

SNAPSHOT

Black bones: a case of incidental discovery of ochronotic arthropathy

A 77-year-old man with no significant medical history presented to the orthopaedic clinic of a metropolitan hospital with longstanding left mechanical knee pain. Examination and x-ray revealed features consistent with degenerative arthritis (Figure, A), and he subsequently underwent total knee arthroplasty. During surgery, a bluish-black pigmentation of the bone and the cartilage of the knee joint was noted (Figure, B). The patient's surgery and recovery were uneventful. It was later discovered that he had a previously undiagnosed rare metabolic disorder, alkaptonuria, which affects one in 250 000 to 1 000 000 people worldwide.¹

Alkaptonuria was one of the first inborn errors of metabolism to be described, in 1908.² It is caused by mutations in the homogentisate 1,2-dioxygenase (*HGD*) gene, which results in a deficiency of HGD, which catabolises homogentisic acid (HGA). This leads to accumulation and deposition of HGA in cartilaginous tissues, causing a bluish-black discolouration (ochronosis).³ Ochronosis is generally asymptomatic, but ochronotic arthropathy due to deposition of pigments in the joints is common.³

The patient also had the characteristic ochronotic discolouration in the sclera (Figure, C), ear cartilage, fingernails and buccal mucosa, but did not report dark urine. The gene defect was not further investigated because of his age and excellent premorbid health status.

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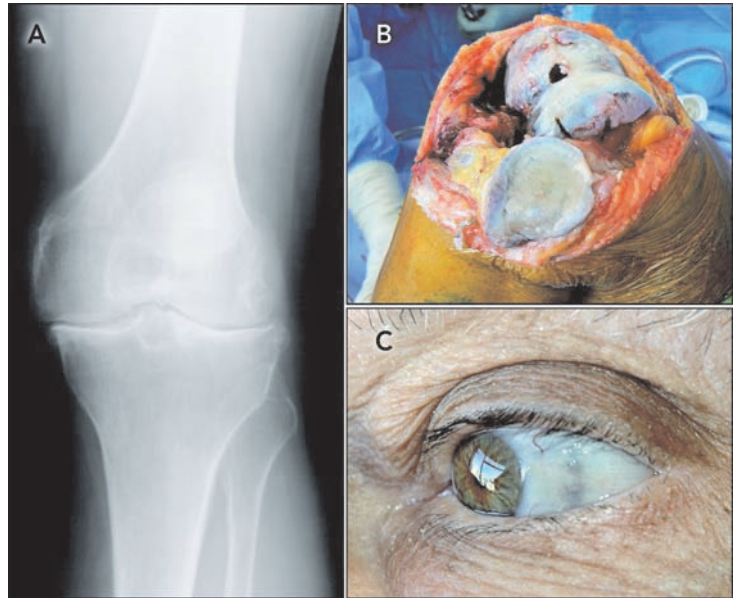
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A: Plain radiograph of the left knee revealing reduced joint space with periarticular sclerosis. **B:** Bluish-black discolouration of the left knee joint. **C:** Bluish-grey scleral pigmentation in the shape of a small ring.

1 La Du BN. Alkaptonuria. In: Scriver CR, Beaudet AL, Valle D, et al, editors. The metabolic and molecular bases of inherited diseases. 8th ed. New York: McGraw-Hill, 2001: 2109-2123.

2 Garrod AE. The Croonian lectures on inborn errors of metabolism. III. Alcaptonuria. *Lancet* 1908; 2: 73.

3 Phornphutkul C, Introne WJ, Perry MB, et al. Natural history of alkaptonuria. *N Engl J Med* 2002; 347: 2111-2121. □