Hereditary spinal osteochondromas in diaphyseal aclasia

Abstract We present cases of cervical cord compression in father and son with diaphyseal aclasia. Both patients were investigated with plain radiography, CT and MRI. We believe this to be the first report of spinal cord compression in first-degree relatives with diaphyseal aclasia.

Key words Diaphyseal aclasia · Osteochondroma · Hereditary multiple exostoses

Introduction

We present what we believe to be the first report of spinal cord compression in first-degree relatives with diaphyseal aclasia.

Case reports

Case 1

A 39-year-old man was investigated for progressive weakness of all four limbs, predominantly on the right, over a period of 3 years. Multiple exostoses had been excised from his long bones since the age of 11 years. His son and daughter had both undergone excision of long bone exostoses.

On examination, he had a reduced range of movement of his neck in all directions, reduced power in the right arm and leg with increased biceps and quadriceps reflexes, patellar clonus, and a positive Babinski sign. There was no definite sensory level.

Plain cervical radiographs (Fig. 1) showed a bony lesion which appeared to originate from the posterior elements of C5 and also to involve the posterior elements of C3, C4 and C6. There was apparent fusion of C4/C5 with loss of disc height. Whether this was congenital, or a result of the disease process, remains unclear. Axial CT (Fig. 2) showed a large, irregular bony lesion which arose from the C5 lamina on the right and involved the lamina of C4 on this side. There was also involvement of the spinous processes of C3 and C6. There was maximum encroachment into the spinal canal at C5, with significant reduction of the anteroposterior diameter. A lesser degree of canal narrowing was seen at C4. T1- and T2-weighted MRI confirmed considerable encroachment into the spinal canal producing marked cord compression at the level of C5, extending to the lower border of C4 and the upper border of C6 (Fig. 3). On T1 weighting the lesion returned high signal, thought to represent fatty marrow within it.

Total excision of an exostosis, which appeared to arise from both the C4 and C5 laminae on the right, was performed. The lesion was variable in consistency and noted to contain a central fatty marrow component, confirming the MRI findings. Histology demonstrated a benign osteochondroma.

The patient failed to make significant improvement following the operation. MRI of the cervical spine 2 years postoperatively showed no evidence of tumour recurrence. There was, however, myelomalacia with marked attenuation of the cord in its anteroposterior diameter as a result of prolonged spinal cord compression.

Case 2

The 15-year-old son of case 1 presented 18 months later complaining of progressive right arm weakness over 10 months following a fall from a trampoline. Past history included removal of a long bone exostosis.

He had reduced lateral rotation of his neck to the right, reduced power in his right arm, with an increased triceps reflex. There was no sensory loss.
Fig. 1 Case 1. Lateral cervical spine radiograph showing a bony lesion which appears to arise from the posterior elements of C5, and to involve the posterior elements of C3, C4 and C6. There is also apparent congenital fusion of C4 and C5, with loss of disc height and dystrophic calcification. The body of C4 has an abnormal shape.

Fig. 2 Case 1. Axial CT at the C5 level showing a large, irregular bony lesion arising from the lamina of C5 on the right. This involved the lamina of C4 on the same side, and the spinous processes of C3 and C6. There is marked encroachment on the spinal canal, maximal at C5. The mass is predominantly of low density, compatible with fat.

Fig. 3 Case 1. T1-weighted sagittal MRI (TR 500, TE 32 ms) showing a bony lesion at C5 encroaching on the spinal canal and producing marked cord compression at the level of C5; it extends up to the lower border of C3 and down the upper border of C6. The lesion returns high signal, consistent with fatty replacement of marrow; this was confirmed at operation. The dysplastic disc at C4/5 returns low signal, and the abnormal shape of the C4 body is confirmed.

Cervical spine radiography was normal. CT showed a bony lesion, 1 cm in diameter, arising in the midline from the posterior elements of C3, extending anteriorly and producing marked reduction in the anteroposterior diameter of the spinal canal (Fig. 4).

MRI (T1-weighted and 2D FLASH) demonstrated a low signal lesion consistent with compact bone, indenting the theca at the level of C3 and C4, producing spinal cord compression (Fig. 5).

Laminectomy of C3 and C4 was performed, with total excision of the bony lesion. Histology confirmed this to be a benign osteochondroma arising from the posterior elements centrally. Postoperatively the patient made a good recovery with complete resolution of neurological signs.

Discussion

Diaphyseal aclasia (hereditary multiple exostoses) is the commonest cause of skeletal dysplasia [1]. It is characterised by anomalous skeletal development, with exostoses usually affecting the long bones around major joints. It can occur sporadically, but is most commonly hereditary, showing autosomal dominant inheritance with variable penetrance. It is more common in males with a male-to-female ratio of 2:1 [1]. Presentation is most commonly in the second decade. Characteristic features are due to stunted bone growth and include short stature in two thirds of cases; bowing of the radius with ulnar deviation (Madelung's deformity); subluxation of the radiohumeral joint; and valgus deformity of the knee [1]. Chondrosarcomatous change is reported in 10% of patients [2].

Spinal osteochondroma causing cord compression in patients with diaphyseal aclasia is rare. To our knowledge this is the first report of first-degree relatives, although thoracic cord compression due to chondrosarcoma in two cousins has previously been reported [2].

The incidence of spinal osteochondromas in diaphyseal aclasia is 9%. Solitary osteochondromas affecting the spine are less common, and are reported to occur in 1–4% of cases. However, as these lesions present only with the onset of neurological symptoms their true incidence is probably underestimated. Half the reported cases were of osteochondromas in the cervical spine, particularly at the level of C2, and most commonly arising from the posterior elements [3]. Neurological sequelae of spinal osteochondromas include cervical myelopathy; sudden tetraplegia [4]; sudden death due to an osteochondroma of the odontoid [5]; occlusion of the left vertebral artery [6]; and Horner's syndrome [7].