A rare cervical spine abnormality associated with neurofibromatosis

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(Index words: Posterior dislocation of C3 vertebra, excision of body of C3 vertebra, fixation of C2 - C4 vertebrae)

Abstract

Spinal abnormalities such as kyphoscoliosis and vertebral scalloping are frequent occurrences in type 1 neurofibromatosis (NF1). We report this rare case of posterior displacement of C3 vertebral body into the spinal canal causing severe cord compression associated with neurofibromatosis in a 13-year old girl. She underwent anterior excision of C3 vertebral body, bone graft and fixation of C2- C4, with improvement of her symptoms.

Introduction

Neurofibromatosis type 1 is a complex disorder frequently (30%-50%) associated with skeletal

abnormalities such as scoliosis, kyphosis and vertebral scalloping. However, there are no reports of posterior displacement of cervical vertebral bodies in such patients. We describe a patient with such an abnormality causing severe cord compression.

Case history

A 13-year old schoolgirl presented with gradually progressing difficulty in walking with numbness in all four limbs of 3 years' duration. When she was seen in the neurosurgical ward she needed support to stand and to walk, and was incontinent of urine. On examination, there was a single neurofibroma in the back measuring 0.5cm and three *café au lait* spots, one on the body and two in

(A)

(B)

Figure 1. Appearance of patient's cervical spine before (A) and after (B) surgery

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the limbs. Muscle power was grade 4 in all four limbs, with a sensory level of C4 to touch and pain, associated with exaggerated reflexes. There was a strong family association of multiple neurofibromatosis involving 6 members of her mother's family, including the patient's mother.

Xray images (Figure 1) showed significant posterior displacement of C3 vertebral body into the spinal canal. MRI images confirmed this, and severe cervical cord compression needing decompression.

The C3 vertebral body was excised by an anterior approach with cervical cord decompression, followed by reconstruction using autologous iliac bone grafts and internal fixation of C2 and C4 vertebral bodies with a plate and interlocking screws (Figure 2). She recovered from surgery uneventfully with improvement of her symptoms.

Discussion

Neurofibromatosis type 1 is an autosomal dominant disorder affecting 1 in 3000 people [1, 2]. About 30%-50% of patients with NF1 have associated skeletal lesions [1, 2]. Among these, scoliosis, kyphosis, thinning of long bone cortex, erosive bone defects, pseudoarthrosis and bone cysts are common [1, 2].

The manifestations in the cervical spine are dystrophic changes in the vertebral body, or pathologic alignment. The most common abnormality observed is a severe cervical kyphosis, which in itself is highly suggestive of the disorder. Dislocation of C1-2 has also been described [2], but there are no previous reported

cases of posterior displacement of a single vertebral body as seen in this patient.

Although plain radiographs may be sufficient for evaluating skeletal lesions, CT and MRI scans are preferred for spinal or intracranial lesions to identify the effects of the anomaly on the nervous system [2].

Previous studies have reported on the effectiveness of anterior correction as opposed to posterior, and the combined approach, to diminish the degree of deformity and achieve solid arthrodesis. A simultaneous anterior-posterior approach for the surgical treatment of severe dystrophic kyphoscoliosis in neurofibromatosis type 1 is better, since it provides proper visualization of the spinal cord, and achieves solid arthrodesis. Since cord compression was mainly anteriorly in we approached the spine in this patient anteriorly, preventing a major surgical procedure, but achieving a stable arthrodesis with spinal mobility. Although the primary pathology of neurofibromatosis is untreatable, we were able to achieve a useful outcome in the patient with improvement of her symptoms.

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