

## Case report

# An Uncommon Variant of an Uncommon Disease: A Caucasian Adolescent with Apical Hypertrophic Cardiomyopathy Diagnosed with Myocardial Perfusion Imaging

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## Abstract

Apical hypertrophic cardiomyopathy (ApHCM) is a subtype of HCM. This variant is more common in the Asian population when compared to North American patients. Patients may present with arrhythmias, heart failure, myocardial infarction, chest discomfort, fatigue, and presyncope or syncope. Initial evaluation requires electrocardiogram and two-dimensional echocardiogram. T-wave inversion in the precordial leads as well as hypertrophy of the left ventricle is hallmarks of the disease. Cardiac magnetic resonance (CMR) imaging is the most specific and sensitive imaging modality. In patients with contraindications for CMR, myocardial perfusion imaging (MPI) has been described to have diagnostic characteristics for ApHCM. MPI images demonstrating a “solar polar” map pattern and increased apical tracer uptake in single-photon emission computed tomography horizontally and vertical long-axis slices are consistent with the diagnosis of ApHCM. Herein, we present a case of a Caucasian adolescent female who underwent a cardiac screening to rule out hypertrophic obstructive cardiomyopathy. Initially, the patient was unable to undergo CMR, and an MPI was utilized to assist with the diagnosis of ApHCM.

**Keywords:** Apical, cardiomyopathy, hypertrophic, myocardial perfusion imaging, myocardial perfusion, solar polar map

## Introduction

Apical hypertrophic cardiomyopathy (ApHCM) is a subtype of HCM, which is characterized by asymmetric myocardial hypertrophy of the apex of the left ventricle. These patients do not have a classical left ventricular (LV) outlet obstruction but may have mid-ventricular obstruction that can manifest similar symptoms to that of hypertrophic obstructive cardiomyopathy (HOCM).<sup>[1]</sup> Apical HCM develops more common in the

Asian population.<sup>[2,3]</sup> In this case, we will review the clinical characteristics of ApHCM and correlate it with different radiographic modalities specifically myocardial perfusion imaging (MPI).

## Case Report

An asymptomatic 15-year-old Caucasian female with no significant medical history underwent a yearly physical examination before participation in sports. She has a history of orthopedic surgery with an unknown metal implant. The patient denies any family history for sudden cardiac death, seizure activity, or syncopal

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episodes. The patient denies any syncope, loss of consciousness, palpitations, or chest discomfort. Physical examination findings were within normal limits with no extra heart sounds, murmurs, rubs, or gallops. The patient electrocardiography demonstrated normal sinus rhythm with LV hypertrophy and T-wave inversion in the inferolateral leads [Figure 1]. The patient was referred for a two-dimensional (2D) echocardiogram (ECHO) [Figure 2], which revealed hypokinesia of the anterior apical segment. The right ventricular apex appeared either foreshortened or hypoplastic. There was no evidence of aortic stenosis. Due to the patients' orthopedic implant, she underwent exercise stress Tc-99m sestamibi MPI. The MPI demonstrated apical thickening with a focal hotspot on the rest images [Figure 3a-c]. Gated image also revealed apical bulging and tardokinesis with a normal ejection fraction of 67%. It was later confirmed the patient did not have any metal implant and underwent cardiac magnetic resonance (CMR) [Figure 4]. This patient was diagnosed with ApHCM based on the electrocardiogram (ECG), 2D ECHO, MPI imaging, and later CMR.

## Discussion

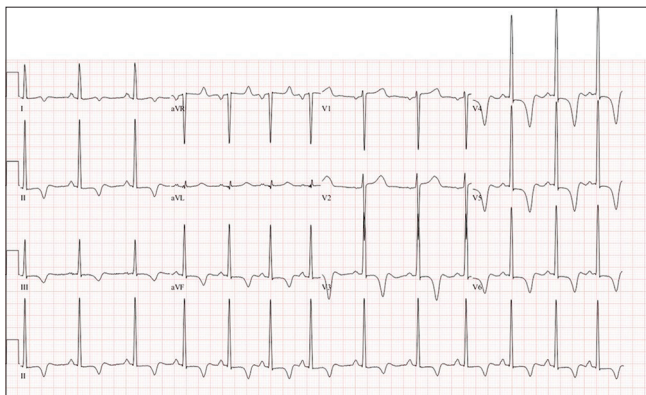
ApHCM is an uncommon subtype of HCM, which is characterized by asymmetric myocardial hypertrophy of the apex of the left ventricle. ApHCM is a more common variant that occurs in Asians than non-Asians, with the prevalence reported to be as high as 41% of the HCM patients in China<sup>[2]</sup> and in 15% of HCM patients in Japan.<sup>[3]</sup> Interestingly enough, in non-Asian populations, reports suggest a prevalence between 1% and 3% of HCM patients.<sup>[3-5]</sup> The pathophysiology is thought to be associated with sarcomere gene mutations.<sup>[6]</sup> However, a direct genetic link has not been established. Patients may present asymptomatic during screenings like in our patient versus symptomatic. Symptoms may include

