Melorheostosis

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Melorheostosis, also known as Leri's Disease, is a rare, nongenetic mesenchymal dysplasia and one of the benign sclerosing disorders of bone. Men and women are equally affected and the prevalence of the disease is 0.9 per million.[1] The age of presentation ranges from 2 to 64 years, with approximately half the cases being diagnosed before 20 years of age.

Clinical manifestations of melorheostosis are variable, ranging from chronic pain and stiffness to contracture and deformity of the involved limb. Patients may be asymptomatic and the disease can present as an incidental radiographic finding. Serum calcium, phosphorus, and alkaline phosphatase are normal. Melorheostosis may involve only one bone (monostotic), one limb (monomelic), or multiple bones (polyostotic). It is almost always unilateral, with only a few cases of bilateral involvement reported. The lower extremity is more commonly affected than the upper extremity, and the disease only rarely involves the spine, skull, or facial bones (Figure 1).

Melorheostosis distributes along a sclerotomal pattern, corresponding to the osseous distribution of spinal sensory nerve segments. The etiology of melorheostosis remains uncertain; however, a widely accepted theory by Murray and McCredie[2] attributes the disease to segmental sensory nerve insult, caused by a mutation of the LEMD3 gene, leading to bone scarring along its distribution.

Diagnosis of melorheostosis is usually made radiographically. The classic radiographic appearance is that of undulating irregular hyperostotic cortical changes along the long axis of bones, which are said to resemble melting wax dripping down the side of a candle (Figure 2). These dense, linear, sclerotic changes can also involve cancellous (trabecular) bone. The disease may also manifest itself as a more focal osteoma-like lesion. A clear demarcation between normal and affected bone is characteristic. Bone scintigraphy invariably shows uptake in areas of sclerotic bone.

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Am J Orthop. 2009;38(7):360-361.

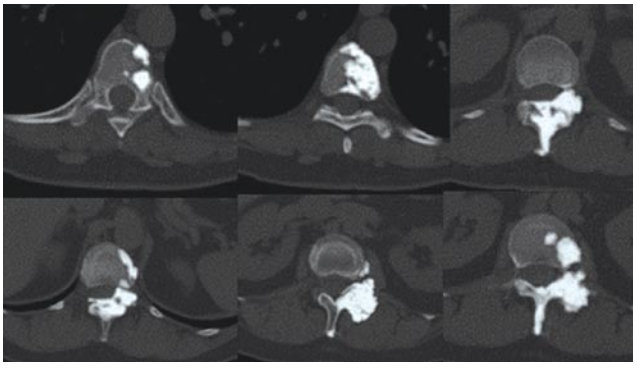

Figure 1. Axial computed tomography images through the lower thoracic and upper lumbar spine in a patient with melorheostosis demonstrate undulating irregular sclerotic changes affecting both cortical and trabecular bones.

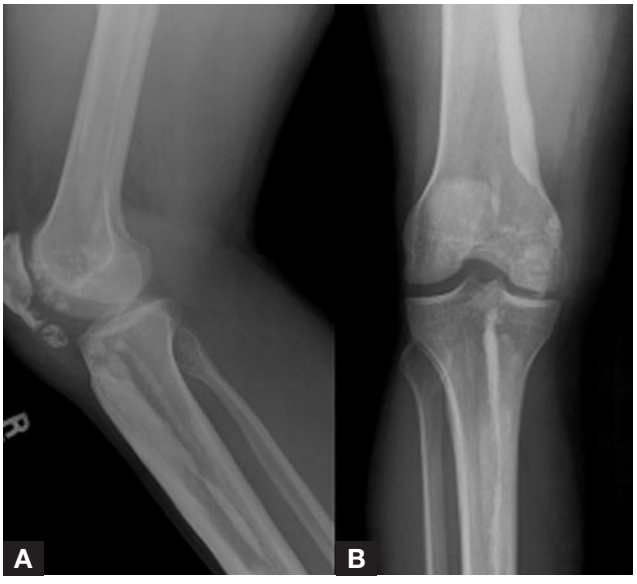

Figure 2. Lateral (A) and frontal (B) views of a knee in a patient with melorheostosis demonstrate undulating irregular hyperostotic cortical changes along the long axis of the femur and tibia resembling melting wax dripping down the side of a candle.

Soft-tissue masses composed of osteoid, chondral, fibrolipomatous, and vascular elements, which may or may not be mineralized, occur in 27% to 76% of patients with melorheostosis.[2,3] These infiltrating, heterogeneous, contrast-enhancing masses are not always contiguous with the hyperostotic lesions and are best evaluated by cross-sectional modalities such as computed tomography or magnetic resonance imaging (Figure 3). Imaging characteristics of these soft-tissue masses may suggest aggressive neoplasm instead of melorheostosis, and failure to recognize the underlying sclerotic bone lesions may prompt unnecessary biopsy (Figure 4).

