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Dystrophic kyphoscoliosis in neurofibromatosis type I: a report of two cases and review of the literature

Received: 1 April 1996
Revised: 2 August 1996
Accepted: 12 August 1996

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Abstract We report two cases of dystrophic scoliosis in neurofibromatosis, each of particular interest. In the first, kyphosis was present with vertebral rotatory subluxation but no neurologic impairment, while the second patient showed manifest paraplegia due to rapidly progressive kyphoscoliosis. The importance of early surgical stabilisation, both front and back if possible, is stressed. Very sharp curves with progressive myelopathy should not be treated with halo-femoral traction because of the potential danger of evoking permanent paraplegia.

Key words Neurofibromatosis · Scoliosis

Introduction

Scoliosis is the most common skeletal deformity in neurofibromatosis, its incidence ranging from 10% to 60% [3, 4]. There is no consistent pattern in its appearance [3, 10]. Dystrophic kyphoscoliosis is usually located in the thoracic area and is feared for its rapid progression with development of paraplegia [2–4, 10, 15]. We describe two cases with manifest dystrophic signs (pencilling of ribs, scalloping and erosion of vertebrae), one presenting with rotatory subluxation, the other with almost complete paraplegia. We present the clinical and radiological features and our treatment of this difficult condition.

A successful fusion (anterior and posterior procedure) was achieved in both cases. The paraplegic boy, who initially deteriorated neurologically in a halo-femoral traction device, recovered completely after an anterior decompression and is now able to ambulate without any supporting device.

Case 1

A 13-year-old boy with neurofibromatosis type I was first seen in our hospital when he was 5 years old. He had ten café-au-lait spots and multiple skin neurofibromata spread over his body. Lisch nodules (iris hamartomas) were present. He had an optic glioma of the left eye. His thorax showed an extreme pectus carinatum. He had

Fig. 1 Vertebral subluxation of T3 on T4

Fig. 2 Scalloping of vertebrae. Intrathoracic tumour. The cord lies free in its canal

Fig. 3 Kyphosis angle corrected to 12° . Vascularised rib graft in situ

Fig. 4 CD instrumentation with scoliosis corrected to 16°

disabling back pain and a high thoracic kyphoscoliosis, convex on the right, with a gibbus height of 2.5 cm. Rotation was 14° , measured with the Bunnel scolimeter. The curve was well compensated. Radiographs revealed a right convex scoliosis T2-T8 of 35° and a kyphosis of 54° (T2-T6). The third thoracic vertebra was partially luxated on the fourth (Fig. 1). There was erosion and scalloping of the corpora of the third, fourth and fifth thoracic vertebrae. MRI showed a large intrathoracic paravertebral mass on the right side (Fig. 2). There was no ingrowth in the intervertebral foramina and no cord compression. Preoperatively the patient complained of paresthesiae in his right hand, probably due to compression of the brachial plexus by the tumour. On 8 January 1992, a posterior spinal fusion was performed from C7 to T10, with Cotrel-Dubousset (CD) instrumentation. Autologous bone grafts were used. On 22 January 1992, an anterior fusion was added. Through a right lateral thoracotomy the T3-T4, T4-T5 and T5-T6 discs were removed and subsequently a vascularised rib graft was inserted from T2 to T7 (Fig. 3). Because of ingrowth into the hilus, the intrathoracic tumour seen on MRI was only partially removed. Histological examination confirmed a ganglioneuroma. Ten days postoperatively the patient was carefully mobilised in a plaster jacket.

Six months later the jacket was abandoned completely. One year postoperatively the patient was free of complaints, went to school every day and was regaining sports activities. The scoliosis measured 16° and the kyphosis 12° . The fusion looked solid (Fig. 4).

Case 2

A teenage boy originating from Surinam was first seen when he was 13 years old for a scoliosis detected during routine school medical screening. At clinical examination we detected several café-au-lait spots and subcutaneous neurofibromata spread over his body. His visus was not disturbed. A severe mid-thoracic left convex kyphoscoliosis was diagnosed with a scoliosis angle of 64° and a kyphosis angle of 50° . Tomography showed severe wedging of the corpora with subluxation of T4 on T5 (Fig. 5). There was pencilling of ribs and scalloping of vertebrae. Because of the progressive character of the neurofibromatosis-related kyphoscoliosis an operative stabilisation was proposed. This was several times refused by the parents and their general practitioner. On 1 January 1986, the patient was admitted, at age 22, with almost complete paraplegia. There was little motor function or sensibility left in either leg. Myelography and CT scanning revealed a complete stop at T5 (Figs. 5, 6). No evidence of an intraspinal neurofibroma was seen. The patient was placed in a halo-femoral traction device. Unfortunately the situation deteriorated and the patient developed complete motor paralysis and loss of sensibility in both legs, combined with compromised bladder function. Anterior spinal decompression, corpectomy of T6 and reconstruction of the spine with autologous rib grafts and allogeneous strut grafts were performed on 10 February 1986 as an emergency procedure (Fig. 7). The patient was replaced in the halo-femoral traction and gradually regained some discrete motor function in both legs. Bladder function recovered quite rapidly and was completely restored a few days af-