arrhythmias such as atrial fibrillation, ventricular fibrillation, fatigue, shortness of breath, and heart failure. Patients may also present with presyncope or syncope.<sup>[1,2,4,7]</sup>

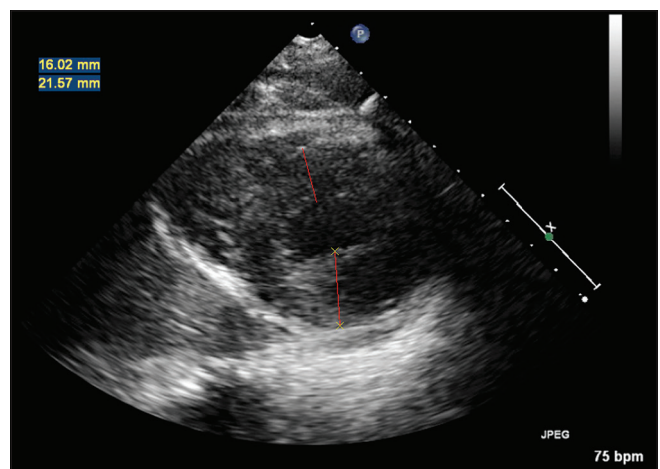
ApHCM has a better mortality prognosis compared to HOCM and other variants. In a study done by Eriksson *et al.* in 2002, a group of 105 patients with ApHCM were followed for 13.6 years. The total cardiovascular mortality rate was 1.9%. These patients had atrial fibrillation as the most common complication. Following the completion of that study, ApHCM in the North American patients did not have an association with sudden cardiac death.<sup>[7]</sup>

Work-up of ApHCM begins with obtaining an ECG. Our patient presented with the classical ECG pattern of giant negative T-waves in the precordial leads (V2-V6).<sup>[2]</sup> A study conducted by Cianciulli *et al.* demonstrated 20/20 patients with confirmed ApHCM as having giant T-wave inversion in the precordial leads. The diagnosis can be confirmed with imaging modalities, specifically, 2D ECHO, computed tomography, or CMR.<sup>[8,9]</sup> However, in our case, the ability to initially utilize CMR was contraindicated with a suspicion for an orthopedic implant.

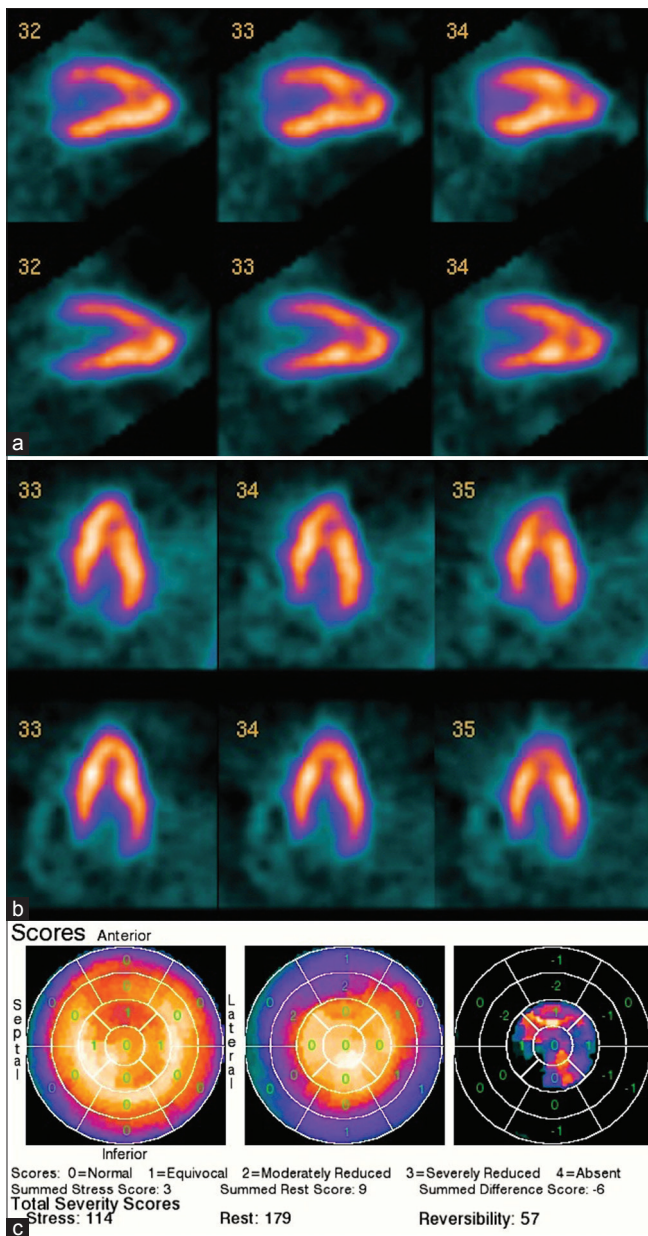
The indication for MPI to characterize HCM is not well described. ECHO and CMR have both been described in the literature to be the most sensitive and specific imaging modalities. Nevertheless, the use of MPI in cases where CMR is contraindicated may be a helpful tool in the diagnosis of ApHCM. Previous studies of stress MPI in patients with ApHCM have demonstrated normal to reversible apical perfusion defects along with normal coronary arteries.<sup>[8]</sup> Our patient demonstrated characteristic ECG abnormalities in association with "solar polar" map pattern and increased apical



