Osteopetrosis: "Sandwich Vertebrae"

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An otherwise healthy woman in her mid-20s presented with low back pain after lifting heavy packages from her car. Physical examination revealed normal strength, sensation, and reflexes in bilateral upper and lower extremities. Palpation of the spine revealed paraspinous spasm and tenderness but no pain with direct percussion of the spinous process. Lumbar spine x-rays (Figure) showed sharply demarcated bands of sclerosis abutting the endplates of all imaged vertebrae. This appearance, called "sandwich vertebrae," is pathognomonic for osteopetrosis and should be distinguished from the "rugger jersey" spine of renal osteodystrophy, which has ill-defined bands of sclerosis with a more gradual transition from sclerotic to osteopenic bone. A pelvic x-ray showed diffuse symmetric bony sclerosis, while dual-energy x-ray absorptiometry yielded strikingly high T scores—5.5, 8.7, and 4.9 for the lumbar spine, femoral neck, and distal radius, respectively. Serum tartrate-resistant acid phosphatase (TRAP) level was grossly elevated at 23. These results, particularly elevated serum TRAP and presence of sandwich vertebrae, are diagnostic for osteopetrosis and obviated the need for a biopsy. An incidental finding of a persistent anterior inferior ring epiphysis of the L4 vertebra (limbus vertebra)[1] was also seen and confirmed on magnetic resonance imaging to be a benign process and negative for fracture or other acute processes. The diagnosis of a lumbar strain was made and conservative care instituted, which included anti-inflammatory medication and physical therapy focusing on lumbar stabilization, abdominal strengthening, and aerobic conditioning. Fortunately, the patient's symptoms resolved without incidence.

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Osteopetrosis is a complex disease with at least 4 different types. Their common denominator is abnormal osteoclast-mediated bone resorption resulting in osteosclerosis.[2,3] The *precocious type,* which is autosomal-recessive, is often lethal in early childhood because of complications of bone marrow cavity obliteration. The *delayed type,* termed *Albers-Schönberg disease* or *marble bone disease,* has autosomal-dominant inheritance and may be relatively asymptomatic. It typically presents with pathologic fracture, mild anemia, or cranial nerve palsies in late childhood or early adulthood. It has 2 subtypes: Subtype 1 has pronounced sclerosis of the cranium with relative sparing of the spine; subtype 2 has sclerosis affecting the skull

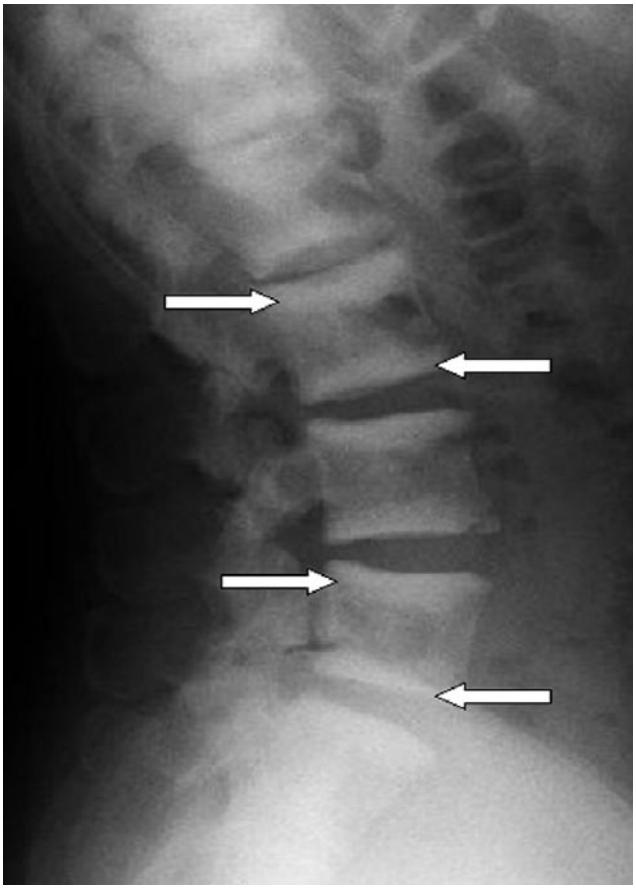

Figure. Lateral plain x-ray of the lumbar spine and sacrum shows vertebral endplate thickening (arrows), representative of the "sandwich vertebrae" manifestation in type 2 osteopetrosis, and a limbus vertebra at L4.

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base, vertebral endplates, and iliac bones.[4,5] Bone marrow function is not compromised in subtype 2. The other 2 main types of osteopetrosis are the *intermediate recessive type* (autosomal-recessive inheritance but with more mild clinical manifestations than the precocious type has) and the *tubular acidosis type* (autosomal-dominant with renal tubular acidosis and mental retardation). The tubular acidosis type is also known as *Sly disease* and, because of the cerebral calcifications involved, *marble brain disease*.

The case reported here emphasizes the importance of recognizing and correctly diagnosing osteopetrosis. The characteristic imaging of sandwich vertebrae combined with supportive laboratory studies confirmed the diagnosis of the autosomal-dominant, type 2 variant.[5] Once the diagnosis is made, it is important to note that these patients are at increased risk for both pathologic and insufficiency fractures because of relatively unopposed osteoblastic activity, despite increased bone density. Therefore, subsequent evaluation should include a search for associated pathologic fractures. If negative, then symptomatic care remains the mainstay of treatment. Improvement can be expected.

Authors’ Disclosure Statement

The authors report no actual or potential conflict of interest in relation to this article.

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