Case Report and Review of the Literature

Pathological sacral fracture in type 1 neurofibromatosis

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Introduction

Neurofibromatosis (NF) type 1 is one of the most common neurocutaneous disorders. This autosomal dominant genetic disorder is due to an abnormality on chromosome 17[1] that leads to abnormal neural crest cell proliferation. Neurofibromas account for 16-30% of all spinal tumours,[2] occurring most commonly in the thoracic region, followed by cervical and lumbar region. Only 1-5% occur in the sacral region. We report a sacral fracture due to a neurofibroma in a child with type 1 neurofibromatosis.

Case report

An 8-year-old male presented with pain over his sacrum one week after falling directly onto his buttock. Prior to the fall the child and the parents noted a deformity for the past two years over his sacrum that was associated with occasional discomfort but they did not seek medical attention. He is a known asthmatic for six years, well controlled on an inhaler.

The child was clinically well and presented with a normal gait. The cutaneous features of NF type 1 were striking, and included both café au lait spots and freckling. His uncle also has similar skin lesions. The child had no other clinical features of NF type 1. The systemic examination was normal.

He had a kyphotic deformity over the upper sacrum without overlying skin abnormality. The deformity at S2/S3 level was tender to palpation and associated with minimal soft tissue swelling. The examination of the rest of the spine, sacroiliac joints and hips were normal. He had no neurological deficit and the rectal examination was normal.

The X-rays revealed an acute kyphotic deformity at S3/S2 and scalloping of the bodies of S2 and S3 anteriorly and posteriorly *(Figure 1)*. The sagittal and coronal reconstructed CT of the lumbosacral area showed a fracture of S3, anterior and posterior scalloping with constriction of the vertebral body, posterior scalloping of S4 and an enlarged sacral foramen on the left between S1/S2 and S2/S3 *(Figure 2)*. The axial and sagittal MRI of the lumbosacral area revealed a sharp kyphosis of the sacrum at S3, a defect of the lower border of S3, anterior and posterior scalloping with constriction of the vertebral body and posterior scalloping of S4 with no evidence of a neurofibroma or a meningocele *(Figure 3)*. The haematological investigations were non-contributory.

The cutaneous features of NF type 1 were striking, and included both café au lait spots and freckling

In view of persistent pain a biopsy and stabilisation were recommended to the parents. A posterior midline approach was used and the deformity at S2/S3 was due to a pathological fracture. Surgery further revealed a large defect anteriorly and posteriorly in the left sacrum with thickened and pale S2/S3 nerve roots and a small soft tissue lesion at the fracture site. Biopsy was taken from the site of the fracture and the surrounding soft tissue. Stabilisation of the fracture was abandoned due to the tenuous nature of the bone. The patient made an uneventful post-surgery recovery.

Histology of soft tissue revealed spindle cell proliferation, representing part of a neurofibroma. The bone histology showed haemorrhage with fracture site reaction.

At eight-month follow-up the patient was asymptomatic. The deformity is static and the fracture has not united.

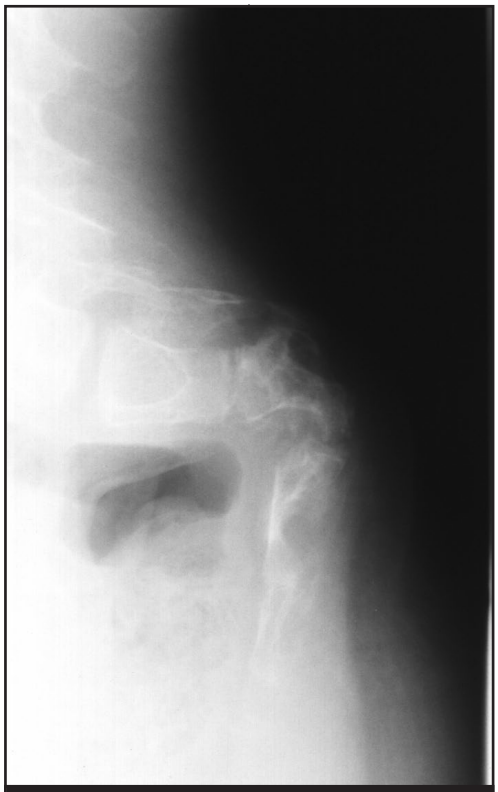

Figure 1: Lateral radiograph of sacrum showing fracture at S2/S3 with dystrophic features

Discussion

Neurofibromatosis type 1 (NF1) has a set of accepted diagnostic criteria,[3] and requires two of seven positive features. Café au lait spots are often the first manifestation and occur in more than 90% of NF1 patients. Freckling in non-sun-exposed areas is also a frequent feature of childhood NF1. Spinal involvement in NF1 is common and may manifest as a kyphosis, scoliosis or kyphoscoliosis. Scoliosis occurs in 10-60% of cases.[6]

The suggested causes of spinal deformity include erosion or pressure by neurofibromas, mesodermal dysplasia, osteomalacia, and endocrine disturbances.[4,5] NF1 spinal deformities encompass dystrophic and non-dystrophic vertebral body changes. Modulation is unique to spinal NF1 and is referred to as a deformity in evolution, as demonstrated in our patient. There is a distinct possibility that it occurs across the spectrum of dystrophic and non-dystrophic curves without a consistent pattern.[7]

Focal kyphoscoliosis due to neurofibroma is described only in the thoracic spine. Spondylolisthesis in NF1 is very rare and is thought to be due to pathologic elongation and erosion of the pedicles or pars interarticularis by foraminal neurofibromas or dural ectasia.[8] Lumbosacral dural anomalies occur in NF1 with considerable morphological variation.[9] Sacral involvement in NF1 is rare, and a pathological fracture due to a dystrophic spinal lesion has not been described.

