

Cardiac MRI in the Diagnosis and Prognosis of patients with Hypertrophic Cardiomyopathy (HCM) - A Case Report

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Abstract

Hypertrophic cardiomyopathy (HCM) is a condition of genetic mutation in the cardiac sarcomere that is defined by cardiac hypertrophy, a non-dilated left ventricle, and a normal or increased ejection fraction. It presents with a heterogeneous clinical picture which continues to challenge clinicians to diagnose it clinically. Cardiac magnetic resonance (CMR) - a non-invasive imaging technique is an essential diagnostic tool that reliably confirms the diagnosis, differentiates hypertrophic cardiomyopathy from other etiologies of left ventricular hypertrophy as well as identifies the individuals who are most at risk of sudden cardiovascular events. We present a case of 15 years old female patient with complaints of left-sided chest pain, dyspnea, palpitations, and orthopnea with a history of several episodes of syncopal attacks in the past. Following echocardiography, cardiac magnetic resonance imaging was done for the proper interpretation of the presenting clinical manifestations.

Keywords: Hypertrophic cardiomyopathy, Cardiac Magnetic Resonance, Left ventricular outflow tract obstruction, Systolic anterior motion of the mitral valve.

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Introduction

Hypertrophic cardiomyopathy (HCM) is a sarcomere protein-related cardiomyopathy defined by cardiac hypertrophy, a non-dilated left ventricle, and a normal or increased ejection fraction.¹ Genetic mutation in the cardiac sarcomere may eventually result in left ventricular hypertrophy (LVH) and fibrosis as a result of increased myocyte strain and compromised function.² HCM is identified as a diffuse or segmental LVH irrelevant to other cardiac or systemic illnesses that induce hypertrophy of the left ventricle while the right ventricle may as well be occasionally involved. The clinical manifestations of HCM vary from being asymptomatic to sudden cardiac death where the four major associated pathophysiologic scenarios are diastolic ventricular dysfunction, obstruction to the left ventricular outflow tract (LVOT), the discrepancy between myocardial oxygen supply and demand, and cardiac arrhythmias.¹ Hence, the wide range of clinical symptoms makes it challenging to diagnose HCM clinically. However, diagnosing HCM has been much easier in recent years after the introduction of cardiac magnetic resonance (CMR) - a non-invasive imaging technique that is increasingly recognized as an essential diagnostic tool in the analysis of suspected HCM. CMR can reliably confirm the diagnosis, differentiate HCM from other etiologies of LVH, as well as identify the individuals who are most at risk of sudden cardiovascular events.³ Here we present a case of HCM diagnosed with the help of CMR performed at our center.

Case report

A 15 years old female patient was brought to our emergency department with complaints of left-sided chest pain, dyspnea, palpitations, and orthopnea. She was given a history of several episodes of syncopal attacks in the past. On physical examination, she had an irregular pulse rate with tachycardia and systolic murmur. Strong precordial impulse and vigorous peripheral arterial pulses were evident. Her baseline investigations showed echocardiographic (ECG) changes suggestive of left ventricular hypertrophy. Further, echocardiography (ECHO) was done which showed concentric hypertrophy of the left ventricle with presence of left ventricular outflow tract obstruction. Hence, the patient was referred for a CMR study to confirm the diagnosis and to look for the prognostic parameters.

Imaging findings: Cardiac MRI with contrast was performed for the patient on a 3Tesla platform. The CMR sequences of transverse black blood and bright blood images, vertical long axis, four-chamber, short-axis cine images, left ventricular outflow tract (LVOT) views along with three-chamber cine images were obtained. Phase-contrast imaging of the aorta and pulmonary artery was done for flow quantification. The delayed gadolinium enhancement phase-sensitive inversion recovery (PSIR) sequences were obtained in short axis, four-chamber, and vertical long-axis views. The imaging findings showed diffuse thickening of the left ventricular wall involving the basal, mid cavity, and apical segments. The thickening was concentric but asymmetrical in distribution predominantly involving the basal interventricular septum (Figure 1). Maximal

left ventricular wall thickness was measured to be 44.1 mm. There was no evidence of motion abnormalities such as hypokinesia, dyskinesia, or akinesia. There was no evidence of apical insertion of the papillary muscles or abnormal insertion of the papillary muscles into the mitral valve or into the septum. Patchy scattered mid-myocardial delayed gadolinium enhancement (DGE) was noted in the left ventricular myocardium predominantly involving the asymmetrically thickened septal segments (Figure 2). The systolic anterior motion (SAM) of the mitral valve was noted leading to obstruction of the LVOT. There was no complete obliteration of the mid cavity and the apical cavity noted during the systole or diastole. There was no dilatation of the left ventricle and left ventricular ejection fraction (LVEF) was noted to be 63%. Mild mitral regurgitation was noted. The percentage of the myocardial scar was calculated by semiautomatic segmentation of the myocardium and scar contours in late gadolinium enhancement images which showed a percentage enhanced volume of 4% representing the myocardial fibrosis (Figure 3).

