlanto-axial instability. J Bone Joint Surg (Am) 1976;58(3):400–7.

Gallie W E. Fractures and dislocations of the cervical spine. Am J Surg 1939;46:495–9.

Itoh T, Tsuji H. Technical improvements and results of laminoplasty for compressive myelopathy in the cervical spine. Spine 1985;10(8):729–36.

Kopits S E. Orthopedic complications of dwarfism. Clin Orthop 1976;(114):153–79.

Lipson S J. Dysplasia of the odontoid process in Morquio's syndrome causing. quadriparesis. J Bone Joint Surg (Am) 1977;59(3):340–4.

McKusick V A. Heritable disorders of connective tissue. 4th ed. C.V. Mosby Co. St Louis 1972:583–611.

Rimoin D L. International Nomenclature of Constitutional Diseases of Bone: revision May 1977. Birth Defects 1978;14(6B):39–45.

Roach J W, Duncan D, Wenger D R, Maravilla A, Maravilla K. Atlanto-axial instability and spinal cord compression in children – diagnosis by computerized tomography. J Bone Joint Surg (Am) 1984;66(5):708–14.

Semine A A, Ertel A N, Goldberg M J, Bull M J. Cervical spine instability in children with Down syndrome (trisomy-21). J Bone Joint Surg (Am) 1978;60(5):649–52.

Stanescu V, Stanescu R, Maroteaux P. Pathogenic mechanisms in osteochondrodysplasias. J Bone Joint Surg (Am) 1984;66(6):817–36.

Winter R B. Congenital deformities of the spine. Thieme Stratton, New York 1983:229–62.

Acknowledgements

We thank Ms Annette Bäckman for typing the manuscript. This study was supported by grants from Sven Jerring's foundation.