

In-vivo Aortic Valve of a Patient with Alkaptonuria

Válvula Aórtica de um Doente com Alcaptonúria

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Palavras-chave: Alcaptonúria; Estenose de Válvula Aórtica; Implante de Prótese de Válvula Cardíaca

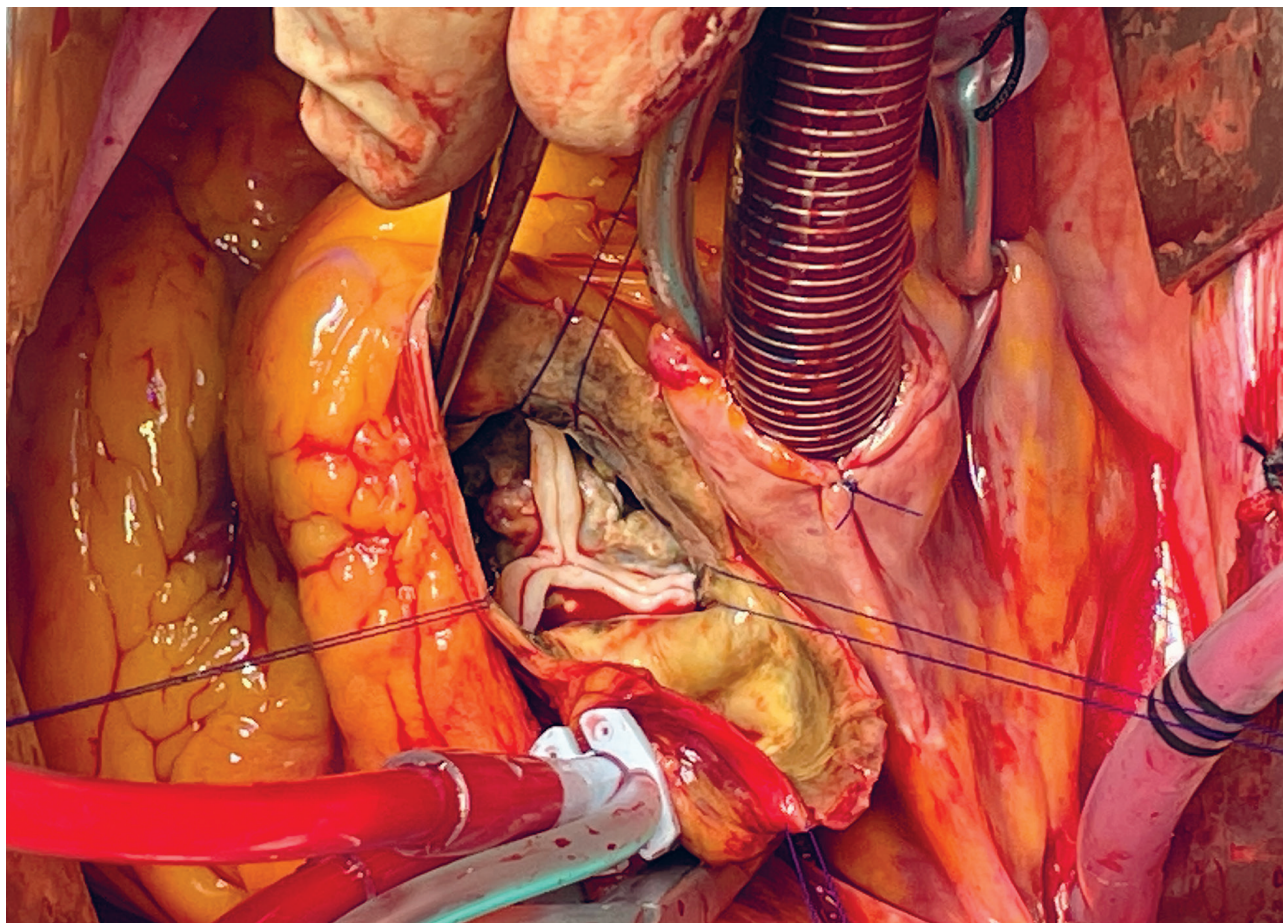


Figure 1 – Intraoperative photograph showing the dark discoloration in the aortic valve cusps and the ascending aorta wall

A 73-year-old man underwent surgery due to severe aortic valve disease. The aortic valve cusps and the wall of the ascending aorta, which are usually pearly white, had a dark discoloration (ochronosis). He also presented typical dark spots in his sclerae and ears and suffered from severe osteoarthritis of weight-bearing joints, having undergone bilateral knee and hip replacements years before. Histology of the aortic valve cusps confirmed the suspicion of cardiac

ochronosis. The patient had an uneventful recovery and was discharged home five days after valve replacement. In alkaptonuria, deficiency of homogentisate dioxygenase,^{1,2} an enzyme involved in tyrosine metabolism, results in high levels of circulating homogentisic acid (HGA). HGA is oxidized to benzoquinones that polymerize, yielding a dark pigment.¹ From birth, the urine from these patients turns black after exposure to air for several hours due to homogentisic

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aciduria; however, with ageing, pigment deposition leads to multisystemic disease.² Alkaptonuria affects one in 250 000 to 1 000 000 live births.²

AUTHOR CONTRIBUTIONS

JM: Conception, investigation and drafting of the work.

VM, PP: Drafting and critical review of the work.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in 2013.

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DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT

Obtained.

COMPETING INTERESTS

The authors have declared that no competing interests exist.

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