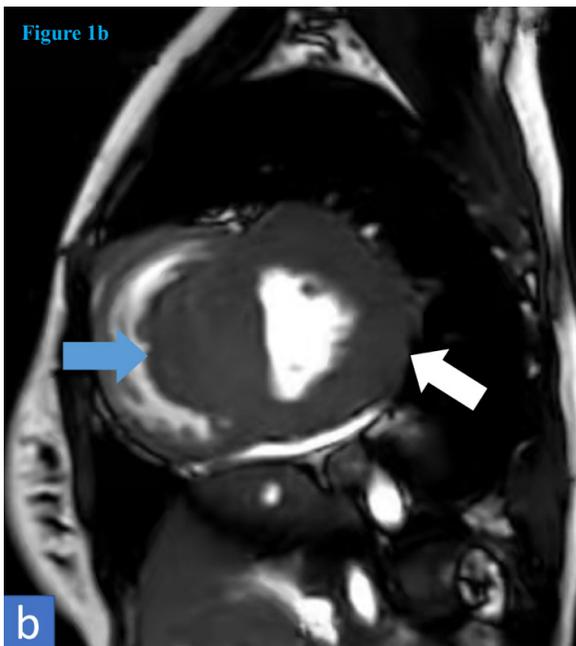
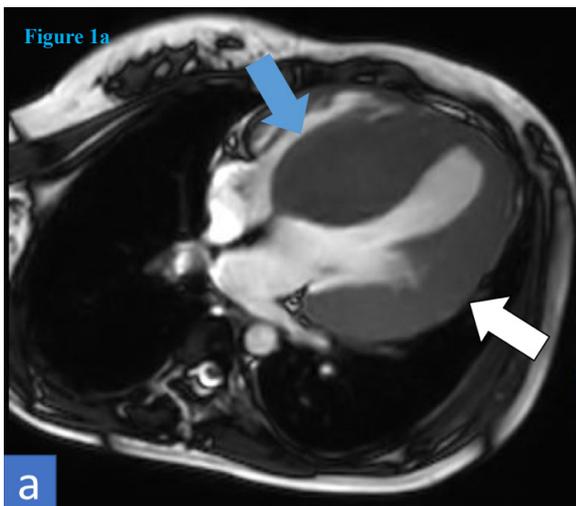


Figure 1. Cardiac Magnetic Resonance (CMR) bright blood images four-chamber view (a) and short-axis view (b) showing diffuse concentric thickening of the left ventricular wall (white arrows), predominantly involving the basal interventricular septum (blue arrows).

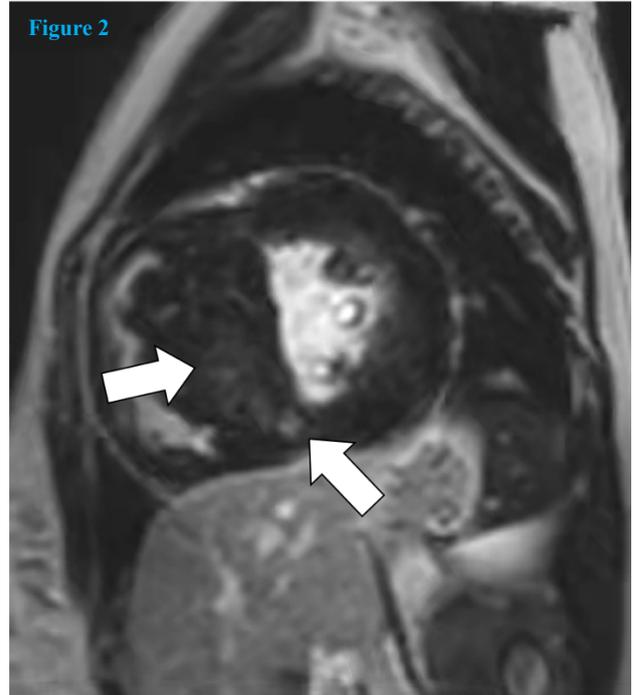


Figure 2. Cardiac Magnetic Resonance (CMR) post-contrast delayed gadolinium-enhancement (DGE) image short-axis view showing patchy scattered mid-myocardial delayed enhancement in the left ventricular myocardium (arrows).

