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# Black cartilage: Incidentally discovered articular ochronosis during arthroplasty

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A 53-year-old man was addressed to the orthopedic clinic for a knee replacement surgery.

The patient complained of pain in several joints starting at age 30, mostly affecting the lumbar spine. Over several years, he developed non-inflammatory arthritis of the hips and knees, leading to the indication of total replacement of the right knee joint. Upon opening of the joint capsule, the surgeon was startled to observe a striking brown-black discoloration of the cartilage. This unexpected finding led to the suspension of the surgery



FIGURE 1 Hyperpigmentation of the scar following the uncompleted total knee replacement surgery

pending clarification of the etiology. Clinical examination revealed ochronosis of the ear cartilage bilaterally, sclera and teeth, together with a hyperpigmented postoperative scar (Figure 1). Urine organic acid analyses unveiled homogentisic acid elevation. Molecular analysis showed two pathogenic variants in the *HGD* gene [c.133delT, p. (Ser45GInfsTer66); c.1078G>C, p.(Gly360Arg)], hence confirming the diagnosis of Alkaptonuria (AKU; OMIM #203500).<sup>1</sup> Joint replacement surgery was rescheduled and performed successfully with uncomplicated postoperative follow-up (Figure 2).

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## **CONFLICT OF INTEREST**

The authors have disclosed no conflicts of interest.

### **INFORMED CONSENT**

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the

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FIGURE 2 Ochronotic deposits of the joint cartilage discovered intraoperatively, (A). Difference between ochronotic cartilage and nonpigmented bone after shaping of the distal femoral bone, (B)

Helsinki Declaration of 1975, as revised in 2000. An informed consent was obtained from the patient included in the study.

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## REFERENCE

1. Ranganath LR, Cox TF. Natural history of alkaptonuria revisited: analyses based on scoring systems. J Inherit Metab Dis. 2011;34:1141-1151.

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