

Metabolism

Pigmented sclera: a diagnostic challenge?

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Figure 1 shows the eyes of a 53 year old man who has suffered with severe generalised arthritis for more than 15 years, requiring left knee replacement at the age of 45 years. He has noticed dark urine on several occasions and passed many renal stones. His sister (aged 52 years) also has generalised arthritis especially in the large joints of the lower limbs and the back. In addition, she has been passing dark urine and has dark pigmentation of the cartilage of her ear (fig 2).



Figure 1 Eyes of male patient showing scleral pigmentation (published with permission).

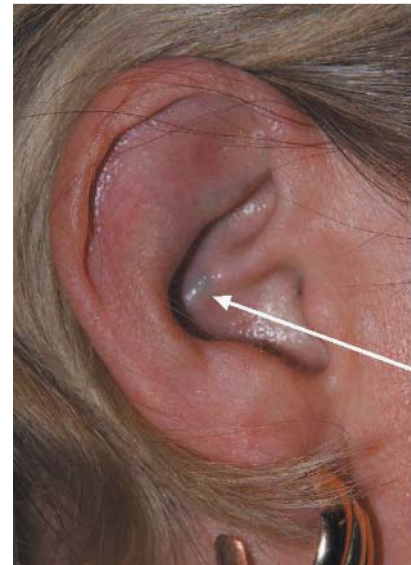


Figure 2 Ear of patient's sister showing pigmentation of cartilage (published with permission).

QUESTIONS

- (1) What is the diagnosis in the male patient?
- (2) Does his sister have the same condition?
- (3) What further investigations would you perform to confirm your diagnosis?

- (4) What are the clinical features of this disease?
- (5) How would you manage this condition?

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