

Ochronotic Spondyloarthropathy Mimicking Diffuse Idiopathic Skeletal Hyperostosis

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INTRODUCTION

A 59-year-old man presented with 20 years history of low back pain. The pain was mechanical in nature with early morning stiffness of less than 15 minutes. The patient was followed-up with a diagnosis of diffuse idiopathic skeletal hyperostosis (DISH) by the other physicians. He had partial response to analgesics.

On inquiry, the patient reported that since childhood his urine had turned a dark color on exposure to air. Physical examination revealed blue-black pigmentation of the sclera and ear pinna bilaterally (Figure 1). On laboratory, full blood count, calcium, phosphorus, alkaline phosphatase, erythrocyte sedimentation rate, C-reactive protein and rheumatoid factor were normal. HLA B-27 was also negative. Measurement of 24 hours urinary homogentisic acid levels was $957 \mu\text{mol}/\text{mmol creatinine}$ (reference value, $<1 \mu\text{mol}/\text{mmol creatinine}$). X-ray showed multilevel intervertebral disc space narrowing associated with disc calcifications of the thoracolumbar spine (Figure 1). The diagnosis of ochronosis was established, ascorbic acid and nitisinone were started for the patient. Protein restriction was made in his diet.

Ochronosis is a rare hereditary autosomal recessive disorder characterized by an excess of homogentisic acid deposits in the tissues. The incidence of ochronosis (alkaptonuria) is estimated at 1 case in 250,000 to 1 million live births [1]. Ochronotic arthropathy generally appears in the third decade, commonly affects in which weight bearing joints and the thoracolumbar spine. Narrowing of the intervertebral spaces and widespread,

thin, linear, wafer-like disc calcification are diagnostic findings seen on spine radiographs [2]. A potential agent called 2 mg of daily nitisinone has been shown to decrease clinical progression of ochronosis [3]. Disc calcifications

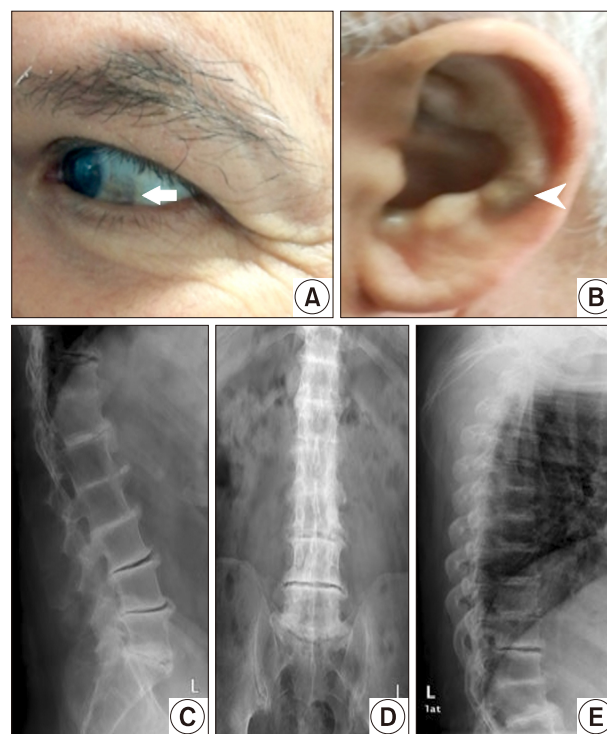


Figure 1. Photographs show blue-black pigmentation of the sclera (arrow) (A) and ear pinna (arrowhead) (B), X-ray of the thoracolumbar spine, multilevel intervertebral disc space narrowing associated with disc calcifications (C~E). We received the patient's consent form about publishing all photographic materials.

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must be distinguished from those of the following: ankylosing spondylitis, juvenile idiopathic arthritis, DISH, calcium pyrophosphate dehydrate crystal deposition disease, hyperparathyroidism, Klippel-Feil syndrome, and congenital and acquired fusions of the spine [4]. The diagnosis of ochronosis must be considered for any patient with multilevel vertebral-body disc involvement.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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