A metabolic cause of spinal deformity

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A 38-year-old man presented to our clinic with a 6-year history of chronic low back pain. Physical examination showed limited spine mobility; radiographs of the spine demonstrated narrowed disk spaces and calcifications. Lumbar spine magnetic resonance imaging showed Modic type II signal intensity changes in the bone marrow consistent with chronic disk degeneration. The finding of a massively elevated excretion of homogentisic acid (HGA) in the patient's urine confirmed the suspicion that the complaints were due to underlying alkaptonuria. Alkaptonuria (ochronosis) is an uncommon cause of backache and results from mutations in homogentisate 1,2-dioxygenase, an enzyme involved in tyrosine catabolism. Homogentisic acid accumulates in the plasma of the affected individuals, and HGA polymers deposit in connective tissues where they cause cartilage degeneration. So far, there is no proven treatment; but preclinical and phase I data with nitisinone, an inhibitor of HGA formation, are promising. Currently, the effects of nitisinone on joint mobility are being evaluated in a randomized trial. Clinicians involved in the care of musculoskeletal problems should be aware of this rare disorder, particularly because the correct diagnosis may have therapeutic implications.
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