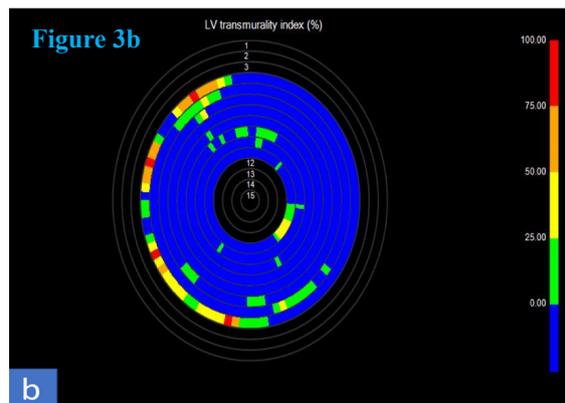
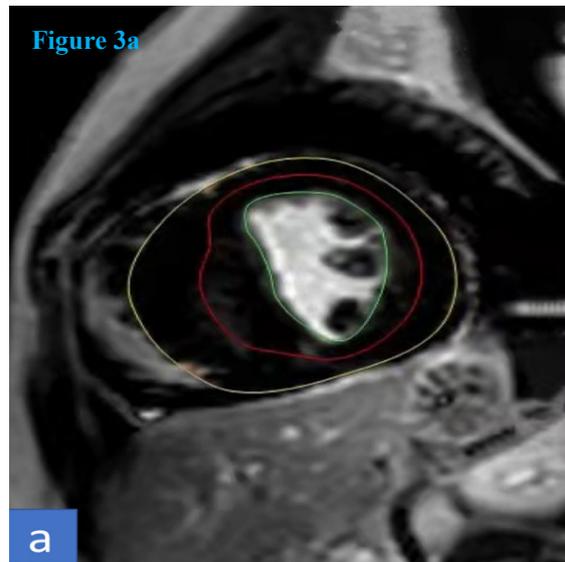


Figure 3c

Results Summary	
	Values
Myocardial mass	244.66 gr
Enhanced mass	9.91 gr
Percentage enhanced volume	4 %
Myocardial volume	233012 mm ³
Enhanced volume	9339 mm ³
Image type used	M,FFE
Threshold method	FVMH
Method parameters	Min Threshold Value: 4, Max Threshold Value: 346
No Reflow volume	0 mm ³
No Reflow mass	0.00 gr

Figure 3. Segmentation of late gadolinium enhancement image in short-axis view (a and b) showing segmentation of myocardium and scar contours, (c) displays the summary of the enhanced myocardial mass and the fibrotic burden.

Discussion

HCM and its subtypes: Hypertrophic cardiomyopathy (HCM) is the most prevalent genetic cardiac condition seen in about 0.2-0.5% of the overall population. It is considered one of many causes of sudden cardiac death among young individuals, especially athletes.^{3,4} The most widely used diagnostic parameter for HCM is a maximal left ventricular wall thickness of ≥ 15 mm in the end-diastolic phase, which can also present with lesser degrees of wall thickening (13 – 14 mm).^{5,6} HCM can affect any part of the left ventricle with a highly variable morphologic picture. It can be asymmetric (interventricular/septal), symmetric (concentric), apical, midventricular, mass-like LVH, and end-stage or burned-out HCM.⁷ The most prevalent form of HCM is basal asymmetrical (interventricular/septal) involvement with the second most common being symmetric (concentric) one.

CMR features: Asymmetric HCM usually involves asymmetrical hypertrophy of basal interventricular septum commonly associated with dynamic LVOT obstruction. The LVOT obstruction is induced by a combination of septal hypertrophy and systolic anterior motion (SAM) of the mitral valve, where the mitral valve leaflets are driven toward the septum by increased flow velocities in an LVOT, blocking the outflow tract. The elongated leaflets and aberrant placement of the mitral valve apparatus cause the atypical SAM of the mitral valve. Such misaligned mitral leaflet lengths and associated systolic anterior motion of the mitral valve may cause mitral regurgitation.^{1,6,7} Furthermore, using the delayed gadolinium enhancement sequences, CMR can differentiate an infarcted or fibrotic myocardium from normal healthy myocardium. HCM may frequently be associated with fibrotic changes affecting the myocardium which is usually depicted in the cardiac MR study in the form of mid myocardial patchy delayed GAD enhancement that is not restricted to any particular vascular territory.⁸

