A mimic of hypertrophic cardiomyopathy

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A 52-year-old man with atrial fibrillation, chronic kidney disease (status-post renal transplant), and a presumed diagnosis of hypertrophic cardiomyopathy (HCM) underwent cardiac magnetic resonance (CMR) prior to catheter ablation for refractory atrial fibrillation. An electrocardiogram during sinus rhythm suggested an accessory pathway (Panel 1). The CMR revealed normal ejection fraction (Panel 2A, Supplementary material online, Movie S1), concentric left ventricular hypertrophy with wall thicknesses of 16–18 mm (Panel 2B), moderate mitral and tricuspid regurgitation, and patchy transmural late gadolinium enhancement (LGE) predominantly involving the basal inferolateral wall (Panel 2C).

On the basis of the CMR findings and electrocardiogram, endomyocardial biopsy was performed and yielded results consistent with Fabry’s disease, ultimately confirmed by a leucocyte alpha-galactosidase level of 0.4 nmol/mg/h (normal > 23.1 nmol/mg/h). The patient was started on enzyme replacement therapy.

Fabry’s disease is characterized by accumulation of globotriaosylceramide in affected organs, resulting in myocardial cell hypertrophy and leading to a phenotype that can be indistinguishable from HCM by traditional imaging modalities.

Cardiac magnetic resonance can aid in the diagnosis of Anderson-Fabry’s disease on the basis of LGE imaging, which usually demonstrates mid-wall enhancement of the basal inferolateral wall. Our case highlights the importance of considering mimics to HCM, particularly when there exists multi-organ disease.

Panel A: Electrocardiogram showing sinus rhythm, right axis deviation, left ventricular hypertrophy, suggestion of right ventricular hypertrophy, and possibility of accessory pathway.
CMR imaging
Panel B: Four-chamber cine image showing biventricular enlargement and pleural effusion.
Panel C: Short-axis cine showing concentric hypertrophy.
Panel D: Contrast-enhanced CMR showing transmural LGE in the lateral wall.
Histopathology
Panel E: Haematoxylin and eosin stain shows perinuclear vacuolization of myocytes (red arrow).
Panel F: Immunohistochemical staining for LMP2 to exclude Danon’s disease.
Panel G: Toluidine blue staining highlights extensive vacuolization of myocytes and endothelium.
Panel H: Electron microscopy reveals lamellar bodies (blue arrow) within the vacuoles.

Supplementary material is available at European Heart Journal online.