**Figure 1:** The electrocardiogram demonstrates normal sinus rhythm with left ventricular hypertrophy. There is a marked ST abnormality with T-wave inversion in leads II, III, arteriovenous fistula, and V3-V6. The T-wave inversion in our patient follows a characteristic pattern of inferolateral pathology

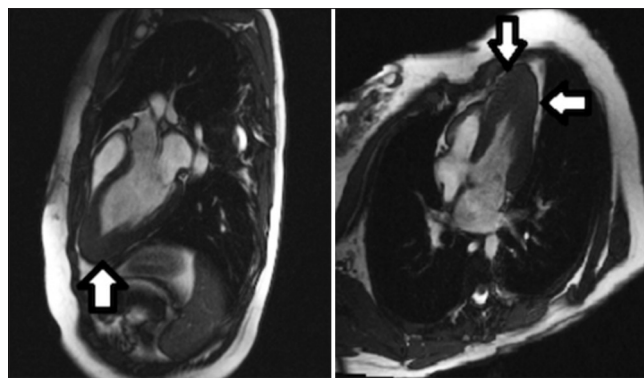


**Figure 2:** The two-dimensional echocardiogram displayed above demonstrates apical hypertrophy with apical obliteration with systole. The overall global left ventricular function is normal. There is normal right ventricular chamber size and wall thickness



**Figure 3:** The single-photon emission computed tomography images: (a) Septal-lateral. Rest (top images) and stress imaging (bottom images) demonstrate unusual morphology with the apex appearing prominent. The perfusion at the apex appears decreased on stress compared to rest; however, this is likely an artifact. (b) Inferior-superior. Rest (top images) and stress imaging (bottom images) demonstrate unusual morphology with the apex appearing prominent. The perfusion at the apex appears decreased on stress compared to rest; however, this is likely an artifact. (c) 17-segment bullseye model. Rest (middle) and stress imaging (left) demonstrate the “solar polar map” and is characteristic of apical hypertrophy

tracer uptake in single-photon emission computed tomography horizontally and vertical long-axis slices that are consistent with the diagnosis of ApHCM.<sup>[8,10]</sup> The apical hypertrophy produces a focal hotspot, with apparent decreased intensity in the mid and basal segments, which is accentuated on the resting images due to loss of resolution from a decreased count rate.



**Figure 4:** Cardiac magnetic resonance images in the sagittal view showing end diastole (left side) and short-axis view in end systole (right side). The apical myocardial thickening extends approximately 3 cm in length and is between 3.5 and 4 cm in thickness

On the stress images, this effect is minimized with a three-fold higher count rate which results in improved resolution.

The polar map of the resting images [Figure 3c] also reflects this perfusion artifact, producing apparent reversibility. This polar plot appearance is colloquially known as a “solar polar map” and is characteristic of apical hypertrophy. The MPI appearance in concordance with ECG findings is of diagnostic value regarding the evaluation of ApHCM. Using MPI as a modality can be invaluable when a patient cannot undergo CMR.

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Nil.

### Conflicts of interest

There are no conflicts of interest.

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