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IMAGES IN CARDIOLOGY

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Hypertrophic cardiomyopathy and left ventricular hypertrabeculation: evidence for an overlapping phenotype

Two female siblings (A and B) were referred for clinical screening following the sudden death of their mother aged 45 years from hypertrophic cardiomyopathy (diagnosed post-mortem). The sisters complained of fatigue, exertional dyspnoea (New York Heart Association functional class II), and infrequent palpitations. Their 12 lead ECGs are shown in panels A and B.

In patient A, the echocardiogram demonstrated asymmetric hypertrophy of the interventricular septum (maximum thickness 25 mm) with prominent trabeculations distally. In patient B there was very extensive hypertrabeculation in the mid and apical segments of the left ventricle with two myocardial layers visible in the parasternal short axis views (ratio of non-compacted to compacted layer 2.3). Their echocardiographic images are shown in panels C and D.

Left ventricular hypertrabeculation or left ventricular non-compaction (LVNC) is an increasingly recognised myocardial disorder characterised by excessive and prominent trabeculation of the left and/or right ventricle. Rare familial cases caused by mutations in cytoskeletal proteins (dystrobrevin, dystrophin), nuclear envelope proteins (emerin, lamin A/C), and tafazzin are described, but in the majority of adult patients with familial disease, the cause is unknown. In most adults with LVNC, the left ventricle is normal in size or dilated with impaired systolic function. Cases of hypertrophy are described in children, but we are unaware of any reports of adults within the same family with LVNC and hypertrophic cardiomyopathy. This family suggests that some cases of LVNC and hypertrophic cardiomyopathy may share a novel molecular mechanism related to disordered cardiac morphogenesis.

(A) The 12 ECG of sibling A shows right axis deviation with repolarisation abnormalities in inferior leads and V6 with deep S waves in leads V1–V3. (B) The ECG of sibling B shows normal axis with negative T waves in inferior and precordial leads.

(C) Apical four chamber view of sibling A demonstrating asymmetric hypertrophy of the interventricular septum with prominent trabeculations distally. (D) The same view of the younger sister (sibling B) revealing prominent distal septal, and apical and free wall hypertrabeculation.

A A Pantazis
S K Kohli
P M Elliott
perry.elliott@uclh.nhs.uk