Fig. 5 Wedging of vertebral corpora and complete stop of contrast at T5

Fig. 6 Myelo-CT scan showing the arrest of contrast continuity in the *bottom right* picture

Fig. 7 Postoperative CT scan showing continuity of contrast, the rib and the strut graft on T6

Fig. 8 Harrington fixation and solid posterior fusion

ter cord decompression. Two weeks later the patient was mobilised in a plaster jacket. One month later a posterior fusion from T4 to T9 with Harrington instrumentation was performed.

Histological examination of the tissue surrounding the excised vertebral body confirmed the neurofibromatosis type I. The patient was kept in a plaster cast for 9 months. Intensive physiotherapy was started as early as possible. At follow-up the scoliosis angle measured 37° with a kyphosis angle of 32°, both between T5 and T8. The fusion looked solid radiographically (Fig. 8). The patient returned to his daily activities with some residual paraparesis and pyramidal symptoms. There was complete restoration of sensibility. He was able to walk without any supporting device.

Discussion

In 1976 Rezaian distinguished at least two different forms of neurofibromatosis-related scoliosis which he called a "moderate form", with a slow onset, and a "severe form", with progressive onset leading to severe kyphoscoliosis and possible paraplegia. He was unable to find definite criteria for differentiating these two forms [8]. Nowadays we recognise two major groups of neurofibromatosis scoliosis: first, a dystrophic (classic) spinal deformity, and, second, a minor form behaving similarly to idiopathic scoliosis [4, 10, 15]. The dystrophic form is usually a short angular kyphoscoliosis located in the thoracic area [2, 3, 10]. The term "kyphoscoliosis" is used when the kyphosis dominates the scoliosis or when it exceeds 49°. This entity is feared for its rapid progression and poor prognosis, especially when there are severe dystrophic changes such as pencilling of ribs, foraminal enlargement, scalloping, rotation and wedging of apical vertebrae [2, 4, 15]. Subluxation or dislocation with rotatory gliding of vertebrae, as seen in case 1, is a rare though severe complication in neurofibromatosis-scoliosis [9, 13, 14]. It is our belief that one should not wait to perform an operative stabilisation of the spine in this condition, even when the degree of scoliosis and kyphosis is still quite modest.

Paraplegia may result from osseous deformities, e.g. angular kyphosis, kyphoscoliosis and vertebral (sub-)lux-

ation, or from an intraspinal tumour, e.g. neurofibroma. Sometimes a combination of both anomalies contributes to the development of paraplegia [6, 15]. In our second case the paraplegia was due to rapidly progressing kyphoscoliosis. There was no evidence of an intraspinal tumour. Winter [14] states that paraplegia due to osseous deformity is seen only in patients with dystrophic kyphoscoliosis, the kyphosis being the main cause of the development of paraplegia [15].

Brace treatment is not effective in dystrophic kyphoscoliosis. Initially we treated the paraplegic boy (case 2) with halo-femoral traction. This is advised by Winter in patients with minor cord compression before operative stabilisation [15]. No traction should be applied to a rigid kyphosis [5]. This may be the reason why the patient in case 2 became worse with the halo-femoral traction. Thus, to avoid initiating permanent paraplegia it is important never to apply a halo traction device to a sharp dystrophic curve. For dystrophic scoliosis, with kyphosis and severe dystrophic vertebrae, a combined anterior and posterior fusion should be carried out. Both our patients were treated this way. Often more than one procedure is necessary. A posterior fusion alone is insufficient and leads to failure [2, 5, 11, 15]. A vascularised rib graft augmentation was used in case 1. Betz et al. [1] suggested doing so whenever possible. Due to the ignorance of the parents and their general practitioner we were not allowed to operate on the second patient before the paraplegia developed. One cannot be persistent enough in trying to convince every party involved of the importance of early operation in neurofibromatosis-related dystrophic kyphoscoliosis. We used the CD instrumentation in the first case and had a satisfying result. The experience with CD instrumentation in neurofibromatosis-related dystrophic kyphoscoliosis is still limited. There are a few publications which report promising results [7, 12]. It is agreed that, in contrast to the management of idiopathic scoliosis, bracing postoperatively remains absolutely necessary for a longer period.

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