

Hypercholesterolaemic valvulopathy in a young woman with heterozygous familial hypercholesterolaemia

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DESCRIPTION

A 19-year-old woman presented with dyspnoea, angina and exertional syncope for the past 1 year. She had an ejection systolic murmur in the aortic area radiating to the carotids. She had multiple skin xanthomas over her feet, knees, hands and buttocks (figure 1). Echocardiography revealed severe calcific aortic stenosis (figure 2). Her serum low-density lipoprotein (LDL) was 391 mg/dL, while triglycerides were normal. Her father and brother also had very high LDL levels and a few xanthomas

but no features of aortic stenosis. Her mother and two sisters had normal LDL levels. The patient was thus diagnosed as having heterozygous familial hypercholesterolaemia (FH). Coronary angiography revealed plaques in the left anterior descending artery but no significant stenosis (figure 3). The aortic root was severely calcified causing valvular stenosis along with supravalvular stenosis at the sinotubular junction (figure 4 and video 1). The patient underwent successful aortic valve replacement with aortic root enlargement. Histopathological examination of the valve showed lipid deposition and calcification in the aortic cusps. She was put on high-dose statins and ezetimibe post surgery. FH is an autosomal dominant disorder in which the defect lies in the LDL receptor gene. Incidence is approximately 1:500 and 1:1 000 000 in its heterozygous and homozygous forms, respectively.¹ Affected individuals develop early coronary artery disease and severe aortic root calcification, despite optimisation of therapy. Previous studies have demonstrated 2.6–16% prevalence of supravalvular and/or valvular stenosis in patients with heterozygous FH.^{2 3} This case describes an extremely rare presence of both the lesions together in a patient with heterozygous FH with characteristic skin lesions.

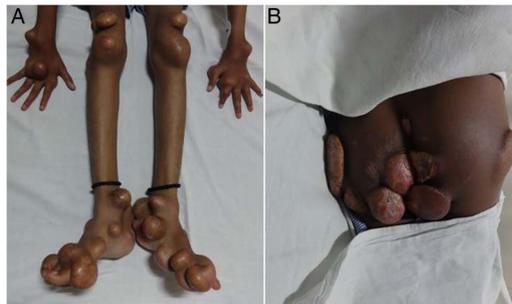


Figure 1 Multiple skin xanthomas over hands, feet, knees (A) and buttocks (B).

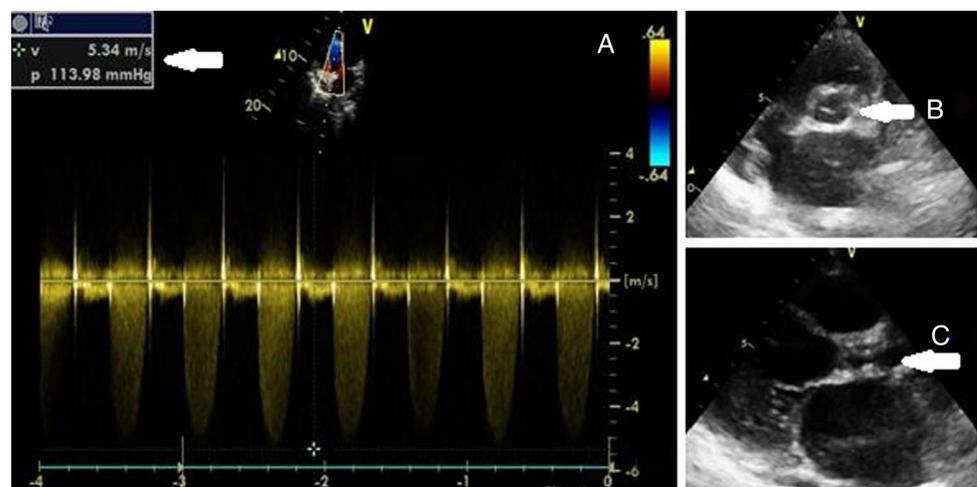


Figure 2 Echocardiogram showing a very high gradient across the aortic valve (A). Note the severely thickened and calcific aortic valve and aortic root (B and C).



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Figure 3 Coronary angiography revealing mild disease (plaquing) of proximal and mid-LAD but no significant stenosis (A–C). Left circumflex is normal. Aortic root angio showing normal right coronary artery (D). Note calcification of aortic root, outlined by multiple arrows (D).

