

CLINICAL IMAGE

Operating the blues

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Case History

The case of patient with aortic stenosis due to ochronosis is presented. A 63-year-old man with known history of alkaptonuria presented with critical aortic stenosis. His medical history included a prosthetic total hip replacement and cervical spondylosis due to degenerative arthritis (Fig. 1). Physical examination showed bluish black pigmentation of the face. At operation, a severely calcified tricuspid aortic valve was observed with bluish black pigmentation involving the entire luminal surface of the ascending aorta (Fig. 2A). Prosthetic aortic valve replacement was performed. Postoperative course was uneventful with the expected remarkable finding of dark urine (Fig. 2B).

Alkaptonuric ochronosis is a rare autosomal recessive genetic disorder of tyrosine metabolism and is associated with homogentisic acid oxidase enzyme deficiency due to chromosome 3q mutations resulting in the accumulation of homogentisic acid [1]. Degenerative arthritis, ochronotic connective tissue pigmentation including structures of the cardiovascular system, and urine darkening are the predominant findings. Aortic valvular stenosis is the most frequent cardiac manifestation [2].

Key Clinical Message

A 63-year-old man bearing most signs and symptoms (facial pigmentation, degenerative arthritis, and dark urine) pertinent to his known history of alkaptonuria underwent aortic valve replacement for critical aortic stenosis. Although rare, aortic stenosis is the most common cardiac manifestation of alkaptonuric ochronosis.

Keywords

Alkaptonuria, aortic stenosis, ochronosis.

Conflict of Interest

None declared.

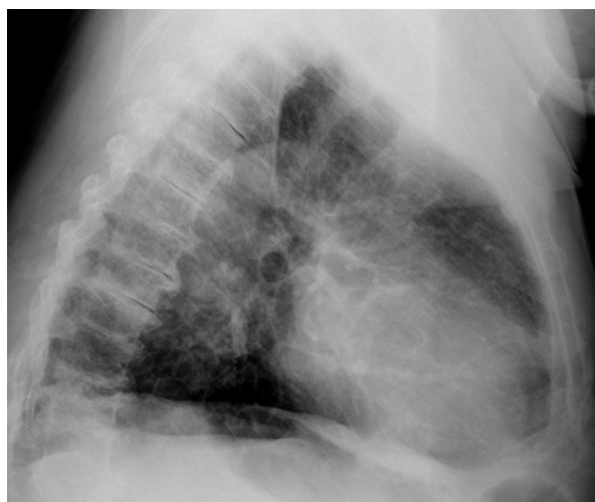


Figure 1. Cervical spondylosis.

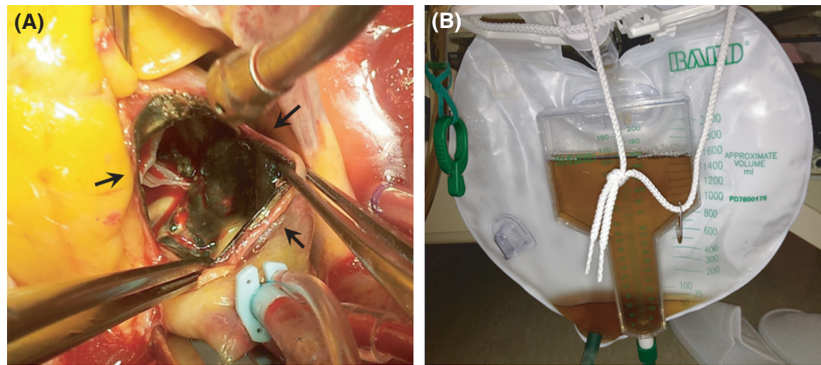


Figure 2. (A) Calcified “blue” aortic valve signified by arrows (cephalic view). (B) Dark urine.

References

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