

Cardiac Imaging

Supravalvular Aortic Stenosis in Homozygous Familial Hypercholesterolemia

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A 21-year-old asymptomatic male patient with known homozygous familial hypercholesterolemia was referred to our department for the evaluation of a heart murmur. The patient had been treated with bimonthly LDL-apheresis since the age of 9, along with lipid-lowering medical therapy (ezetimibe and rosuvastatin), which resulted in LDL cholesterol levels between 150 and 200 mg/dL. Cardiac auscultation revealed an ejection systolic murmur in the right upper sternal border that radiated to the neck. Chest X-ray and 12-lead ECG revealed normal findings, while transthoracic echocardiography identified a mild supravalvular aortic stenosis (Figure 1) with a peak Doppler velocity of 3.5 m/s and an estimated mean pressure gradient of 27 mmHg (Figure 2). CT angiography confirmed the diagnosis and revealed a supravalvular aortic wall atherosclerotic thickening of up to 3.6 mm, consisting of predominantly soft plaque with scattered calcifications (Figure 3). The plaque extended from the level of the sinuses of Valsalva to the sinotu-

bular junction, and resulted in a narrowed aortic lumen measuring 1.8×1.7 cm (Figure 4).

Premature malignant atherogenesis leading to aortic valve and root abnormalities is a recognized complication of homozygous familial hypercholesterolemia.¹⁻³ Noninvasive imaging should be included in the routine assessment of the disease, given that hemodynamically severe involvement may require surgical intervention.

References

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Figure 1. Two-dimensional parasternal long-axis echocardiographic image demonstrating the aortic valve (large arrow) and mild supravalvular aortic stenosis at the level of the sinuses of Valsalva (small arrow).

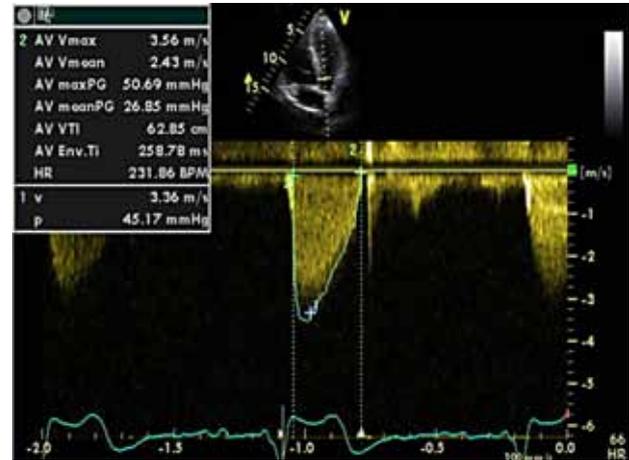


Figure 2. Continuous wave Doppler showing the pressure gradient across the level of the stenosis.

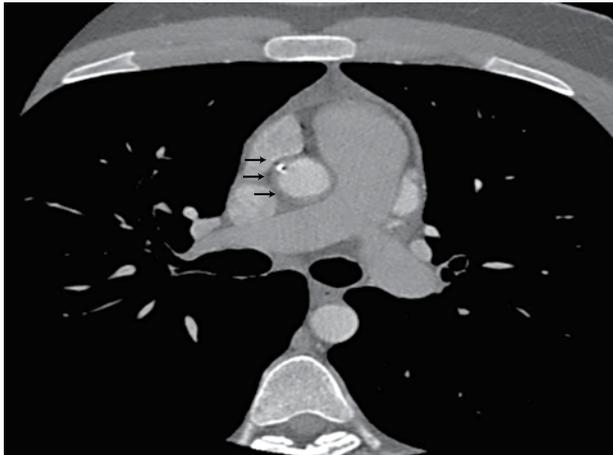


Figure 3. Cardiac CT demonstrating the soft atherosclerotic plaque (arrows).

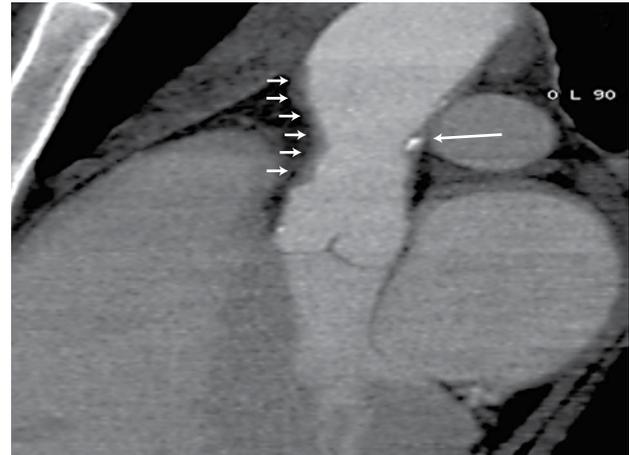


Figure 4. Aortic wall atherosclerotic lesion extending from the level of the sinuses of Valsalva to the sinotubular junction (small arrows) with calcification (large arrow).