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CASE REPORT

Hypertrophic cardiomyopathy with heart failure and ST-segment elevation of the lateral wall

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Abstract

Hypertrophic cardiomyopathy (HCM) is the most common cardiovascular disease that is inherited from a single gene. Its clinical manifestations range from asymptomatic mutant gene carriers to patients with severe left ventricular effluent tract obstruction and end-stage HCM with motor restriction. In this case, we present a patient with the main presentation of heart failure and ST-segment elevation of the lateral wall, as determined by electrocardiogram. The patient was finally diagnosed with HCM because of genetic testing and the presentation of extensive myocardial fibrosis with reduced systolic function on cardiac magnetic resonance imaging. The patient's clinical findings, electrocardiogram, and cardiac magnetic resonance imaging were different from those of typical patients with HCM.

Keywords Hypertrophic cardiomyopathy; Heart failure; ST-segment elevation; Cardiac magnetic resonance; Genetic test

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Introduction

Hypertrophic cardiomyopathy (HCM) is the most common cardiovascular disease that is inherited from a single gene. The clinical manifestations of HCM range from asymptomatic mutant gene carriers to patients with severe left ventricular effluent tract obstruction and end-stage HCM with motor restriction.[1] The majority of cases of HCM and chronic, drug-resistant heart failure are mainly caused by left ventricular outflow tract obstruction, but a small number of cases without obstruction still have progressive end-stage heart failure.[2] Most of these cases have unique phenotypic alterations that lead to systolic pump failure, which is often accompanied by ventricular dilation, increased end-diastolic or end-systolic volume, and hypertrophy regression due to diffuse myocardial scars.[3,4] Cardiac magnetic resonance (CMR) multisequence and multiparameter imaging provide information on cardiac morphology, function, myocardial perfusion, and myocardial tissue characteristics.[5,6] Genetic testing for HCM and other genetically unknown cardiac hypertrophies (HCM phenotype) is also recommended for the diagnosis and prognosis of patients with atypical clinical manifestations of HCM or when other genetic diseases are suspected.[5,6]

Case report

A 41-year-old male with no history of cardiovascular disease presented to our hospital due to exhaustion after activity for more than 1 month. At admission, the patient's temperature was 36.4°C, his blood pressure was 148/108 mmHg, his pulse was 108 per minute, and his respiratory rate was 20 per minute. Physical examination revealed engorgement of the neck veins, enlargement of the heart boundary, and diastolic gallop rhythm. The electrocardiogram (ECG) at admission suggested ST-segment elevation and pathological Q waves in leads I, II, AVL, and V5-V6, with fragmented QRS waves in leads II, V5 and V6 (*Figure 1A*). Laboratory studies revealed increased levels of cardiac troponin-T (44.3 ng/L, reference 0–14 ng/L) and N-terminal brain natriuretic peptide (1911 ng/L, reference 0–88 ng/L). Lateral myocardial infarction was first suspected; however, coronary angiography demonstrated only mild atherosclerotic plaque formation. The echocardiogram suggested that the whole heart was enlarged, the anterior wall of the right ventricle and ventricular septum thickened, and the systolic (ejection fraction 35%) and diastolic (mitral E/e' 25) function of the left ventricle was significantly reduced (*Figure 1B*).

Cardiomyopathy or myocardial amyloidosis was suspected, and CMR was completed to confirm the diagnosis. The CMR illustrated that the left atrium and the left ventricle were enlarged and the left ventricular wall was thickened (*Figure 2A*). There was a small perfusion defect in the septal wall under first-pass perfusion, and the de-

Figure 1 Electrocardiogram and echocardiogram of the patient at admission. (A) 12-lead electrocardiogram indicated ST-segment elevation (blue arrows) and pathological Q waves (green arrows) in the leads I, II, aVL, V5, and V6, ST-segment depression (red arrows) in aVR and V1, with fragmentation QRS waves (orange arrows) in leads II, V5, and V6. (B) Still frame of apical four-chamber and left ventricle short axis view suggested the whole heart enlarged, anterior wall of the right ventricle, and ventricular septum (yellow arrows) thickened.

