NEUROFIBROMATOSIS OF THE CERVICAL SPINE

A REPORT OF EIGHT CASES

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Eight patients with neurofibromatosis presented with symptoms of cervical spine involvement over a period of 17 years, five of them within the second decade of life. The symptoms included neurological deficit in five, a neck mass in four, and deformity in three; only two complained of pain. Osteolysis of vertebral bodies with kyphosis of more than 90° was the most common radiological feature.

Posterior fusion failed in the one patient in whom it was performed. Good results were achieved by anterior fusion, alone, or combined with posterior fusion. Surgical complications included one death in a patient with a malignant neurofibroma, and one case of transient neurological deterioration.

Involvement of the cervical spine in neurofibromatosis has only rarely been reported (Curtis et al 1969; Winter et al 1979; Yong-Hing, Kalamchi and MacEwan 1979; Verbiest 1983), although the spine is the part of the skeleton most commonly affected (McCarroll 1950). The symptoms generally described are pain on neck movement and neurological deficit, but the majority of cases are asymptomatic (Yong-Hing et al 1979). Neurological complications include tetraparesis, hemiparesis and radicular symptoms (Verbiest 1983). The radiological features, which may occur in isolation or in various combinations, are vertebral scalloping, foraminal enlargement, collapse of entire vertebral bodies, loss of the normal lordosis, cervical kyphosis, fixed anterior spondylolisthesis, atlanto-axial rotatory subluxation and atlanto-occipital subluxation. Surgery has been undertaken for spinal-cord or nerve-root compression, atlanto-axial rotatory subluxation, and increasing pain.

The cases reported here showed the additional radiological features of massive spondylophyte formation and dural ectasia of the cervical spine.

PATIENTS AND METHODS

Between 1974 and 1990 we treated eight patients with neurofibromatosis who had symptoms related to the cervical spine (Table I). There were seven men and one woman. Their average age was 26 years (10 to 54); five were aged between ten and 20 years.

The presenting symptoms included neurological deficit, a mass in the neck, deformity and pain. The former was the most common; two patients had quadriplegia, one hemiparesis, one monoparesis of the right arm, and one had paraesthesia, increased muscle tone and brisk tendon reflexes. A mass in the neck was present in four patients. Biopsy showed it to be a malignant neurofibroma in one, a benign neurofibroma in two and a plexiform neurofibroma in one. The last was 15 cm in diameter, sessile, and had to be incised to gain access to the posterior arches of the atlas and axis to perform an arthrodesis. Pain was the presenting feature in two patients and deformity of the neck in three. In one patient pain was the only symptom.

The cutaneous manifestations of neurofibromatosis, including café-au-lait spots and neurofibromata, were seen in all patients. A tibial pseudarthrosis had been successfully treated in one of the patients at a younger age. The only woman in the series had Lisch nodules in her irises. In six patients radiographs of the cervical spine showed deficiencies of one to three vertebral bodies. The resultant anterior bone deficiency produced a kyphosis of more than 90° in five (Fig. 1a); the remaining patient had osteolysis of a single vertebral body, biopsy of which showed a malignant neurofibroma (Fig. 2). One patient had a kyphosis of 15° at the C5 to C6 level without

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Case 2. Cervical kyphosis in a 31-year-old man. Figure 1a - Deficiency of the C6 vertebral body. Figure 1b - There is a sound anterior fusion from C2 to C7 nine years after anterior fusion.

Fig. 1a

Fig. 1b

vertebral body deficiency, but with large spondylophytes on the adjacent vertebral margins (Fig. 3). A 15-year-old boy (case 7) had a gross anterior dislocation of the atlas on the axis (Fig. 4a), suggesting a disruption of the transverse ligament of the atlas. A CT scan confirmed the displacement (Fig. 4b).

In three patients we performed a cervical myelogram; in two no abnormality was noted and in the third there was dural ectasia at C5 to C7 in a spine with

deficient vertebral bodies at C3 and C4 and a kyphosis of more than 90° (Fig. 5).

We performed posterior cervical fusion alone, supplemented by a wire loop, in case 1. Four patients had anterior spinal fusion operations (Fig. 1b), one combined with simultaneous posterior fusion. The patient with the atlanto-axial dislocation was treated by reduction with skull traction followed by a posterior Brooks and Jenkins (1978) type of arthrodesis (Fig. 4c).

Table I. Details of eight patients with neurofibromatosis involving the cervical spine

Case	Age (yr) and sex	Symptoms*			Neural defect	Other features of neurofibromatosis	Radiological findings			Treatment		
		Pain	Deformity	Mass			Vertebral deficiencies	Kyphosis	Myelogram	PSF† (yr)	ASF‡ (yr)	Result
1	37/F	+	+	+	-	Cutaneous Lisch nodules in eyes Thoracic scoliosis	C5, 6, 7	90°	-	1974 1987	1988	Asymptomatic Fused C3 to C6
2	31/M	-	-	-	+	Cutaneous	C6	90°	Normal	-	1981	Asymptomatic Fused C2 to C7
3	54/M	-	-	+	+	Cutaneous	C5	10°	-	-	1985	Died, 8 days, of pneumonia
4	15/M	+	-	-	-	Cutaneous, tibial pseudarthrosis Dystrophic T12 to L1	-	15°	-	-	-	Asymptomatic
5	18/M	-	+	-	+	Cutaneous	C3, 4	90°	Normal	-	1987§	Quadriplegia 1990
6	10/M	-	-	-	+	Cutaneous	C3, 4	90°	Dural ectasia	1987	1987	Monoparesis arm, non-progressive Fused C2 to C5
7	15/M	-	-	+	+	Cutaneous plexiform neuroma, occiput and neck	Atlanto-axial dislocation	5°	-	1987	-	Asymptomatic Fused C1 to C2
8	16/M	-	+	+	-	Cutaneous	C3, 4	90°	-	-	1990¶	Asymptomatic Fused C2 to C5

