Apical Hypertrophic Cardiomyopathy in an African American: A Case Presentation and Literature Review

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Abstract

We present a case of a patient with a two-month history of night sweats, nausea and vomiting, fatigue and shortness of breath on exertion as well as significant weight loss. Prior to arrival at our facility he was diagnosed with apical aneurism and was offered an evaluation with cardiac catheterization but the patient decided on a second opinion and thus came to our facility. The apical aneurism turned out to be apical variant hypertrophic cardiomyopathy, which is a common echocardiographic misinterpretation and thus necessitating additional imaging modalities such as cardiac MRI.

We perform a comprehensive and up to date literature review of apical variant hypertrophic cardiomyopathy as it relates to epidemiology, diagnosis, prognosis and treatment as some variants portend poorer prognosis and it is often misdiagnosed on initial echocardiography as described in our case presentation.

INTRODUCTION

Apical Hypertrophic Cardiomyopathy is a rarely occurring variant of hypertrophic cardiomyopathy in western population with variable presentation although some classic diagnostic findings. Certain variants may portend poorer prognosis and thus proper diagnosis with prompt evaluation is important. We review the different variants of this disease entity and examine approach to diagnosis and prognosis as it is often misdiagnosed on echocardiography.

CASE DESCRIPTION

A 50-year-old African American male with a past medical history significant for hypertension and dyslipidemia presented with a two-month history of night sweats, nausea with intermittent vomiting, fatigue, and shortness of breath on exertion.

A chest roentgenogram demonstrated bilateral hilar lymphadenopathy, confirmed on chest Computed Tomography (CT). Electrocardiogram demonstrated left ventricular hypertrophy with associated ST changes. Given these latter findings, an echocardiogram was performed and despite normal global left ventricular systolic function, a left ventricular apical aneurysm was suggested. The patient refused recommended coronary angiogram and instead came to us for a second opinion.

The initial electrocardiogram (ECG) performed at the time of admission was unchanged when compared to the outside hospital’s tracing (Figure 1). Troponin I was elevated initially at 0.5 ng/mL and increased up to 0.95 ng/mL within the next two days before stabilizing to a mean of 0.91 ng/mL. Repeat echocardiogram with doppler revealed apical hypertrophy with a small, focal, akinetic region, but normal left ventricular ejection fraction (Figure 2).

Subsequently the patient underwent a cardiac MRI due to his abnormal Echocardiogram to evaluate for scar burden and for possible concomitant cardiac sarcoid. It revealed diffuse late gadolinium enhancement of the left ventricular apical segment in a pattern most suggestive of myocardial fibrosis, but not classic for cardiac sarcoidosis (Figure 3A,3B). The overall left ventricular size and function was normal. The patient was later discharged home with a diagnosis of apical variant hypertrophic cardiomyopathy and pulmonary sarcoidosis placed on beta-blockers and directed to follow-up closely with cardiology.

DISCUSSION

Apical hypertrophic cardiomyopathy (AHCM) is a rare form...
of non-obstructive hypertrophic cardiomyopathy (HCM) first described in Japanese patients by Sakamoto, et al in 1976 [1]. This group performed left ventricular scanning by echocardiography and ultrasono-cardiotomography to search for possible muscular abnormalities in nine cases that demonstrated giant T wave inversion (>1.2mV) in the precordial leads on an ECG without known documented cause. The group was then compared to nine control cases with documented hypertrophic cardiomyopathy with obstruction. The group found that the degree of T-wave inversion differed between the two groups, and that those with the apical variant had much larger degree of inversion. In 1983, Yamaguchi, et al subsequently published a report determining that apical hypertrophy is the only specific hypertrophic pattern that shows characteristic ECG abnormalities i.e. giant negative T waves and high QRS voltage in the left precordial leads [2].

Currently it is estimated that AHCM has an incidence of approximately 25% of Japanese [2] and Taiwanese populations [3] as well as constituting 16% of all HCM Chinese patients as documented in a study by Yan, et al [4]. Whereas the incidence in western world does not exceed 3% of the entire HCM population [5]. It is believed that this number is underestimated in the US given autopsy as well as cardiac magnetic resonance imaging.

**Figure 1** Electrocardiogram depicting normal sinus rhythm at 79 bpm with left ventricular hypertrophy, prolonged QTc, and deep inverted T-waves in the infero-lateral leads.

**Figure 2** A four chamber echocardiographic view depicting the classic end-diastolic “spade-like” cavity typically associated with apical hypertrophic cardiomyopathy.
(MRI) data, suggesting that echocardiography often misses this diagnosis [6]. Male gender is the most frequently sex affected in the Japanese population but this gender difference does not translate to populations outside Japan [7].

The most commonly found presenting symptoms associated with AHCM are chest pain, palpitations, and/or dizziness. Diagnostic evaluation may be initiated by an incidentally found abnormal ECG where the most frequent findings are negative T-waves in the precordial leads found in 93% of patients (notably a depth > 10 mm are found in 47%). Sixty five percent of patients also have documented left ventricular hypertrophy on imaging [8].

