

Denouement and Discussion

Carpal-Tarsal Osteolysis

Joint hypermobility is a relatively common pediatric finding the causes of which range from normal variants to any number of heritable disorders. More familiar genetic causes include the various types of Ehlers-Danlos syndrome, osteogenesis imperfecta (types I and IV), Marfan syndrome, and fragile X syndrome. Less well-known causes of joint hypermobility include pseudoxanthoma elasticum syndrome and cutis laxa syndrome, as well as a variety of skeletal dysplasias such as pseudoachondroplasia and spondyloepiphyseal dysplasia congenita. When patients do not meet clinical criteria for one of these syndromes, they are often diagnosed with benign joint hypermobility syndrome.

For this patient, the radiographic images of the hands and wrists revealed small distal radial epiphyses with absence or absorption of the carpal bones and bases of the metacarpals (Figure, B). Imaging of the lower extremities similarly demonstrated midtarsal bone loss and deformities of the talus and calcaneus. These studies are diagnostic of type 2 carpal-tarsal osteolysis (CTO), also known as idiopathic osteolysis. Carpal-tarsal osteolysis is a rare disorder that belongs to a spectrum of so-called disappearing bone diseases. The condition was first described by Froelich in 1937 in an 18-year-old woman with a progressive dissolution of the carpal and tarsal bones that started when she was 2 years of age.¹ These diseases are characterized by a destructive resorption of bone, but they vary in their pattern of bony involvement, in the age of individual at presentation, in inheritance pattern, in organ involvement, and in the presence of other associated symptoms.²

Patients with CTO often present in early childhood, typically at 2 to 3 years of age with joint pain, joint swelling, and decreased mobility at the wrists or ankles.^{3,4} Pain commonly decreases with disease progression. Joint involvement is usually bilateral, although, at initial presentation, it may be asymmetric. With progressive bone resorption, the natural course of the disease leads to joint deformity with hypermobility and subluxation, and eventually to significant musculoskeletal impairment. The results of laboratory investigations are typically normal, including inflammatory markers. Occasionally, facial dysmorphism has been described, including micrognathia, hypertelorism, triangular face, frontal bossing, and other skull deformities.^{3,5} Radiographic studies typically confirm the diagnosis and can help distinguish the disease from other arthropathies such as juvenile idiopathic arthritis. Plain radiographs reveal progressive osteolysis of the carpal and tarsal bones, as well as adja-

cent tubular bones (eg, metacarpal bones and phalanges). Resorption of other bones, including elbow involvement, although uncommon, has been reported. At presentation, radiographs may be normal, making it difficult to establish a diagnosis unless radiography is performed again months or even years later. In general, the overall health and well-being of the individual are not typically affected, and the disease process often stabilizes between the ages of 20 and 30 years for most individuals.

Treatment of CTO is largely supportive in nature and may include pain relief, orthotics or prosthetics, and physical or occupational therapy. Steroids and other anti-inflammatory medications have not been shown to be beneficial. One report⁶ describes the successful treatment of the nephropathy associated with CTO with cyclosporine A. In another report,⁷ a patient with osteolysis of the metacarpal bones in both hands was found to have an increased number of stimulated osteoclasts in biopsy specimen. Treatment with bisphosphonates halted further progressive osteolysis.⁷

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