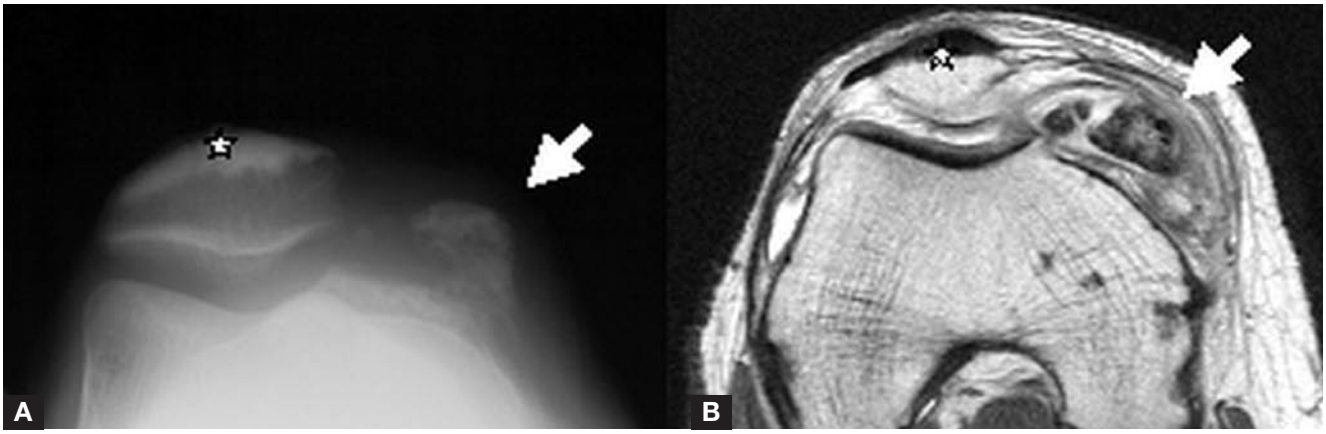

Figure 3. Radiographic Merchant view (A) and axial proton-density magnetic resonance image (B) of a knee demonstrate a partially mineralized mass (arrow) with intra-articular extension into the patellofemoral compartment. There are hyperostotic cortical changes along the anterior aspect of the patella (star), consistent with melorheostosis.

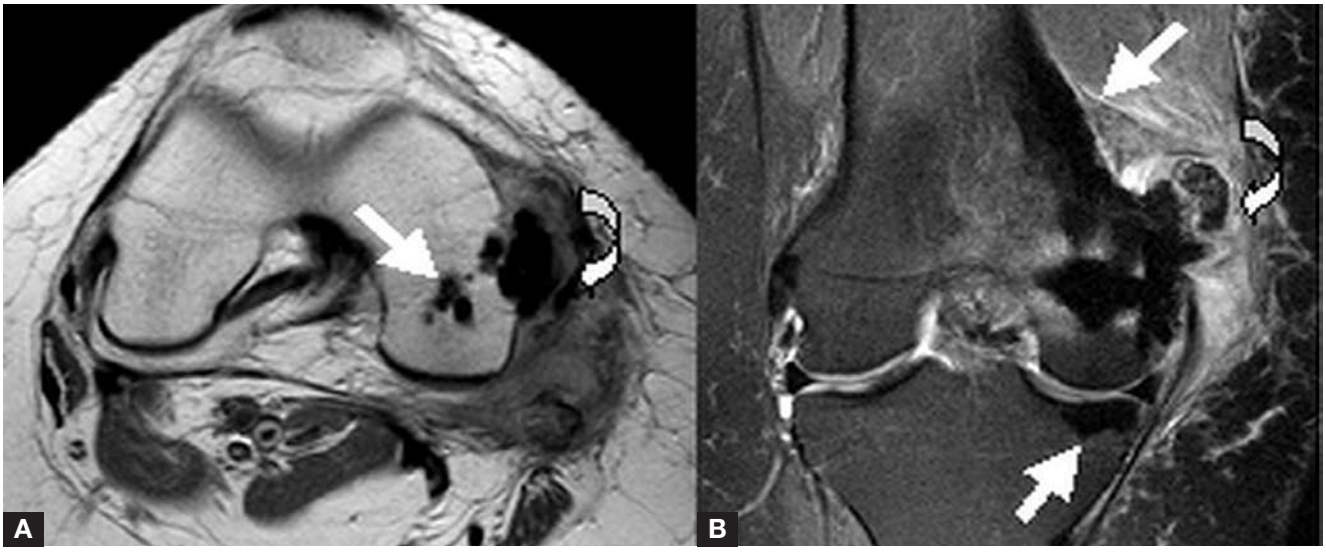

Figure 4. Axial proton-density (A) and coronal fat-saturated proton-density (B) magnetic resonance images of a knee demonstrate an infiltrating, partially mineralized, and aggressive-appearing soft-tissue mass along the distal femur (curved arrow) with surrounding reactive changes. Failure to recognize the characteristic underlying cortically and medullary based sclerotic bone lesions within the femoral metaphysis and tibial plateau (arrows) may prompt unnecessary biopsy.

Intra-articular extension of disease occurs in 35% of patients and classically includes both cortical hyperostosis and extensively mineralized masses.[3] Morris and colleagues[4] described a number of soft-tissue abnormalities associated with melorheostosis, including limb shortening or lengthening, limb deformities, muscle atrophy, scleroderma, lymphatic abnormalities, hemangiomas and other vascular malformations, neurofibromatosis, and skin pigmentation.

Many anecdotal approaches have been offered to treat the pain and soft-tissue contractures associated with this progressive disease. Conservative measures, such as manipulation, analgesia, physical therapy, bisphosphonates and casting, have been utilized. More invasive therapies, including surgical sympathectomy, fasciotomy, resection of hyperostotic regions, arthrodesis, total arthroplasty, and amputation, have also been attempted with varying degrees of success.[5,6]

Authors' Disclosure Statement

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

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