MR findings of fibrodysplasia ossificans progressiva complicated by acute cord compression: Case report and literature review

Nathalie V. Gebara, MD; Gordon D. Heller, MD; C. Douglas Phillips, MD; and Vaios Hatzoglou, MD

We report a case of fibrodysplasia ossificans progressive in a 34-year-old male who presented with bilateral lower-extremity numbness. This rare disease results in abnormal ossification of various muscles, tendons, and ligaments. In this case, there was ossification of the posterior longitudinal ligament and resultant thoracic spinal-cord compression. Recognizing the MR features of this disease and this serious complication is very important in the management of these complex patients.

Introduction

Fibrodysplasia ossificans progressiva (FOP) is a very rare and disabling genetic disorder characterized by progressive heterotopic ossification of skeletal muscle and connective tissue. The disease has no sexual, racial, or ethnic predilection (1). Recently, the gene for FOP has been mapped to human chromosome 4q 27-31 (2). Characteristic radiographic features have been described primarily with plain film radiography (3). In this case report, we discuss the MR features of this disorder resulting in acute spinal cord compression.

Case report

A 34-year-old male with a known history of FOP presented with bilateral lower-extremity numbness. He reported no recent history of trauma and no other symptoms. Plain radiographs of the thoracic and lumbar spine (Fig. 1) demonstrated ossification of multiple intercostal and paraspinal muscles in sheet- and ribbon-like configurations. MRI of the entire spine without contrast was performed one week later. These images (Fig. 2) revealed extensive...
MR findings of fibrodysplasia ossificans progressiva complicated by acute cord compression

Discussion

FOP is a rare genetic disorder that can lead to significant, progressive immobility. First described nearly 200 years ago, this disease is characterized primarily by heterotopic ossification of soft tissues throughout the body and malformations of the great toes (present at birth). The prevalence is approximately one in two million worldwide. Most cases result from a spontaneous mutation, but autosomal dominant transmission has been described (3).

Patients typically present clinically within the first decade of life with inflammatory soft-tissue swelling that is later replaced by heterotopic ossification, classically in ribbon- and sheet-like configurations (4). FOP is both episodic and progressive and can be triggered by minor trauma or otherwise trivial surgical procedures. Consequently, patients become increasingly immobile and often wheelchair-bound by adulthood (3).

Although the disease can affect muscles, tendons, and ligaments throughout the body, the tongue, diaphragm, esophagus, extra-ocular, and cardiac muscles are characteristically spared (5). In the classical form of FOP, for which a gene mutation has been identified, developmental anomalies of the cervical spine frequently manifest as enlarged posterior elements, tall narrow vertebral bodies, and fusion of the facet joints between C2 and C7 (3). Additional skeletal manifestations have been described, including deformities of the femurs as well as osteochondromas in the medial proximal tibias (4). To our knowledge, no reports have described the MRI findings of the specific complication of spinal-cord compression resulting from a heterotopic ossification of interspinous ligaments in these patients.

Although the majority of these patients are identified early in life, it is important that radiologists are able to recognize both the radiographic appearance and possible complications of this disease in adults. For example, traditional management of patients with acute spinal-cord compression resulting from degenerative disease, trauma, or neoplasm may involve emergent surgical decompression, possible fusion, or other surgical intervention. In FOP, surgical attempts at removing sites of heterotopic ossification often result in explosive and painful bone formation at the operative site, often worsening the condition (6). Accurate diagnosis and careful management of these patients is therefore critical in avoiding these unfortunate and often debilitating consequences.

References

1. Mahoubi S, Glaser DL, Shore EM, Kaplan FS. Fibrodysplasia ossificans progressiva. Pediatric Radiology. 2001; 31(5):307-14. [PubMed]
2. Shore EM et al. A recurrent mutation in the BMP type 1 receptor ACVR1 causes inherited and sporadic fibrodysplasia ossificans progressiva. Nature Genetics. 2006; 38(5): 525-527. [PubMed]
3. Cremin B, Connor J, Beighton P. The radiological spectrum of fibrodysplasia ossificans progressiva. Clinical Radiology. 1982; 33(5):499-508. [PubMed]

Figure 2. (A) Axial T1- and (B) T2-weighted images at the L4 level demonstrate multiple heterotopic ossifications (arrows) in the paraspinal musculature on the left.

Ossifications of the paraspinal musculature and ligamentum flavum, which demonstrated signal characteristics of bone. Additional MR and CT (Fig. 3) imaging of the thoracic spine revealed ossification of the ligamentum flavum at the T9/10 level, resulting in spinal-cord compression and spinal-cord edema. The patient was referred to neurosurgery for T9-10 laminectomy and spinal-cord decompression.
Figure 3. (A) Sagittal T1-weighted and (B) T2-weighted MR images of the thoracic spine demonstrate hypertrophy (arrows) of ligamentum flavum at the T9-10 level resulting in narrowing of the canal and compression of the cord at this level. (C) Corresponding sagittal CT reconstruction confirms the presence of heterotopic ossification (arrow) of the ligamentum flavum at this level.

4. Diermengian GK, Hebela NM, O’Connell M, Glaser DL, Shore EM, Kaplan FS. Proximal tibial osteochondromas in patients with fibrodysplasia ossificans progressiva. *The Journal of Bone and Joint Surgery*. 2008; 90:366-74. [PubMed]

5. Kaplan FS, Strear CM, Zasloff MA. Radiographic and scintigraphic features of modeling and remodeling in the heterotopic skeleton of patients who have fibrodysplasia ossificans progressiva. *Clinical Orthopaedics*. 1994; 304: 238-247. [PubMed]

6. Caron KH, DiPietro MA, Aisen AM, Heidelberger KP, Phillips WA, Martel W. MR Imaging of early fibrodysplasia ossificans progressiva. *The Journal of Computer Assisted Tomography*. 1990; 14(2):318-21. [PubMed]