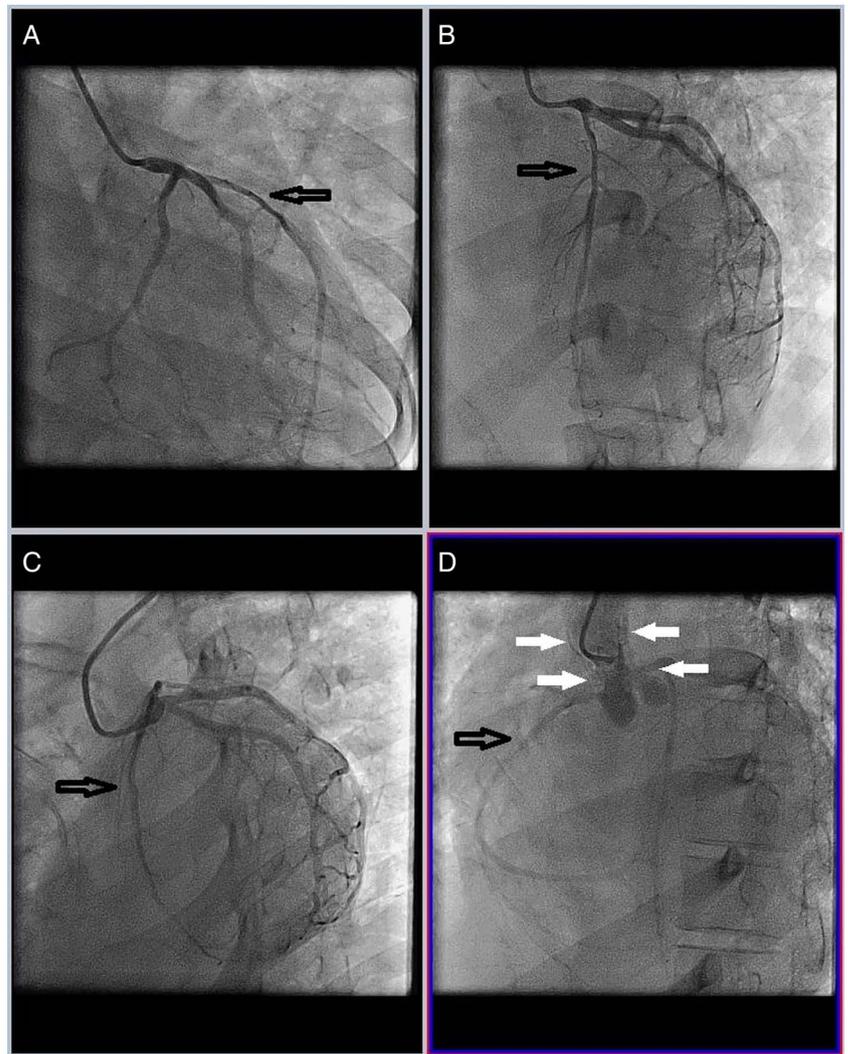
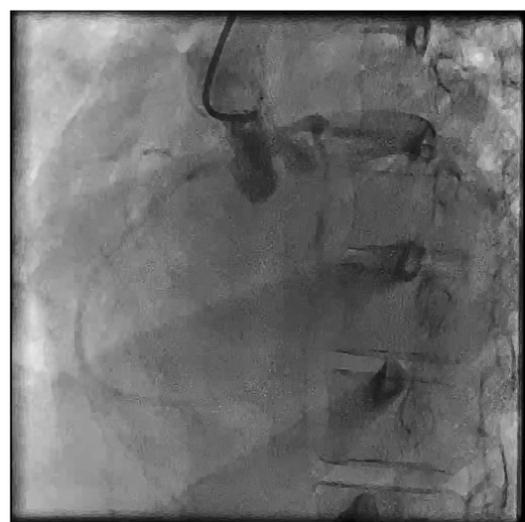


Figure 4 Heavily calcified aortic root (outlined by multiple white arrows) and narrowing at the level of the sinotubular junction (marked by black arrows) suggestive of supra-aortic stenosis.



Video 1 Aortic root angiography showing calcification of the aortic root and coronary arteries, and level of supra-aortic stenosis.

Learning points

- ▶ Heterozygous familial hypercholesterolaemia is a disorder caused by mutation in the low-density lipoprotein (LDL) receptor gene, and is characterised by high-serum LDL levels and skin xanthomas.
- ▶ Premature coronary involvement and valvulopathy are common.
- ▶ Simultaneous occurrence of supra-avalvular in addition to valvular stenosis in heterozygous familial hypercholesterolaemia is extremely rare.
- ▶ Early recognition is warranted to achieve optimal treatment results.

Competing interests None declared.

Patient consent Obtained.

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