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Osteopetrosis – Classic Imaging Findings in the Spine

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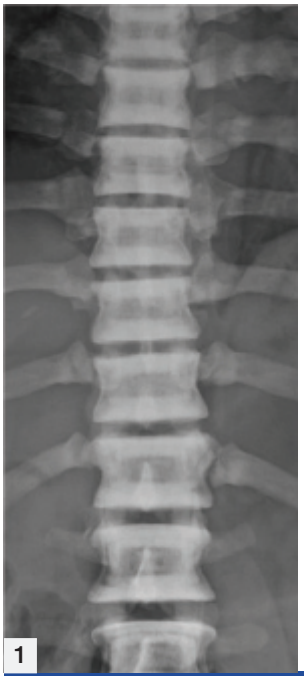
A 27-year-old man presented to his primary care clinic with chronic, progressive mid to lower back pain. Neurologically, the patient was normal with no focal deficits. A radiographic examination of the thoracic and lumbar spine was performed [Table/Fig-1,2], demonstrating diffuse endplate sclerosis with a sharp delineation between the peripheral bony sclerosis and relative lucency of the central vertebral bodies, producing the characteristic "sandwich vertebrae" of osteopetrosis. In addition, several lumbar vertebrae demonstrated sclerosis along the inner margin of the remainder of the vertebral bodies, resulting in "bone within a bone" appearance, also characteristic of osteopetrosis.

Osteopetrosis is an inherited disease with a variety of genetic causes related to dysfunctional bone remodeling, which results in abnormally dense bones. Although dense, the involved bones are weakened and prone to pathologic fractures. There are many sub categories of osteopetrosis; however, they are largely divided into two main groups Autosomal recessive (ARO) and Autosomal dominant (ADO). The autosomal recessive subtype is typically severe, resulting in either fetal demise or discovered early in infancy with pathologic fractures, neurologic deficits, or bone marrow abnormalities. The autosomal dominant form of osteopetrosis (Albers-Schönberg disease), on the other hand, is less severe and often discovered in young to middle aged adults. Patients may be asymptomatic with incidental findings on radiographic examinations or present with fractures, scoliosis, arthritis, delayed healing, or osteomyelitis. There are two phenotypic variants of ADO. Subtype 1 (ADO1) is rare and secondary to a defect of the low density lipoprotein receptor 5 (LRP5), which results in increased uniform sclerosis [1]. This subtype primarily involves the cranium. Subtype 2 (ADO2) is far more common and secondary to a chloride channel 7 (CLCN7) deficiency [1]. Radiographically, ADO2 presents with "bone within a bone" pattern most often within the pelvis and long bones and less frequently within vertebrae, as well as the characteristic "sandwich vertebrae" appearance in the spine, as seen in this case [2]. Current treatment for ADO is primarily supportive, though, it is hypothesized that gene therapy may play a role in the future with an identifiable gene defect [3].



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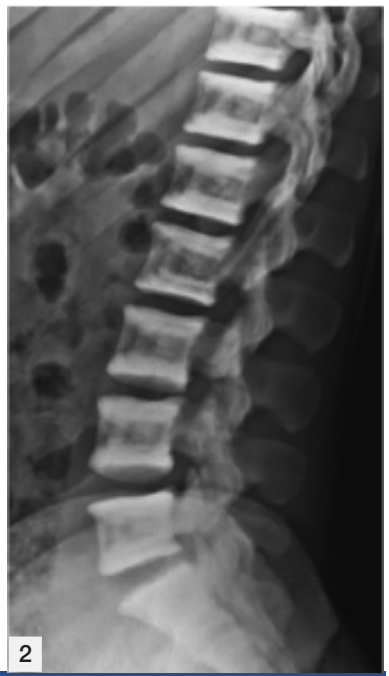

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[Table/Fig-1]: Lower thoracic spine frontal view: diffuse vertebral endplate sclerosis with well-demarcated margins between the peripheral sclerotic and central relatively lucent bone, consistent with a "Sandwich vertebrae" appearance. The visualized lumbar vertebrae also demonstrate a "bone within a bone" appearance secondary to sclerosis along the inner margins of the lateral aspect of the vertebral bodies, along with the endplate sclerosis
[Table/Fig-2]: Lumbar spine lateral view: similar findings are demonstrated with both the "Sandwich vertebrae" and "bone within a bone" appearance

The "Sandwich vertebra" appearance, while classical for osteopetrosis, bears similarity to vertebral sclerosis patterns seen in other disease processes. Hyperparathyroidism is the primary differential consideration for this appearance in the spine, as both processes cause endplate sclerosis. In contrast to osteopetrosis, the demarcation between the peripheral sclerotic and central lucent bone is indistinct or smudgy with hyperparathyroidism; this pattern is colloquially referred to as the "Rugger-jersey" spine, which should be readily distinguished from the "sandwich vertebrae" appearance on imaging [4]. In radiographically equivocal cases, a clinical work-up for hyperparathyroidism establishes the correct diagnosis. Paget's disease affects middle aged to elderly adults and may also result in endplate sclerosis, along with other characteristic imaging features. Plain radiographs demonstrate bony expansion with peripheral cortical and central trabecular thickening, resulting in a characteristic "picture frame" appearance of the vertebral bodies, as well as diffuse bony sclerosis [4,5]. This differs from the "bone within a bone" appearance of osteopetrosis, which typically lacks bony, cortical, or trabecular thickening. Patient age and imaging appearance are distinguishing features of Paget's disease.

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"The views expressed in this material are those of the authors, and do not reflect the official policy or position of the U.S. Government, the Department of Defense, or the Department of the Air Force."

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