

VERTEBRAL COLLAPSE AND CORD COMPRESSION IN A YOUNG ADULT WITH PAGET'S DISEASE OF THE SPINE

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HISTORY AND PHYSICAL EXAM

A 38-year-old Caucasian male presented to our institution with a four year history of intermittent low back pain. There was no trauma or inciting event and his pain progressed over the course of months to radiate to both of his lower extremities. Initially he was self-treated with conservative measures such as physical therapy, ergonomic chairs, and massages. He was capable of going to work, playing golf, working out at the gym, and was able to sit for prolonged periods of time at work with ibuprofen. His associated symptoms included a ten pound weight loss over the preceding months and a progressive kyphosis. He denied fevers, chills, night sweats, headache, visual/auditory changes, bowel or bladder dysfunction and fatigue. History was negative for environmental exposures (radiation, asbestos, etc).

On examination, his thoracolumbar junction was non-tender and kyphotic with a compensatory increase in lumbar lordosis. There was no atrophy of the lower extremities and no sensory deficits in the lower extremities. Dorsalis pedis pulses were palpable and equal bilaterally. Babinski's reflex was absent on the right and upgoing on the left. There were hyperreflexic patellar and Achilles reflexes, no clonus, and a negative straight leg raise.

Initial work-up included plain radiographs which revealed a destructive process at T12 and L1 with severe compression deformity and loss of vertebral height. There was 30 degrees of kyphosis on the lateral radiograph. A grade I listhesis to the right at the thoracolumbar junction and retropulsion of fracture fragments into the spinal canal were also visualized. CT scan showed pedicle destruction and eccentric soap bubble appearance of the involved levels with posterior element sparing (Fig. 1).

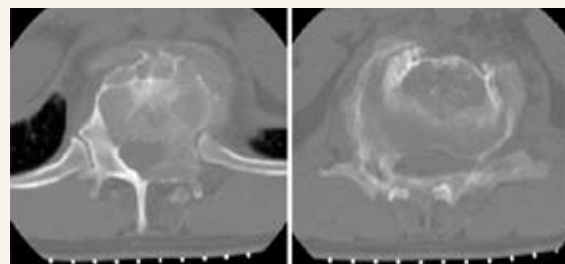


Figure 1:
Axial CT scan images showing soap-bubble appearance of pagetic bone with invasion of paraspinal muscles and intervertebral disk.

An MRI with gadolinium was obtained which confirmed a destructive process involving T12-L1 causing spinal stenosis and cord compression from T11 to L2 and involving the associated paraspinal soft tissues (Fig. 2). A CT guided transpedicular biopsy was performed of T12 which showed fragments of woven bone with reactive changes and was negative for malignant cells. A joint fluid sample was obtained from the T12/L1 disc space which was negative on gram stain, fluid culture, and acid fast smear. His alkaline phosphatase was elevated (249 IU/L).



Figure 2:
Sagittal T2 and T1 weighted images of T12-L1 showing L1 vertebral collapse and cord compression.

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HOSPITAL COURSE

The patient was taken to the operating room for vertebrectomy of T12-L1, anterior cage replacement of T12 and L1 and fusion with rib autograft. Intra-operatively, a large soft tissue mass was visualized covering the spine on the left side of the vertebral body which appeared to be bony callus. This was resected and sent to pathology. There was severe collapse of the T12 and L1 vertebral bodies with bone and disc retropulsed against the conus. The fractured vertebrae were gently elevated off the spinal cord, resected, and a thorough decompression

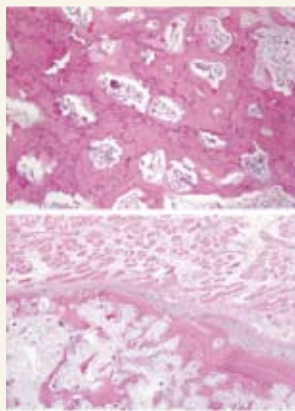


Figure 3:
H&E staining of T12 tissue biopsy demonstrating classic “mosaic pattern” of disorganized pagetoid bone, loss of organized lamellar structure, and osteoclast-like giant multinucleated cells. b) disorganized blastic activity with extension through cortical surface into adjacent paraspinal muscles, new bone/osteoid formation and reactive changes causing mechanical instability and compression fracture.

was performed. There were also extensive adhesions to the posterior longitudinal ligament by calcified pagetic tissue. On post-operative day #2 he was taken back to the OR for posterior fusion from T10 to L3 for his kyphotic deformity. Post-operatively the patient reported neurological improvement.

The histopathological diagnosis revealed characteristic “mosaic” pattern of disordered bone formation and osteoclast-like giant multi-nucleated cells characteristic of Paget’s disease (Fig. 3).

DISCUSSION

Osteitis deformans or Paget’s disease of bone (PD) is a common disorder of abnormal bone homeostasis in the elderly population. PD is generally diagnosed by exam, serum alkaline phosphatase (AP) levels, plain radiographs, and bone scintigraphy and treated medically¹. The prevalence of PD increases with age and few reports exist in the literature of severe spinal deformity secondary to PD in persons under age 40. Here we report a case of focal PD at the thoracolumbar junction causing progressive deformity, mechanical overload of the T12 and L1 vertebral bodies and cord compression. PD uncommonly presents as pathological vertebral compression fracture causing myeloradiculopathy and requiring acute decompression. Despite the high prevalence of PD in the geriatric population, isolated and symptomatic pagetic lesions of the spine are rare to occur in young adult patients.