Concentric HCM generally represents a diffusely hypertrophied left ventricle (LV) with decreased LV cavity. The differentials causing symmetrical thickening of LV wall such as athlete's heart, sarcoidosis, amyloidosis, Fabry disease, hypertrophy secondary to hypertension, or aortic stenosis need to be identified from concentric or symmetric HCM. This is where CMR facilitates the possible accurate detection of HCM through the myocardial hypertrophy patterns and characteristic DGE features.^{3,6,7}

However, a combined form of concentric HCM along with asymmetrical involvement of basal interventricular septum is not an uncommon finding. A symmetric or concentric variant associated with an asymmetric septal involvement may lead to narrowing of the left ventricular cavity and an invasion of LVOT, along with associated SAM of the mitral valve, and patchy mid myocardial delayed gadolinium enhancement as depicted in our case.

Apical HCM represents localized hypertrophy of the apex of the left ventricle (LV) giving off a spade-like configuration of the LV cavity. It may be associated with complications such as apical infarctions and aneurysm formation. A delayed enhancement in CMR DGE images confirms the presence of apical aneurysms and helps predict prognosis in apical HCM.⁷

Midventricular HCM is an uncommon form of LV hypertrophy predominantly involving the middle third of the left ventricle

wall. On CMR, this variant together with thinning of the apex causes the LV cavity to resemble an hourglass or dumbbell-shaped configuration. Among various phenotypes of HCM, apical and midventricular types are associated with apical LV aneurysms with a higher risk of thromboembolic events.⁷

Mass-like HCM is characterized by focal segmental hypertrophy of the LV wall mimicking a cardiac mass. CMR DGE sequences are vital to differentiate between mass-like HCM and a cardiac mass, where a mass-like lesion shows differential enhancement with the adjacent myocardium while mass-like HCM is consistent with the homogeneous signal intensity of the surrounding normal LV myocardium.^{6,7}

End-stage HCM or burned-out phase HCM is the type with progressive thinning of the LV wall, increased LV end-systolic dimensions, and reduced left ventricular ejection fraction.⁷ This subtype is considered to have a poor prognosis with associated systolic dysfunction and extensive myocardial fibrosis which can be reliably evaluated with CMR.^{3,7}

Management and Prognosis

Management strategies of HCM differ among individuals depending upon their presenting stage, etiology, and obstructive or non-obstructive physiology. Hence, treatment varies from without the need for extensive medical interventions to prophylactically implantable cardioverter/ defibrillators (ICD) to prevent sudden death. Surgical septal myectomy, or alcohol septal ablation (ASA) to relieve LVOT obstruction, use of anticoagulative and antiarrhythmics to prevent thromboembolic events and atrial fibrillation are other treatment options to be considered in selected patients.^{7,9} In addition, HCM being relevant to the incidence of sudden cardiac death, it is essential to be familiar with the related prognostic indicators. The major parameters that determine the prognosis of HCM comprise of LV maximal wall thickness, presence/absence of LVOT obstruction, the fibrotic burden, right ventricular hypertrophy, and diastolic cavity obliteration (Table 1).¹⁰ Our current instance here had poor prognostic parameters of maximal LV wall thickness of 44.1 mm and the presence of LVOT obstruction, while the fibrotic burden was only 4 %. Amidst all the variability and challenges, CMR is indeed an established modality to evaluate the prognosis in terms of disease progression and risk of sudden death as well as to determine an approach to clinical management. Our patient was advised for ICD placement based on the prognostic evaluation.

Table 1. Poor Prognostic Parameters of Hypertrophic Cardiomyopathy

Maximal wall thickness	>30mm indicative of poor prognosis
Fibrotic burden	>15% indicative of poor prognosis
LVOT obstruction	Present- poor prognosis
RV involvement	Present- poor prognosis
Diastolic cavity obliteration	Present- poor prognosis

Conclusion

A heterogeneous clinical picture of HCM continues to challenge clinicians to diagnose it based on ECHO findings which is considered the initial imaging modality in the evaluation of hypertrophic cardiomyopathy. However, several advantages of CMR over ECHO such as accurate measurement of wall thickness, detailed interpretation of cardiac structures in different views, and the capability to identify myocardial fibrosis with late gadolinium enhancement images, make it a preferred radiological modality to diagnose and evaluate the vulnerability of HCM.

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