Pelvic neurofibromas as found in our patient are rare, even in association with NF1, and the overwhelming majority of sacral nerve sheath tumours are Schwannomas.[10] This may be due to the silent nature of these tumours, as seen in our case, that would not have presented to us if he did not sustain a pathological fracture. Neurofibromas may grow to a considerable size and cause poorly differentiated pain and some sacral nerve numbness with only occasional urinary symptoms.[11]

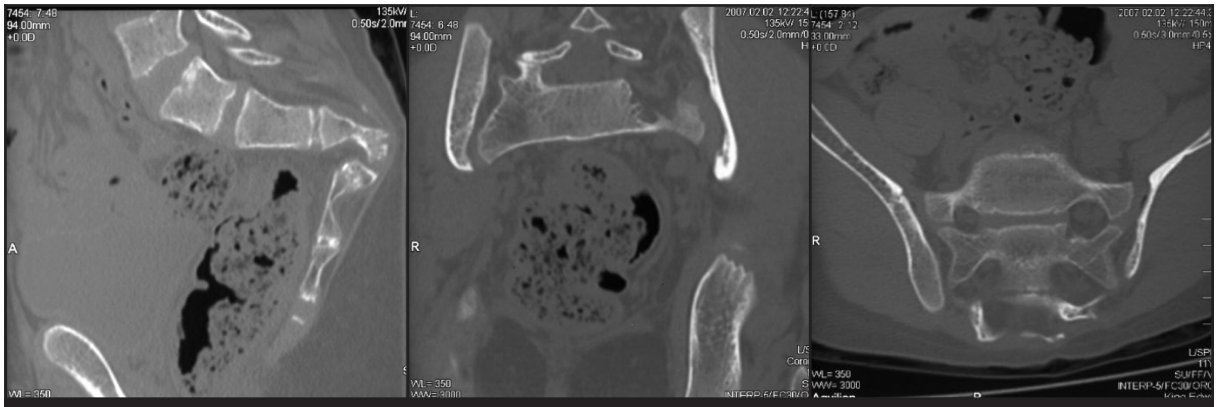

Figure 2: CT sagittal axial coronal
Shows pathological fracture with enlarged neural foramina on the left side

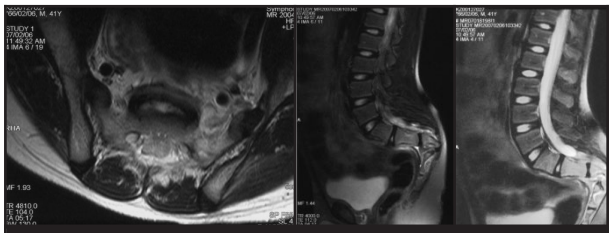

Figure 3: MRI axial sagittal T1 and T2 Demonstrates a sharp kyphosis of the sacrum at S3, a defect of the lower border of S3, anterior and posterior scalloping of S3 and S4, with no evidence of a meningocele

Spinal neurofibromas are difficult to differentiate from neurosarcomas and rare dumb-bell tumours such as meningiomas, ganglioneuroma and haemangioendothelioma.[12] CT is a useful modality and is said to display both intra- and extradural components of tumours as well as skeletal changes,[13] although we could not find any evidence of the intra- or extradural components on the CT in this case. Retroperitoneal plexiform neurofibromas have a characteristic appearance on CT scans. They are typically bilateral, symmetric, low-attenuation masses in a parapsoas or presacral location. Asymmetry in size and attenuation of a larger mass suggests the possibility of a malignant tumour of the nerve sheath.[14] MRI may demonstrate a discrete central low intensity focus on T1-weighted image with contrast (Dot-sign) and is characteristic of neurofibroma or Schwannoma.[15]

Transverse fractures of the sacrum due to trauma are exceptional in children. These high energy fractures occur more commonly at the S1/S2 level and are associated with neurology, in contrast to our patient.[16,17] The absence of neurological deficit in our patient may be attributed to the low energy transfer of the injury, lower level of the injury and the dystrophic nature of S2, S3 and S4 resulting in enlarged neural foramina. The high incidence of neurological injury described in sacral fractures may thus not be applicable if they are pathological.[18]

The incidence of delayed and non-union in patients with NF1 presenting with fractures is not reported. Associated pseudarthrosis, specifically of the tibia, is well known in NF1 and classified by Boyd *et al*.[19] Although the underlying pathology in our case is the neurofibroma and no pre-injury radiology was done, the ensuing pseudarthrosis may be comparable to a Boyd type II, and we expect similar poor outcome in terms of union. Luckily, union in our case may not be essential for a good outcome.

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