*+ yes; - no, †PSF, posterior spinal fusion, ‡ASF, anterior spinal fusion, §refused operation, ¶transoral approach

Fig. 2a

Fig. 2b

Case 3. Figure 2a - Osteolysis of C5 vertebral body in a 54-year-old man. Figure 2b - Anterior iliac strut grafting was performed. Histological examination showed a malignant neurofibroma.

Fig. 3

Case 4. A 15-year-old boy with angular kyphosis at C4 to C5 and large spondylophytes at C5 to C6.

Fig. 4a

Fig. 4b

Fig. 4c

Case 7. Figure 4a - The radiograph shows gross anterior atlanto-axial dislocation in a 15-year-old boy with 'drop attacks' and a hemiparesis. Figure 4b - A CT scan further emphasises the displacement. Figure 4c - Stability was achieved by a posterior Brooks-Jenkins wedge compression arthrodesis.

Fig. 5a

Fig. 5b

Case 6. Radiographs of a ten-year-old boy with deficiency of vertebral bodies C3 and C4, retrolisthesis of C4 on C5, and a localised kyphosis of more than 90°. Figure 5a - The dots on the posterior margins of the vertebral bodies and the spinolaminar junctions define the limits of the spinal canal. Figure 5b - The myelogram shows the dural ectasia.

Two patients had no operation. One (case 5) refused the proposed anterior spinal fusion after skull traction had been started. The other patient complained only of pain, had no deformity or neurological defect, and required only conservative treatment (case 4).

RESULTS

The first patient in the series, treated by posterior cervical fusion and a wire loop between the spinous processes of C5 and T1, was found, 13 years later, to have a pseudarthrosis, for which we performed a further posterior surgical repair and removal of the wire loop. One year after this she had an anterior iliac strut graft for a persisting posterior pseudarthrosis. The bleeding during this operation was excessive. Three years later the graft had fused to the anterior vertebral bodies of C3 to C6. Ideally, the fusion should have extended to T1, but the patient has remained asymptomatic.

Three of the anterior spinal fusions including the one combined anterior and posterior fusion, have resulted in solid bony arthrodeses.

The patient with severe anterior atlanto-axial dislocation was first treated by skull traction in extension, an incomplete reduction resulting. Stability was achieved by a Brooks-Jenkins type of posterior atlanto-axial arthrodesis. Two patients were treated postoperatively in a halo body jacket, and both had successful fusion.

Neck pain, in the patient without vertebral body deficiency and with only a mild kyphosis, responded well to conservative treatment, and he was asymptomatic two years later. The patient who refused surgery was readmitted to hospital two years later with quadriplegia (Frankel grade D). He still refused surgical treatment.

Complications. The patient with quadriplegia and a malignant neurofibroma (case 3) died eight days after anterior spinal fusion from bronchopneumonia. The patient with partial paralysis of the right arm developed a complete monoplegia of that limb and weakness of the right leg immediately after simultaneous combined anterior and posterior fusion. The neurological status had recovered to the pre-operative level six months after surgery. This patient's posterior fusion mass was seen to extend to the occiput.

DISCUSSION

It has been said that cervical deformities in neurofibromatosis are found only in young adults (Holt 1978), but five of our cases presented in the second decade of life, two in the fourth and one in the sixth. Yong-Hing et al (1979) have reported a 30% incidence of abnormalities of the cervical spine in patients with neurofibromatosis referred for treatment of spinal curvatures. While all our cases primarily had complaints referable to the cervical spine, three were found to have coincidental deformities in the thoracic and lumbar spine.

Dural ectasia has been previously reported in association with spontaneous dislocation of the thoracic spine (Stone et al 1987). It has been suggested that the spinal cord may escape damage by virtue of the pathological enlargement of the spinal canal. Our patient with dural ectasia had a greatly widened cervical canal at the C5 to C7 levels. The ectasia occurred immediately below a kyphotic deformity of more than 90°, itself due to deficiency of the C3 and C4 vertebral bodies (Fig. 5). He deteriorated neurologically after a combined anterior and posterior cervical fusion procedure and neurological recovery to the pre-operative level took six months.

Atlanto-axial dislocation due to neurofibromatosis has been previously reported in three patients, two of whom had hemipareses (Isu et al 1983), as in our case. Reduction by skull traction and posterior atlanto-axial arthrodesis proved to be successful and there were no complications.

Posterior cervical fusion for a kyphosis of more than 90° was unsuccessful on two occasions in the same patient. In four patients anterior cervical fusion in the presence of severe kyphosis resulted in stable fusions in three; the fourth developed a distal pseudarthrosis but was asymptomatic three years after operation. The patient with severe kyphosis who refused surgery suffered progressive neurological deterioration.

Cervical spine involvement may be asymptomatic, hence the recommendation that radiographic examination of the cervical spine should be done in all patients with neurofibromatosis before general anaesthesia is given for any reason and before applying skull traction for treatment of a scoliosis.

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