Fabry disease deposition mimicking a cardiac tumour and precipitating heart block

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A 39-year-old female presented with syncope. She has Fabry disease (del.Ile239 mutation) on enzyme replacement therapy and also has a renal transplant secondary to Wegener’s granulomatosis. An electrocardiogram demonstrated a complete heart block with a slow ventricular escape rhythm. Cardiovascular magnetic resonance imaging showed global left ventricular hypertrophy with more prominent thickening of the basal septum, left ventricular outflow tract (LVOT) and aortic root, and increased T₂ signal intensity in these areas (Panel A). Late gadolinium enhancement was observed in the basal inferolateral myocardium (Panel B, white arrow), the classic pattern of Fabry disease, and also in the basal septum in the vicinity of the Bundle of His (Panel B, black arrow and Panel C, Bundle of His indicated by star). A dual-chamber permanent pacemaker was inserted. Months later, an echocardiogram showed increased thickening of the LVOT myocardium and interatrial septum, which appeared like a mass on transoesophageal echocardiography (TOE) (Panels D—F). A computed tomography/positron emission tomography scan showed a soft tissue mass surrounding the aortic root (Panel G) with extension into the interatrial septum that demonstrated increased FDG uptake (Panel H), suggestive of a neoplasm or active inflammatory mass. However, TOE-guided endomyocardial biopsy excluded neoplasia and demonstrated typical histological features of cardiac Fabry disease including marked sarcoplasmic vacuolization (Panel I—H&E stain, original magnification 600 × ).

This case demonstrates the ongoing importance of multimodality assessment in complex cardiac pathology.

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