CLINICAL IMAGE

Cardiac magnetic resonance imaging in a woman suspected of hypertrophic cardiomyopathy based on genotyping

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Hypertrophic cardiomyopathy (HCM) is a common genetic cardiac disease with a diverse clinical phenotype and imaging results; therefore, genotyping is of key importance in establishing a proper diagnosis.[1] To illustrate these diagnostic challenges, we report a case of a 61-year-old woman referred for family (including genetic) screening after diagnosing her daughter with a fully developed phenotype of HCM and typical echocardiographic findings at the age of 30 years. An MYBPC3 c.2541 C>G, p.(Tyr847*) mutation in the myosin-binding protein C gene was detected in the proband (FIGURE 1A).

The MYBPC3 p.Tyr847* mutation was discovered in 2004. In the ClinVar database, it has been reported several times in HCM patients from different countries as pathogenic or likely pathogenic. In the mother of the proband, echocardiography failed to show typical abnormalities, partly due to problems with proper visualization and measurements (the maximum wall thickness was about 12–13 mm without left ventricular outflow tract obstruction). The patient was asymptomatic.

Electrocardiography (FIGURE 1B) showed no signs typical of HCM (according to the European Society of Cardiology guidelines).[1] On two 24-hour Holter examinations, 3 consecutive ventricular beats were recorded (a short episode of nonsustained ventricular tachycardia with a heart rate of 138 bpm). No sudden death was reported in family history. To improve the quality of imaging, we used cardiac magnetic resonance (CMR) imaging with gadolinium contrast. It demonstrated left ventricular hypertrophy suggestive of HCM: diastolic maximum wall thickness in the inferior wall of 15 mm and late gadolinium hyperenhancement in the segment with maximum thickness (FIGURE 1C and 1D). Left ventricular ejection fraction calculated from high-quality CMR images was normal (79%). On a treadmill exercise test, the patient achieved a workload of 7 metabolic equivalents at a maximum heart rate of 86%. Maximum oxygen uptake was normal (96% of the predicted value). The test was terminated due to fatigue. The asymptomatic status corresponded with normal results of exercise

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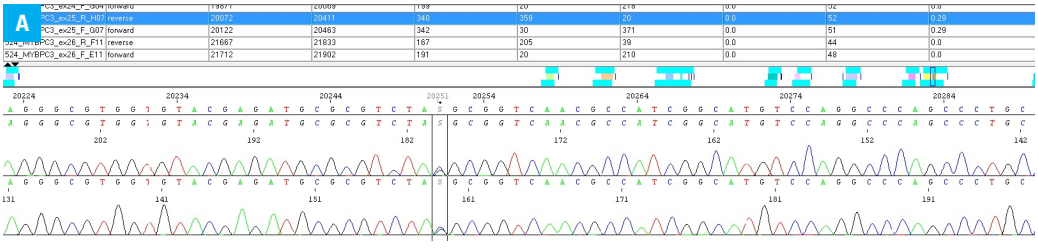

FIGURE 1 Examinations in a patient suspected of hypertrophic cardiomyopathy: **A** – direct sequencing of gene encoding

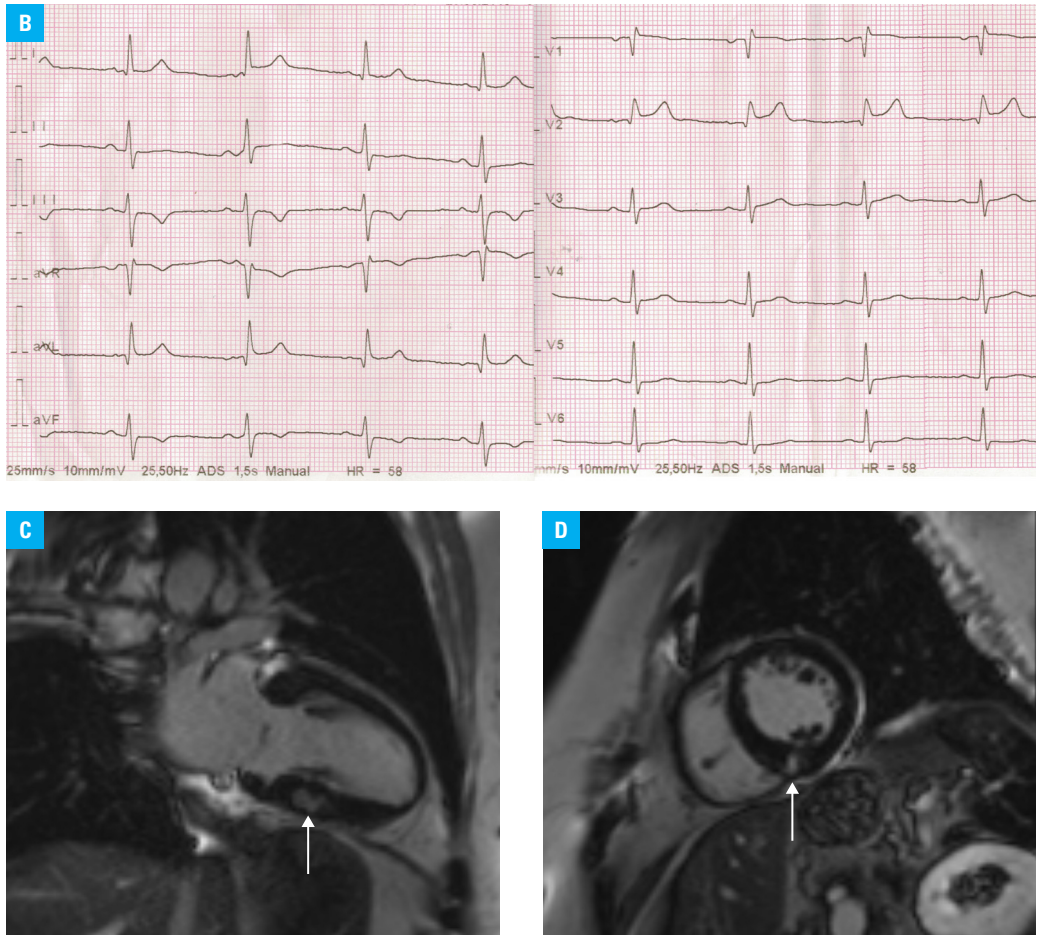

FIGURE 1 Examinations in a patient suspected of hypertrophic cardiomyopathy (HCM): **B** – electrocardiograms showing no signs typical of HCM; **C** – cardiac magnetic resonance (CMR) image: hypertrophied myocardial segment with late gadolinium enhancement (arrow; long-axis view); **D** – CMR image: hypertrophied myocardial segment with late gadolinium enhancement (arrow; short axis-view)

test (96% of the predicted value of maximum oxygen uptake).

The relevance of CMR and exercise testing in a differential diagnosis of HCM was reported previously.[2] CMR imaging is helpful in 20% of HCM patients in whom echocardiography is inconclusive, especially when showing borderline left ventricular hypertrophy.[3] CMR imaging allows also a visualization of pathological late gadolinium enhancement (potential arrhythmogenic substrate).[4] To our knowledge, only one paper reported the use of CMR imaging in a single patient with this mutation, but no typical features of the late gadolinium hyperenhancement pattern were described.[5] In conclusion, CMR imaging may be helpful when diagnosis is uncertain, for example, when the proband's mother is suspected of HCM and HCM is confirmed in the proband using genetic testing. However, this case shows that genotyping should be performed in case of inconclusive imaging findings.

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