encouraged. Further studies are required to identify whether and how subjects with SVPCs should be treated to improve their cardiovascular outcome.

Supplementary material

Supplementary material is available at European Heart Journal online.

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References

The list of references is available in the online version of this paper.

CARDIOVASCULAR FLASHLIGHT

A novel MYH7 mutation in a family with cardiomyopathy presenting with restrictive physiology and varying degrees of left ventricle hypertrophy

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The proband, a 25-year-old female, was presented to our institution with exertional dyspnoea and oedema in lower extremities. Her electrocardiogram showed prominent dilated atria 2 years ago (Panel A), followed by sustained atrial fibrillation 1 year later. Transthoracic echocardiography revealed preserved systolic function, massive biatrial enlargement, and moderate left ventricular hypertrophy (LVH) localized to the mid-septum (maximal thickness = 18 mm) with no left ventricular outflow tract obstruction (Panels B and C, arrow). Cardiac CT ruled out constrictive pericarditis. Cardiac catheterization revealed a normalized left ventricle with a ‘dip and plateau’ ventricular filling pattern. Endomyocardial biopsy obtained from the right ventricular septum showed myocyte degeneration and disappeared complicated by obvious interstitial fibrosis (Panel D, arrow), no myocyte hypertrophy and disarray. All the members of her family were studied, which comprised four affected individuals aged 10–52 years presenting with restrictive phenotype but with minimal or no LVH (maximal mid-septum thickness range 10–13) (Panel E). Genetic analysis of all family members identified a novel mutation (c.755T>C/p.F252S) (F) in exon 9 of sarcomeric cardiac beta-myosin heavy chain gene (MYH7) in four clinically affected individuals.

Despite variable degrees of LVH, all affected members of our family exhibited restrictive physiology characterized by echocardiography and haemodynamic catheterization. Thus, it is conceivable that this mutation of MYH7 can present with restrictive cardiomyopathy (RCM) or mixed cardiomyopathy (RCM plus hypertrophy) within the same family, implying the necessity of comprehensive cardiac assessment in familial cardiomyopathy screening.

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