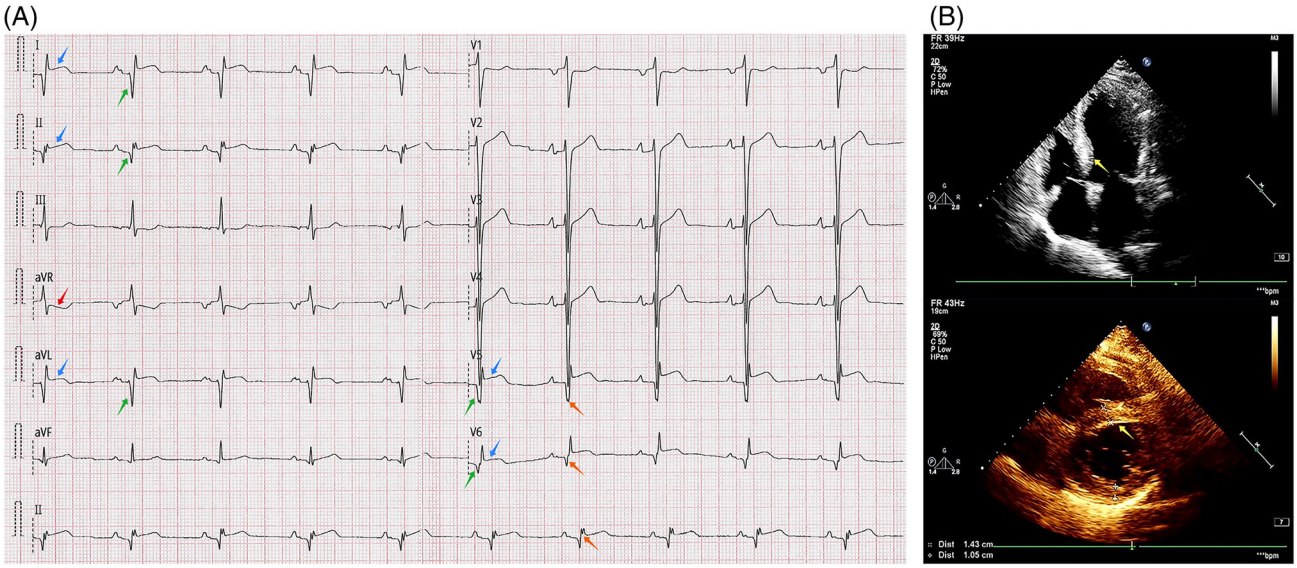

Figure 2 Cardiac magnetic resonance of the patient. (A) True fast imaging with steady-state precession of cardiac magnetic resonance. Blue arrows, the thickened left ventricular wall. (B) Phase-sensitive inversion recovery imaging of cardiac magnetic resonance. Green arrows: A little perfusion defect in the septal wall under first-pass perfusion. Orange arrows: The delayed scan showed diffuse and transmural abnormal enhancement of the left ventricle, with sheet-like abnormal enhancement of the right ventricle.

