Review

Valvular and Aortic Diseases in Osteogenesis Imperfecta

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Osteogenesis imperfecta (OI) is an inheritable connective tissue disorder caused by defective collagen synthesis with the principal manifestations of bone fragility. OI has been associated with left sided valvular regurgitation and aortic dilation. Valve and aortic surgery are technically feasible in patients with OI but are inherently high risk due to the underlying connective tissue defect. This report reviews the valvular and aortic pathology associated with OI and their management. We describe two cases of patients with OI who have significant aortic and mitral valve regurgitation, one of whom has been managed conservatively and the other who has undergone successful mitral valve repair and aortic valve replacement. The latter case represents the fifth case of mitral valve repair in a patient with OI reported in the medical literature.

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Case Presentations

Case 1

A 59 year-old male patient with a diagnosis of OI was noted incidentally to have a heart murmur in the context of an upper respiratory tract infection. He reported symptoms of mild dyspnoea (NYHA class II) at presentation. He had no prior cardiac history. Transthoracic echocardiography showed severe mitral regurgitation, moderate aortic regurgitation and a dilated left ventricle (LV) with preserved systolic function (left ventricular end-systolic diameter (LVESD) = 50 mm, left ventricular ejection fraction (LVEF) = 65%). Comorbidity included severe auditory impairment secondary to OI, but relatively few fractures. The family history for OI was positive with one sibling who was also diagnosed with OI and valvular heart disease. The diagnosis of OI was unclear in their father who died from an aortic rupture in his eighties.

Pre-operative trans-oesophageal echocardiography (TOE) revealed ruptured chordae with a flail medial scallop of the posterior mitral leaflet and an eccentric, anterolaterally directed regurgitant jet (Fig. 1a). The aortic valve was trileaflet but the leaflets were redundant. There was moderate aortic regurgitation (Fig. 1b). The ascending aorta was mildly dilated with a trans-sinus diameter of 42 mm. Coronary angiography showed two-vessel coronary artery disease involving the right coronary and left circumflex arteries. The patient was referred for surgery.

Surgery was performed via a median sternotomy. Antegrade cold blood cardioplegia was delivered after cardiopulmonary bypass was established with aortic and two stage venous cannulation. The mitral annulus was moderately dilated and there were ruptured chordae tendineae in keeping with the echocardiographic findings. The mitral valve was repaired by attaching two sets of Gore-Tex® loops from the posteromedial papillary muscle to the medial scallop of the posterior leaflet. A 34 mm Physio® annuloplasty ring was implanted around the mitral annulus with circumferential 2-0 Tycron® sutures. The right coronary cusp of the aortic valve was perforated and prolapsed. A running stitch with a 6-0 Gore-Tex® suture was placed on the cusps. Unfortunately the attempt to repair the aortic valve was unsuccessful. Therefore the aortic

