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The patient, a 56-year-old woman with progressively worsening back pain, was referred to our rheumatology clinic for ankylosing spondylitis (AS) evaluation after being informed that she had spinal ankylosis on radiographs. Her medical history was significant for a childhood diagnosis of alkaptonuria. On physical examination, she had bluish-black discoloration of her external ears (left) and sclerae. Spine radiographs showed severe disc space narrowing with calcification of the degenerated lower lumbar spine discs (middle), fusion of the upper lumbar spine disc spaces (right), and severe osteopenia; the sacroiliac joints were patent. Alkaptonuria is a rare autosomal-recessive metabolic disorder in which a homogentisate 1,2-dioxygenase enzyme deficiency leads to an accumulation of homogentisic acid (HGA) (1,2). The classic clinical triad of alkaptonuria is homogentisic aciduria, ochronosis, and ochronotic arthropathy. The tissue deposition of the pigmented oxidation products of HGA results in ochronosis, which is the dark pigmentation of connective tissues (e.g., external ear, sclera, and intervertebral disc) in alkaptonuria. Ochronotic arthropathy commonly affects the thoracolumbar spine and the weight-bearing joints, and contributing mechanistic factors include deposition of pigmented HGA polymers, loss of cartilage elasticity, altered mechanical loading transmission, and aberrant bone remodeling (3). This case highlights an important mimic of AS.


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