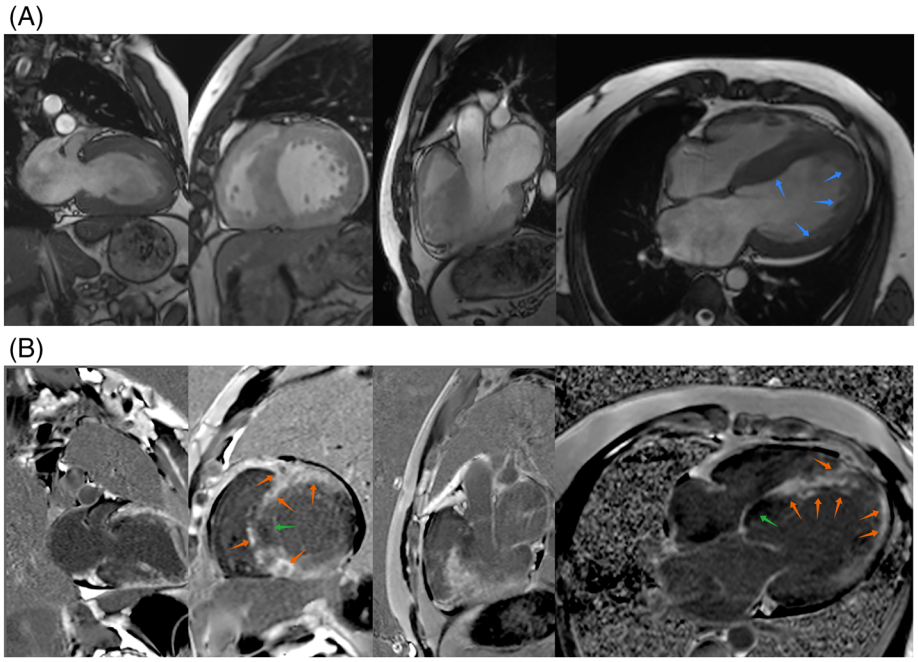

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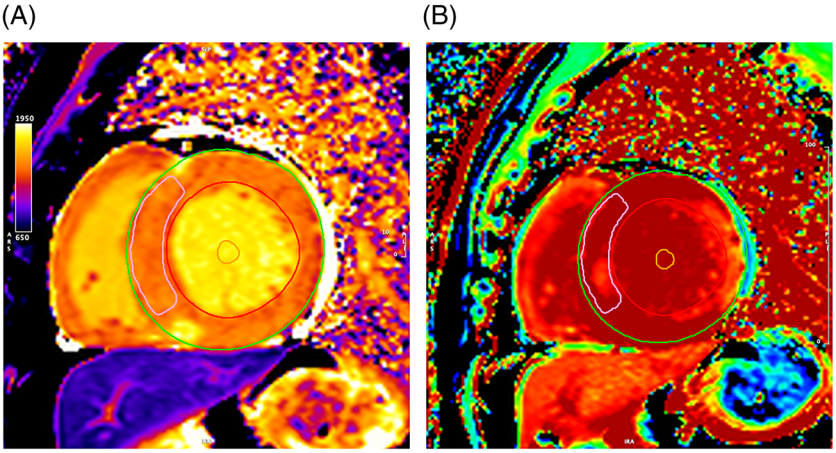
Figure 3 Cardiac magnetic resonance T1 mapping of the patient. (A) Native myocardial T1 image of the patient. (B) Quantitative extracellular volume fraction image of the patient.



layed scan suggestive of extensive fibrosis showed diffuse and transmural abnormal enhancement of the left ventricle. It was dominated by subepicardium and progressing to endocardium, with sheet-like abnormal enhancement of the right ventricle (*Figure 2B* and *Video S1*). The cardiac movie indicated weakened heart movement and significantly reduced systolic function of both ventricles (left ventricular EF: 18.7%, right ventricular EF: 34.5%) (*Video S2*). Moreover, T1 mapping indicated a native T1 value of 1415 ms with an extracellular volume of 67% (*Figure 3*).

Myocardial amyloidosis and non-ischaemic cardiomyopathy still could not be distinguished. Further analysis showed no obvious abnormality in the quantification of kappa light chain and lambda light chain from blood and urine. Serum protein electrophoresis also indicated no increase in immunoglobulin M. Genetic testing demonstrated that the MYBPC3 gene inherited from his mother had a mutation that changed a cytosine to thymine at nucleotide 622 (c.622C>T), causing a nonsense amino acid mutation (p.Q208X). The genetic background also suggested autosomal dominant inheritance.

Myocardial biopsy or implantable cardioverter defibrillator implantation was refused. The patient was eventually diagnosed with HCM and treated with sacubitril/valsartan, bisoprolol fumarate, spironolactone, furosemide, and statin.

Discussion

In cases of HCM, ECG is the most sensitive routine test with only 6% of HCM patients demonstrating a normal ECG.[7] However, ECG lacks specificity.[7] ECG abnormalities usually include abnormal P wave, Q wave (especially in the inferior and lateral leads), left axis deviation, and deeply inverted T waves in V2-V4 leads (patients with apical HCM).[8] ST-segment elevation in HCM is relatively rare, and the occurrence may be due to apical aneurysm (particularly in V4–V6 leads) and left ventricular hypertrophy (especially in V1–V3 leads).[9] However, HCM characterized by ST-segment elevation of the lateral wall as the main manifestation, such as this case, is very rare. In addition, extensive myocardial fibrosis may be another reason for myocardial inactivation, resulting in ST-segment elevation. The ST-segment elevation in this case could be due to the disorder of the arrangement of hypertrophic cardiomyocytes and interstitial fibrosis, which blocks the depolarization of local myocardium and the generation of locally oriented ST vectors. It could also be due to the abnormal polarization of hypertrophic abnormal cardiomyocytes that produces injury current similar to that of acute myocardial infarction. The fragmented QRS waves in leads II, V5, and V6 may be another ECG manifestation of extensive myocardial fibrosis.

Another interesting feature of this case is that the patient's CMR findings are mainly characterized by diffuse delayed enhancement of the left ventricle involving the right ventricle. These findings are similar to the findings of myocardial amyloidosis but not typical of HCM. This increases the difficulty of diagnosis. Late gadolinium enhancement in typical HCM mainly results from patchy delayed enhancement of the middle wall, especially at the junction between the interventricular septum and the free wall of the right ventricle, whereas myocardial amyloidosis often shows diffuse delayed enhancement.[10]

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Conflict of interest

None.

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Supporting information

Additional supporting information may be found online in the Supporting Information section at the end of the article.

Video S1. Delayed scan of cardiac magnetic resonance.
Video S2. The cardiac movie of the patient.

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