The diagnostic criteria for AHCM include demonstration of asymmetric left ventricular hypertrophy confined predominantly to the LV apex. This hypertrophy must present with an apical wall thickness ≥15 mm and a ratio of maximal apical to posterior wall thickness ≥1.3. The diagnosis can be based on either echocardiogram, which can utilize contrast medium such as Optison™ microspheres to improve endocardial border visualization or cardiovascular resonance imaging (CMR). AHCM is generally divided into two groups, based on whether they had isolated asymmetric apical hypertrophy (pure AHCM) or had co-existent hypertrophy of the inter-ventricular septum (mixed AHCM) [7]. Phenotypic expression of the two main types of AHCM have been linked to geographic as well as racial factors. The "pure" form of AHCM (hypertrophy of only the apical segments) is predominant in Japanese patients, while the mixed form has been linked to Caucasian patients [9,10].

The preferred initial imaging test is a transthoracic echocardiogram (TTE). Image quality can vary however, as it is dependent on many factors including body habitus and the skill of the sonographer to focus on the left ventricular apex. When the baseline images are suboptimal, a contrast echocardiogram may be utilized depicting a “spade-like” configuration of the apical segment (similar to what is seen with left ventricular angiography) [11]. Other imaging modalities have been proven useful such as single photon emission computed tomography (SPECT) myocardial perfusion imaging. This modality usually presents with three distinct findings; 1) an increased apical tracer uptake, 2) a spade-like configuration of the LV chamber, and 3) the “Solar Polar” map pattern. This presentation may be seen in 75% of patients [12]. The “Solar Polar” map pattern on resting Tl-201 volume-weighted polar maps described by Ward, et al. is the first description of the typical findings on dual-isotope rest and stress SPECT perfusion images [13]. This pattern demonstrates an intensely bright spot of counts in the apical segment surrounded by a circumferential ring of decreasing counts. Cardiac computed tomography (CCT) can also be used to diagnose AHCM and simultaneously evaluate the coronary artery anatomy [14]. CMR is a valuable tool for diagnosing this entity and it tends to be the modality of choice when echocardiography produces suboptimal images [15]. Furthermore, CMR can accurately measure the myocardial wall thickness, the left ventricular mass, the left atrial volume, and also search for apical aneurysms and myocardial scarring/fibrosis.

Apical hypertrophic cardiomyopathy is generally associated with a good prognosis. This has been demonstrated repeatedly both in Asian and Caucasian populations. The long-term annual cardiovascular mortality in both populations is 0.1 percent. In Asian populations elderly women tend to have adverse outcomes though in western populations younger patients are at higher longer-term risk [8]. The extent of scar burden evaluated with late gadolinium enhancement as well as the presence of an apical aneurysm identified by cardiac MRI were both found to be significant predictors of major adverse clinical events in apical hypertrophic cardiomyopathy patients with a signal towards increased ventricular arrhythmias [16,17].

The approach to management of apical hypertrophic cardiomyopathy depends on symptoms and the risk of sudden cardiac death. The medical regimen in symptomatic patients primarily consists of beta-blockers, which have been shown
to decrease symptoms as well as overall morality [18-20]. The concern associated with overall diagnosis of hypertrophic cardiomyopathy is the risk of sudden cardiac death. Current guidelines do not comment on indications for primary prevention for SCD with ICD implantation specific for apical variants of hypertrophic cardiomyopathy. Most recent ACCF/AHA as well as ESC Guidelines on diagnosis and treatment/management of hypertrophic cardiomyopathy describe the major clinical features associated with increased risk of sudden cardiac death as described for hypertrophic cardiomyopathy in general include Non-sustained ventricular tachycardia, maximum left ventricular wall thickness of ≥ 3cm, family history of sudden cardiac death at young age, unexplained syncope and abnormal blood pressure response during exercise. Other potential SCD risk modifiers are increased left atrial diameter as well as already described increased scar burden as evidenced by LGE on CMR and apical aneurism [21,22]. Certainly any of these clinical features would not be dismissed in a patient with apical variant hypertrophic cardiomyopathy and thus would be used in clinical prognosis.

Apical variant of hypertrophic cardiomyopathy is less prevalent in the Western hemisphere. This variant generally carries a good prognosis, but there are several factors associated with higher risk for morbidity and mortality. Careful evaluation of the patient with regards to personal as well as family history, and a multimodality imaging approach for risk stratification should be employed.

For our patient initial echocardiographic evaluation was sufficient to diagnose the patient with apical thickening. This case was unique due to the patient’s ancestry and further complicated by the underlying presence of sarcoidosis. Here, CMR was obtained not only to further evaluate the extent of apical thickening, but to also determine between the presence of a fibrotic versus infiltrative etiology. LGE is a useful technique as there are specific patterns associated with cardiac sarcoid such as subepicardial fibrosis mostly affecting basal septal and basal lateral walls [23]. Our patient’s CMR showed only fibrosis such as subepicardial fibrosis which was prevalent in the distribution of hypertrophy and otherwise did not exhibit basal lateral walls [23]. As there are specific patterns associated with cardiac sarcoidosis, LGE is a useful technique to decrease symptoms as well as overall mortality [18-20].

REFERENCES