Femoral head ochronotic pigmentation in a patient with alkaptonuria and secondary hip osteoarthritis

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Short title: Ochronotic hip pigmentation in alkaptonuria

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A 59-year old female patient presented to the rheumatology clinic with a longstanding history of shoulder, low back and hip pain. She was diagnosed with alkaptonuria at the age of 45 and since then developed mechanical pain that progressively limited her everyday activities. Radiological imaging confirmed generalized secondary osteoarthritis that was more pronounced in her lumbar spine (Figure 1) and right hip (Figure 2). Pain management required a multimodal pharmacological approach, physical therapy and hip and knee hyaluronic acid injections. Right hip pain remained severe and non-responsive to treatment and a total hip replacement was performed with significant relief and functional improvement. Surgical specimen of the femoral head exhibited the typical dark-bluish (ochronotic) pigmentation on the joint surface (Figure 3).

Alkaptonuria is an autosomal recessive disorder resulting from the deficiency of a tyrosine degradation enzyme named homogentisic acid dioxygenase[1]. The accumulation of homogentisic acid in the connective tissue is responsible for the clinical features of alkaptonuria, including joint damage and secondary osteoarthritis[2]. This ochronotic arthritis can be severe and disabling and half of these patients require large joint replacement by the sixth decade of life[2].

Figures

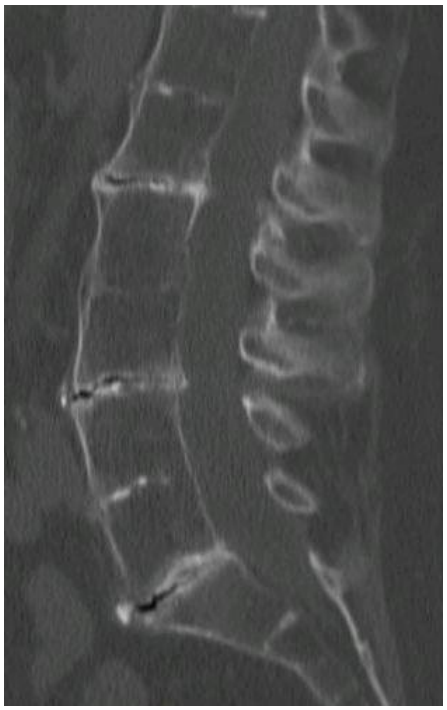

Figure 1 – Lateral view of lumbar spine CT scan with multilevel disc collapse, intervertebral disc calcification and syndesmophytes

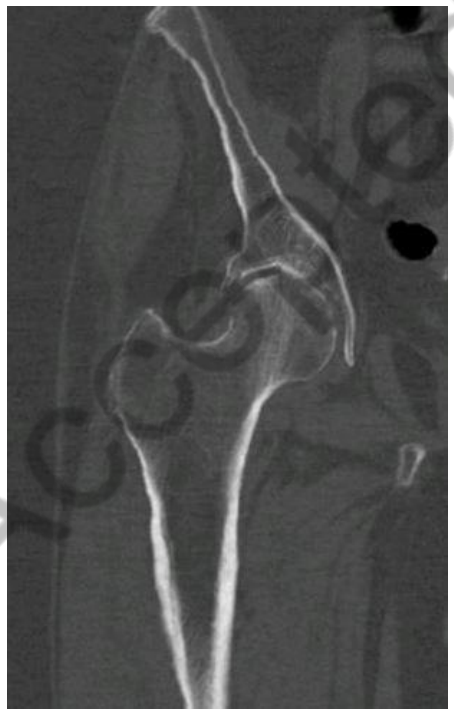

Figure 2 - Frontal view of right hip CT scan showing joint space narrowing, osteophytes and subchondral sclerosis and cysts

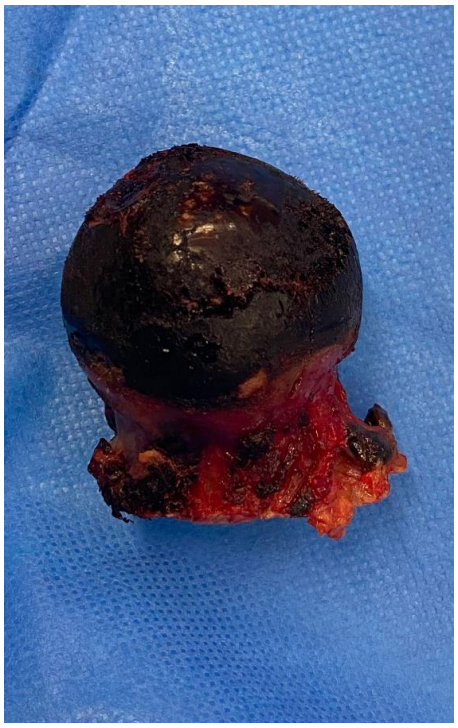

Figure 3 - Surgical specimen of the femoral head showing significant cartilage disruption and the typical dark-bluish (ochronotic) pigmentation.

References

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