

Alkaptonuria with Atypical Joint Involvement

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Ochronosis occurs from a deficiency of the enzyme homogentisic acid oxidase leading to elevated levels of homogentisic acid, which is oxidized, polymerized, and deposited in connective tissue, causing characteristic pigmentation. The arthritis is similar to osteoarthritis (OA), also with characteristic calcification of the discs, and has been confused with other inflammatory arthritis.

A 70-year-old man with ochronosis had destructive OA, but also inflammatory findings in the hands, elbows, and shoulders. He presented 40 years ago with bluish tinged sclerae and OA of the hips and knees. Having already had bilateral knee replacements and one hip arthroplasty, he had accelerated, severe, and atypical OA of the hips, knees, elbows, shoulders, and proximal interphalangeal (PIP) and distal interphalangeal (DIP) joints. Recently the upper extremities (particularly elbows) had been more symptomatic. Examination revealed blue pigmentation of the sclerae, ears, face, palms,

some tendon sheaths, and knuckle pads (PIP, metacarpophalangeals, MCP). He had marked coarse crepitus, restricted range of motion (ROM) of many joints (PIP, MCP, wrists, elbows), boggiess of his wrists, and minimal MCP thickening but no evidence of MCP synovial proliferation or effusions. Shoulders had extremely limited ROM and severe pain. Effusions of the elbows and shoulders were present. Knees had reduced flexion bilaterally and right hip had some flexion but no internal or external rotation. He had no movement at the subtalar joint and marked kyphosis.

Conventional radiographs showed evidence of inflammatory features with erosions in some areas in the hands, elbows, and right shoulder. There was evidence of minimal chondrocalcinosis in the left wrist (Figures 1–3). This arthropathy is atypical for rheumatoid arthritis and OA, and we suggest is from the metabolic alterations in the cartilage with secondary inflammation.



Figure 1. Anteroposterior radiographs of hands and wrists bilaterally. There is an absence of periarticular osteopenia. Left ulnarcarpal area has faint calcification. Large erosions are seen at ulnar side of 5th MCP, radial aspect of left 2nd MCP, and several PIP. Osteophytes and sclerosis are absent.



Figure 2. Radiograph of the subject's elbow reveals no periarticular osteopenia but symmetrical joint space narrowing and large atypical erosions with minimal osteophyte formation. There is no evidence of chondrocalcinosis.



Figure 3. Anteroposterior right shoulder. There is symmetrical glenohumoral joint space narrowing with erosions seen inferiorly along the joint. Sclerosis and osteophytes are not prominent. The head of the humerus is flattened and riding superiorly, indicating probable degenerative rotator cuff. No calcification is seen. The acromioclavicular joint is spared.