FIC Federazione Italiana di Cardiologia
Italian Federation of Cardiology

Black aorta in a patient with alkaptonuria (ochronosis)

Giovanni Concistrè, Brenno Fiorani, Federico Ranocchi, Giovanni Casali, Antonio Loforte and Francesco Musumeci

A rare cause of valvular heart disease is the deposition of foreign material in the valvular tissues, including material accumulating as a result of inborn errors of metabolism of the essential amino acids. Alkaptonuria can result in accumulation of homogentisic acid. We report the case of a patient with alkaptonuria undergoing surgery for aortic valve replacement.

J Cardiovasc Med 2011, **12**:444–445

Keywords: aorta/aortic, aortic valve, prosthesis, replacement

Department of Cardiac Surgery and Heart Transplantation, San Camillo Hospital, Circonvallazione Gianicolense 1, Rome, Italy

Correspondence to Dr Giovanni Concistrè, Department of Cardiac Surgery and Heart Transplantation, San Camillo Hospital, Circonvallazione Gianicolense 1, 00146 Rome, Italy
Tel: +39 06 555 3800; fax: +39 06 555 2221;
e-mail: gioconci@libero.it

Received 25 October 2010 **Revised** 7 January 2011
Accepted 21 January 2011

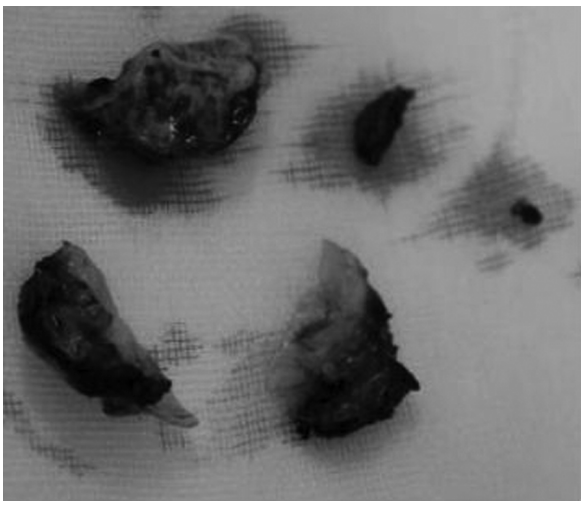
Alkaptonuria is a very rare congenital metabolic disorder that affects about 1 in 1 million births. This disease is transmitted by a single recessive autosomal gene, resulting in an irreversible, progressive, connective tissue disease [1]. Alkaptonuria is associated with a homogentisic acid (HGA) oxidase enzyme deficiency. This deficiency causes the excretion of large quantities of HGA in the urine, which turns dark upon standing. Although the most common clinical feature is severe ochronotic spondyloarthropathy, a wide spectrum of clinical manifestations – including ocular and cutaneous pigmentation, genitourinary obstruction by ochronotic calculi, and cardiovascular system involvement – has been described [2–4]. Deposition of polymerized HGA occurs in the aortic intima, the aortic and mitral valves, the coronary arteries, the subendocardium and the pericardium [2–4]. The exact incidence of cardiovascular disease in patients with ochronosis is not clear; a number of studies suggest a high prevalence of aortic valve stenosis in such patients who are more than 50 years old [1,5].

A 64-year-old woman with known alkaptonuria was admitted to our department with a 4-week history of dyspnea and pulmonary edema due to severe aortic valve stenosis.

Alkaptonuria was evidenced by HGA in the urine and ochronotic arthropathy in vertebrae, hip and knee joints.

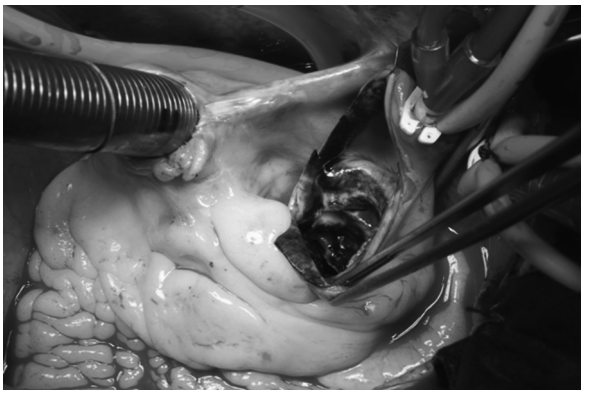
Transthoracic echocardiography confirmed severe aortic stenosis with a mean aortic valve gradient of 80 mmHg. The calculated aortic valve area was 0.3 cm^2, and the left-ventricular ejection fraction was 55%. The mitral annulus was heavily calcified with no evidence of mitral valve stenosis or regurgitation. Cardiac catheterization revealed normal coronary arteries.

Fig. 1



Dark ochronotic pigmentation in the aortic valve leaflets.

Fig. 2



The aortic wall with evident dark pigmentation.

1558-2027 © 2011 Italian Federation of Cardiology DOI:10.2459/JCM.0b013e328344e5d2

The patient underwent aortic valve replacement under cardiopulmonary bypass. Oblique aortotomy revealed a tricuspid aortic valve with thickened dark leaflets (Fig. 1). The aortic wall had a similar appearance (Fig. 2). The aortic valve was excised, and a mechanical prosthesis (19-mm; St. Jude Medical Regent, St. Paul, Minnesota, USA) was sutured in the supra-annular position using 2–0 Ticron interrupted sutures. We implanted a small prosthesis and we excluded indication to annular enlargement because the body surface area was 1.39 m^2. The postoperative course was uneventful, and the patient was discharged from hospital on day 7.

References

1 Van Offel JF, De Clerck LS, Francx LM, Stevens WJ. The clinical manifestations of ochronosis: a review. *Acta Clin Belg* 1995; **50**:358–362.

2 Erek E, Casselman FR, Vanermen H. Cardiac ochronosis: valvular heart disease with dark green discoloration of the leaflets. *Tex Heart Inst J* 2004; **31**:445–447.

3 Butany JW, Naseemuddin A, Moshkowitz Y, Nair V. Ochronosis and aortic valve stenosis. *J Card Surg* 2006; **21**:182–184.

4 Vavuranakis M, Triatafillidi H, Stefanadis C, Toutouzas P. Aortic stenosis and coronary artery disease caused by alkaptonuria, a rare genetic metabolic syndrome. *Cardiology* 1998; **90**:302–304.

5 Phornphutkul C, Introne WJ, Perry MB, Bernardini I, Murphey MD, Fitzpatrick DL, *et al.* Natural history of alkaptonuria. *N Engl J Med* 2002; **347**:2111–2121.