Aortic valve replacement surgery reveals previously undiagnosed alkaptonuric ochronosis

Christian Pfeffera,*, Erik Bagaev, Karl Sotlarb and Christian Hagla

a Herzchirurgische Klinik und Poliklinik der Ludwig-Maximilians-Universität München, Munich, Germany
b Pathologisches Institut der Ludwig-Maximilians-Universität München, Munich, Germany

* Corresponding author. Herzchirurgische Klinik und Poliklinik der Ludwig-Maximilians-Universität München, Marchioninstraße 15, 81377 Munich, Germany. Tel: +49-89-70952951; fax: +49-89-70958898; e-mail: christian.pfeffer@med.uni-muenchen.de (C. Pfeffer).

Received 3 January 2014; received in revised form 13 February 2014; accepted 18 February 2014

Keywords: Alkaptonuria • Ochronosis • Homogentisic aciduria • Aortic valve surgery

A 64-year-old patient with aortic stenosis underwent surgical aortic valve replacement. At surgery, severe annular calcification and blackish-blue discolouration of the valve cusps, aortic wall and coronary arteries were noted (Fig. 1). Microscopic examination revealed dark ochronotic pigment in the valvular tissue (Fig. 2).

Figure 1: Ochronosis of the ascending aortic wall (A) and aortic valve annulus (B).

Figure 2: Ochronosis of the aortic valve leaflets (A) and HE staining of aortic valve leaflets (B and C) showing ochronotic pigment (arrows).