

# Clinical Quiz

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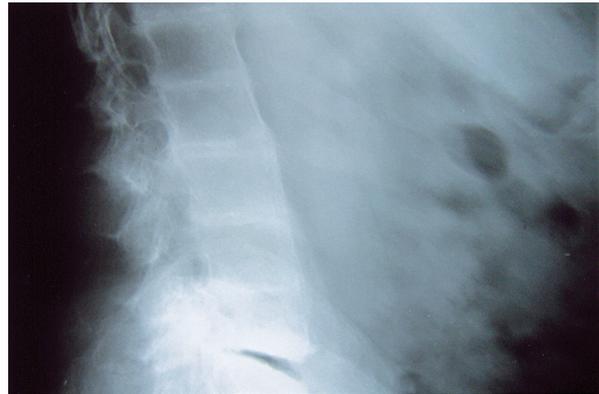
## A 69-year-old man with a thick dark pigmentation on his hands, and low back pain

### Clinical Presentation

A 69-year-old man presented with progressive low back pain, difficulty in walking, and black thick pigmented skin lesions over the palmar aspect of both index fingers and thumbs (Figure 1). Plain radiograph of the lumbar spine showed intervertebral disc calcification (Figure 2).



**Figure 1** - Skin lesions over the palmar aspect of both index fingers and thumbs.



**Figure 2** - Radiograph of the lumbar spine showing intervertebral disc calcification.

## Questions

1. What is the diagnosis?
2. How would you treat this condition?

# Clinical Quiz

## Answers

1. These cutaneous lesions are characteristic of alkaptonuria. The spinal x-ray showed typical ochronotic arthropathic changes (intervertebral disc calcification).
2. There is no specific therapy for ochronosis, supportive therapy such as nonsteroidal anti-inflammatory drugs (NSAIDs), and physical therapy is used for the arthropathy.

## Discussion

Alkaptonuria (ochronosis) is a rare inherited (Mendelian recessive) disorder of metabolism characterized by deficiency of homogentisic acid oxidase, which involved in the catabolism of aromatic amino acids (phenylalanine and tyrosine).<sup>1</sup> This leads to the accumulation of homogentisic acid and its metabolites in collagenous tissues causing ochronosis, characterized by darkened cartilaginous tissues, arthritis, and joint destruction.<sup>2,3</sup> The disease manifested clinically by spondyloarthropathy and soft tissue ochronosis, with a characteristic feature of darkening of urine. Patients might be asymptomatic until the third or fourth decade when scleral pigmentation (Osler's sign) starts or skin pigmentation appears.<sup>4</sup> One of the first sites to be involved is the ear cartilage, which becomes thickened with bluish black pigmentation. There may be widespread dusky discoloration of the skin of the cheeks, forehead, axilla, and genitalia.<sup>5</sup> Ochronotic arthropathy is a particularly troublesome feature, and appears insidiously around the fourth decade resembling osteoarthritis. There is involvement of weight-bearing joints like the spine and knees, as well as the shoulders.<sup>3,4</sup> Detecting and measuring the amount of homogentisic acid in the urine confirms the diagnosis of this disease. Treatment of alkaptonuria is frustrating and involves giving a low protein diet with restriction of phenylalanine and tyrosine. Supportive therapy like NSAIDs, and physical therapy is used for the arthropathy but the course of the disease remains the same.

## References

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