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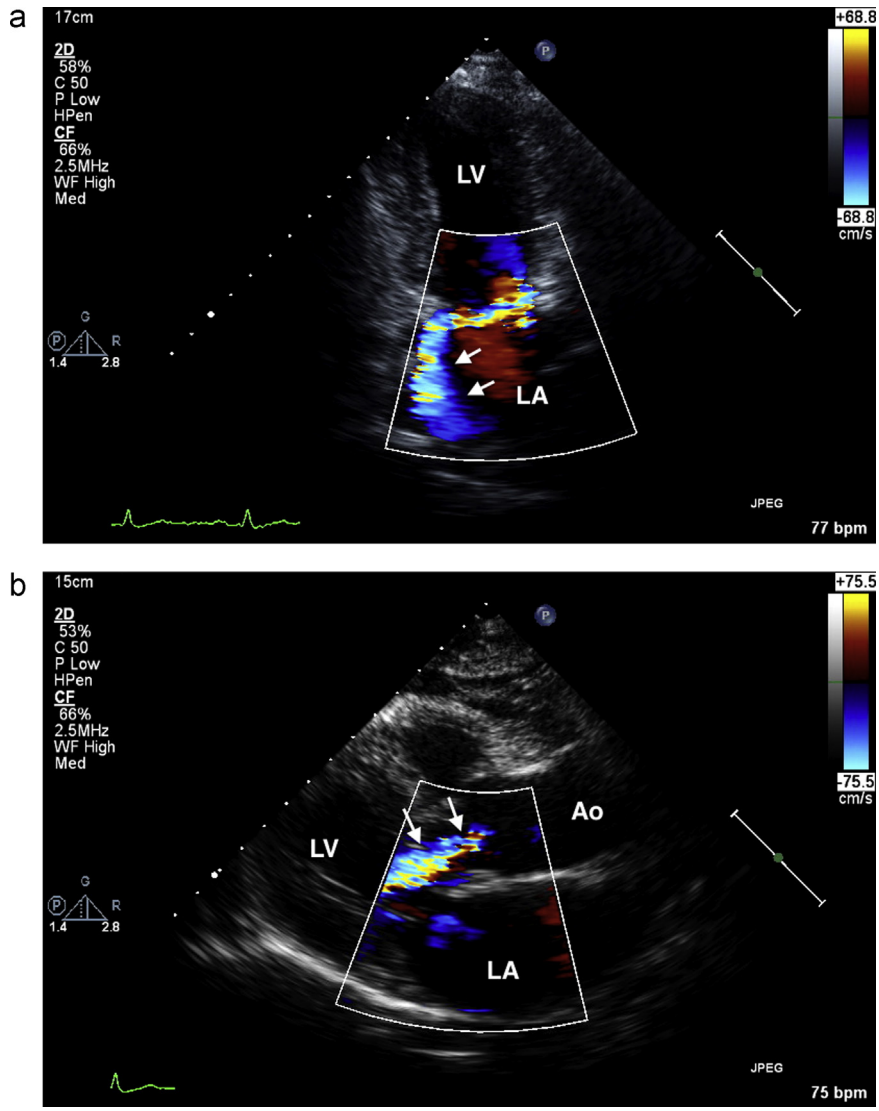

Figure 1. *Two-dimensional echocardiography with colour flow Doppler of Case 2 showing (a) an eccentric jet of severe mitral regurgitation (arrowed) and (b) moderate aortic regurgitation (arrowed). Labels: LV, left ventricle; LA, left atrium; Ao, aorta.*

valve was replaced with a 27 mm Perimount® bioprosthesis anchored with interrupted 2-0 Ethibond® sutures. Two saphenous vein grafts were constructed to the posterior descending, and circumflex obtuse marginal coronary arteries.

Sternal wound bleeding was a problem despite heparin reversal with protamine. This eventually subsided after administration of fresh-frozen plasma and platelets. Intrinsic platelet dysfunction was documented (prolongation of closure times on a PFA-100 analyser) in the context of normal platelet count and hematocrit. Platelet aggregation studies and coagulation profile were normal.

The post-operative period was otherwise uneventful except for atrial fibrillation. Sinus rhythm was restored with electrical cardioversion in the late post-operative period. The patient remains well with no cardiac symptoms and in sinus rhythm two years after his surgery.

Case 2

A 58 year-old man with OI was referred for evaluation of a heart murmur in the absence of any significant cardiac symptoms. His past medical history was notable for more than 40 fractures since the age of two years, osteoporosis, lumbar radiculopathy and hypertension.

Transthoracic echocardiography showed severe mitral regurgitation due to posterior mitral leaflet prolapse and moderate aortic regurgitation secondary to redundancy of the aortic valve leaflets. The left ventricle was not dilated (LVESD = 25 mm) and there was satisfactory systolic function (LVEF = 63%). The aortic root and proximal ascending aortic diameter were 27 mm and 38 mm, respectively (ascending aortic diameter index = 2.12 cm/m^2). The patient was further assessed with dobutamine stress echocardiography which showed good LV contractile reserve.

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