

IMAGES IN CLINICAL RADIOLOGY



Multicentric carpal-tarsal osteolysis

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A 2.5-year-old girl presented with pain and reduced strength in hands and wrists and walking difficulties. Clinical examination revealed a position deformity of both hands and broad elbows with limited extension.

Plain films showed marked changes with a rather symmetric distribution. Radiograph of the left hand revealed absence of ossification of carpal bones with shortening of the wrist (Fig. A, long arrow). Ulnar deviation of the wrist was seen due to erosion and shortening of the ulna (Fig. A, short arrow). The proximal ends of the second and third metacarpal were tapered (Fig. A, curved arrows). An oblique radiograph of the right foot showed underdeveloped medial tarsal bones (Fig. B, arrow). A lateral radiograph of the right elbow demonstrated irregular delineation of the capitulum of the humerus (Fig. C, arrow).

Based on the combination of clinical and imaging findings, the diagnosis of multicentric carpal-tarsal osteolysis (MCTO) (MIM 166300) was made.

Comment

MCTO is a rare congenital osteolysis syndrome. There are two major subtypes: with or without nephropathy. It is considered as an autosomal dominant condition for which the gene defect remains unraveled. The pathogenesis is still a matter of debate, but abnormal proliferation of fibrous tissue or an immune disturbance have been incriminated as potential etiologic mechanisms. Histological evidence of active inflammation has never been shown.

The onset of MCTO may be exacerbated by a mild trauma. The disorder has a characteristic clinical course and begins in childhood with joint pain, swelling and tenderness. In order of frequency, the carpal bones, tarsal bones and other joints (elbow and shoulder) are involved to a variable degree. The involvement may be asymmetric. Typically, in adolescence the symptoms subside with a relatively asymptomatic period. Progressive deformities develop in the third decade. Affected individuals may have a triangular face with mandibular hypoplasia. In some patients nephropathy can occur and may be fatal.

Plain radiographs have a pivotal role in the diagnosis of the disease. The radiological hallmark consists of progressive osteolysis of the carpal and tarsal bones. In early stage disease, there is progressive demineralization with loss of bone contours. Further bone resorption results in collapse, fragmentation and sclerotic remnants of the carpal and tarsal bones. Finally, partial resorption of adjacent tubular bones leads to tapering of the proximal ends of the metacarpals and metatarsals with a characteristic "sucked candy" appearance. In severely affected individuals deformity of metacarpal, metatarsal and interphalangeal joints may occur. Irregular delineation of the epiphyseal centers of elbow and shoulder may occur as well. On magnetic resonance imaging, a fibro-collagenous tissue replaces the carpal or tarsal bones, without evidence of synovial inflammation.

The most significant differential diagnosis includes juvenile rheumatoid arthritis (JRA). MCTO can be clinically differentiated from JRA by absence of parameters of acute inflammation. MCTO is distinguished from Gorham's osteolysis in that the latter is unicentric and histopathologically associated with hemangiomatosis.

Our patient was treated supportively, with physiotherapy and ergotherapy. There is little evidence suggesting that bisphosphonates may retard the natural history or progression of the disease. Hemodialysis applied in case of end-stage renal failure complicating the subtype with nephropathy.

Reference

1. McDonald K., Toms A.P., Armon K., Johnson K., Marshall T.J.: Carpal-tarsal osteolysis with elbow involvement. *Skeletal Radiol*, 2007, 36: 1097-1101.

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