PD is the second most common disorder affecting bone after osteoporosis² and the lumbar (58%), thoracic (45%), and cervical (14%) spines are frequently involved³. The overall prevalence of PD in the United States has been estimated to be between 1 and 2% or approximately 2.5-5 million people. The adult onset form of PD (distinct from juvenile PD) increases in prevalence with age and rarely becomes symptomatic in a young healthy adult⁴.

Maintenance of skeletal homeostasis in normal bone is performed by osteoclastic resorption of bone balanced by osteoblastic activity. The pathophysiology of PD is characterized by focal abnormalities in the balance of bone turnover with increased bone resorption by the osteoclast (an increase in osteoclast number, size, and activity)⁵. The chronic and progressive nature of PD eventually results in the replacement

of normal bone with new disorganized bone that is structurally inferior and prone to pathological fracture as in the present case.

An estimated 70% of radiographically visible pagetic disease sites remain clinically silent³. The majority of patients with PD are asymptomatic and only approximately 30% experience symptoms^{3,6}. PD is diagnosed by exam, serum alkaline phosphatase (AP) levels, plain radiographs, and bone scintigraphy in the majority of patients. Alkaline phosphatase, a blood marker of osteoblastic activity, and urinary N-telopeptide, a marker of osteoclastic activity released during bone resorption in PD are reflective of this increased cellular turnover. Both have been reported to be elevated from 10-20 fold in patients with Paget’s disease⁷. In the present case the AP was elevated 3-6 fold (249 IU/L).

Medications such as bisphosphonates and calcitonin are the mainstay of treatment for PD and have been successful in treating the disease in a variety of settings including spinal stenosis⁸ and cauda equina syndrome⁹. The proven efficacy of pharmacological therapy for pagetic spinal stenosis¹ makes surgical intervention a second line treatment after failure of anti-pagetic medicines. In the present case of vertebral collapse and instability causing myeloradiculopathy, acute operative intervention was necessary.

PD of the spine has been reported to cause spinal stenosis with extradural ossification and involvement of the soft tissues^{10,12} and invasion of the intervertebral disk¹³. In the case described that presented to our institution, there was seen to be a large bony callus that eroded both into the adjacent soft tissues and into the intervertebral disk. Decompressive laminectomy¹⁰ and percutaneous vertebroplasty¹¹ have been described in the setting of spinal PD. The present case involves a patient with pagetic involvement of the T12 and L1 vertebral bodies with invasion of the paraspinal tissues and vertebral collapse causing retropulsion onto the conus. The lack of a clear histopathological diagnosis by CT-guided transpedicular biopsy and the local invasion seen on imaging studies merited an open biopsy to rule out a more sinister lesion. Operative vertebrectomy, decompression, and anterior cage placement was necessary to alleviate impingement on the conus and to stabilize the thoracolumbar junction. The vertebral instability and kyphotic deformity required fusion from T10 to L3.

The etiology of PD is unknown. Genetic factors and infectious agents have both been proposed to play a role. PD is often seen to cluster in families and in certain geographic areas an estimated 15% of affected individuals have at least one other family member affected¹⁴. Pagetic osteoclasts contain nuclear inclusions seen on electron microscopy that are thought to represent the molecular fingerprints of the nucleocapsids of paramyxoviruses¹⁵ suggesting a viral etiology, however an infectious agent has yet to be isolated.

Malignant degeneration has been shown to occur in pagetic lesions with approximately 1% progressing to osteosarcoma (a several thousand-fold greater risk than the general population)^{16,17}. There is evidence to suggest an association

exists between PD and osteosarcoma that may be the result of a single common gene on chromosome 18q or two tightly linked genes that undergo concomitant mutation¹⁸. Careful histologic inspection is therefore warranted in cases of PD to rule-out sarcomatous degeneration.

Few reports exist in the literature of isolated and severe spinal deformity secondary to PD in a young healthy person. Here we present a focal and advanced case of PD in a 38 year-old man causing pathologic compression fracture and cord compression at the thoracolumbar junction. Interestingly, younger patients with PD are more likely to have spinal involvement. A significant difference in patients <40 and in those >40 has been observed in the incidence of pagetic lesions in the thoracic (44% vs. 14%, respectively) and lumbar spines (50% vs 29%, respectively)¹⁹. In patients diagnosed with PD at age 70, subclinical pagetic lesions are estimated to be present before the age of 30 in 64%²⁰, however diagnosis is rarely made this early in life.

The present case demonstrates a rare clinical occurrence of a young healthy man with an advanced form of PD causing vertebral collapse that required acute spinal decompression and open biopsy. Lesions present in this younger age group are often subclinical and represent a diagnostic and therapeutic challenge to the orthopaedic surgeon. Early diagnosis and treatment of PD is necessary in this age group to prevent advanced disease presentations and to avoid neurological and mechanical sequelae.

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