

ABSTRACT

Background

Osteogenesis imperfecta (OI) is an inherited connective tissue disorder with defective synthesis of type 1 collagen resulting in easy bruising and fractures. Cardiovascular manifestations of osteogenesis imperfecta include left sided valvular disease mainly aortic and mitral regurgitation, aortic root dilatation and left ventricular dilatation.

Case presentation

A 34-year male with blue sclera and recurrent fractures clinically diagnosed as osteogenesis imperfecta at the age of 24 years presented with worsening exertional dyspnea and paroxysmal nocturnal dyspnoea. He had clinical signs of cardiomegaly and severe aortic regurgitation. Transthoracic echocardiography confirmed severe aortic regurgitation with a dysplastic three cusp aortic valve, left ventricular dilatation and preserved LV function. He was started on diuretics and ACE inhibitors and referred for minimally invasive aortic valve replacement.

Conclusion

This case highlights the importance of active surveillance for recognized complications of diseases. Minimally invasive aortic valve replacement has revolutionized the management of OI patients requiring AVR as associated bone fragility puts OI patient at high risk for a standard approach with a midline sternotomy.