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Apical Hypertrophic Cardiomyopathy Presenting as Left Ventricular Aneurysm in a 74-Year-Old African American Male

Daniel E Diez[1*], Joel Brooks Tamayo[1], Jennifer Rodriguez-Belen[1], Jorge Hernandez Canciobello[1], Amr El-Talla[2], Kevin Mancheno[2], Jorge X Montilla[1], James D Davenport[1] and Yeissen Godinez[1]

[1]HCA Florida Kendall Hospital, SW 40th Street, Miami, Florida

[2]Nova Southeastern University Dr. Kiran C Patel, College of Allopathic Medicine, S. University Drive, Florida

[*]**Corresponding author:** Daniel E Diez, 1HCA Florida Kendall Hospital, SW 40th Street, Miami, Florida, United States.

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Abstract

A 74-year-old African American male with multiple comorbidities including NSTEMI was evaluated for epigastric pain and found to have the rare apical variant of hypertrophic cardiomyopathy, referred to as Yamaguchi Disease. This variant spares the interventricular septum and patients largely lack the symptoms of traditional cardiomyopathy. This case was of particular significance not only due to the rare presentation amongst a population outside of its usual Asian demographic, but also one with an uncharacteristically more aggressive appearance, meriting a thorough workup and interventional treatment when taking into account the patient's history of acute coronary syndrome and ventricular arrhythmia.

Background

Apical hypertrophic cardiomyopathy (apHCM) is a variant of hypertrophic cardiomyopathy (HCM) involving enlargement of the apical myocardium. This variant typically lacks subaortic left ventricular outflow obstruction. ApHCM is commonly seen in East Asian populations, particularly Chinese and Japanese. Most reports suggest that in non-Asian populations, this variant makes up only 1-3% of patients with HCM [1]. This disease has been shown to arise due to mutations in sarcomeric protein genes that follow an autosomal dominant pattern but can also arise sporadically. Symptomatic patients experience angina, heart failure, myocardial infarction, and ventricular or atrial fibrillation. Although most symptomatic cases follow a mild course, a minority of patients will develop refractory symptoms of angina, dyspnea, and presyncope or syncope due to diastolic ventricular dysfunction and reduced cardiac output.

Common features of apHCM include a fourth heart sound, large negative T waves on EKG mostly in the left precordial leads, a "spade-like" image on echocardiogram with associated apical obliteration, and ventricular wall abnormalities. Diagnostic modalities include echocardiography, computerized tomography, ventriculography, and cardiac magnetic resonance imaging.

Although the American Heart Association lacks diagnostic criteria for apHCM, it is confirmed with imaging displaying apical wall thickness ≥15 mm with a ratio of maximal apical to posterior wall thickness ≥1.5 [2]. Management of apHCM is generally the same as that of traditional HCM. However, given that most patients with apHCM do not consistently display left ventricular outlet or mid-cavity obstruction, septal reduction is typically not indicated [1].

Case Report

A 74-year-old African American male with hypertension, hyperlipidemia, diabetes mellitus, ventricular tachycardia, and NSTEMI status-post left heart catheterization (approximately 8 months prior to presentation and showed no evidence of CAD) presented to the emergency department with midline, non-radiating postprandial epigastric pain. The pain was rated 10/10 in severity prior to the patient being administered morphine which improved to 5/10. of note, the patient was scheduled to have an AICD placed during his most recent hospitalization but left against medical advice. A CT abdomen/pelvis showed prostatomegaly, cholelithiasis, and what was interpreted as a 3.4cm left ventricular apical aneurysm (Figure 1). Electrocardiogram was done and showed no significant ST changes or signs of ischemia. However, large T wave inversions were noted in the precordial leads (Figure 2). Subsequent echocardiography was negative for LV aneurysm, systolic dysfunction, and ejection fraction was preserved. The patient's thickened apex took on the ace-shape seen in Yamaguchi Syndrome, further raising suspicion that this patient had apHCM (Figure 3a-3d). Additional studies identified a systolic anterior movement of the mitral valve leaflet, characteristic of HCM, present in our patient (Figure 4).

The patient had been previously seen by cardiology for monomorphic ventricular tachycardia (VT) and at the time refused AICD placement. At his present admission, cardiology repeated concerns for AICD placement by electrophysiology given the patient's significant history of NSTEMI, monomorphic VT, and current evidence of apHCM, raising the risk for future, potentially fatal, cardiovascular events. Electrophysiology later successfully placed a dual chamber AICD with instructions for close follow-up outpatient. In addition to electrophysiology follow-up, the patient was informed of the necessity for himself and, his children especially, to receive genetic screening tests. At the time, the patient agreed but was lost to follow-up.

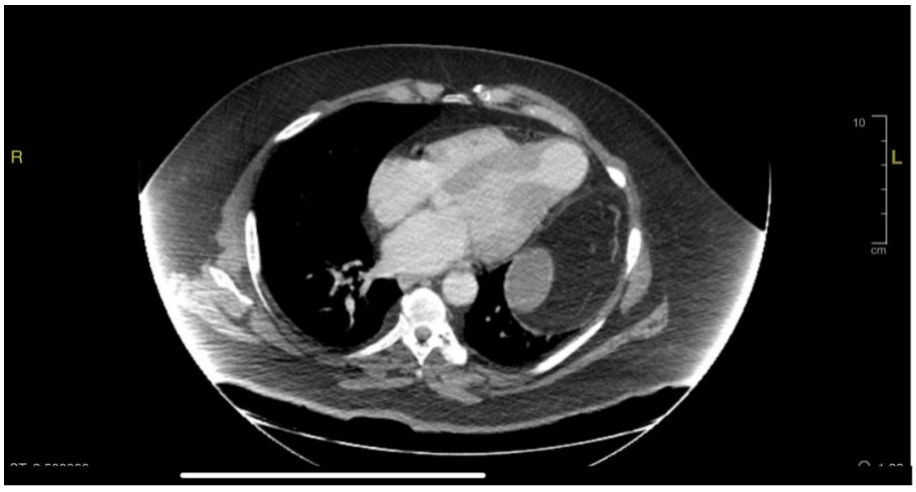

Figure 1: CT abdomen identifying what was believed to be a left ventricular aneurysm.

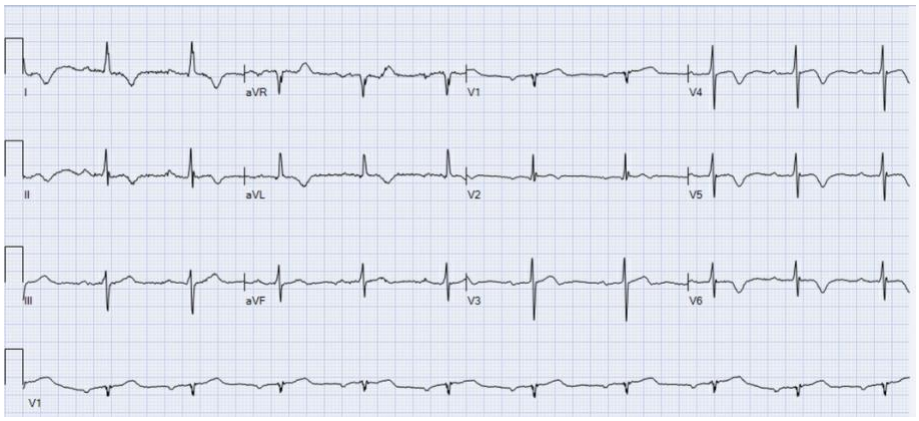

Figure 2: EKG displaying large negative T waves in the precordial leads.

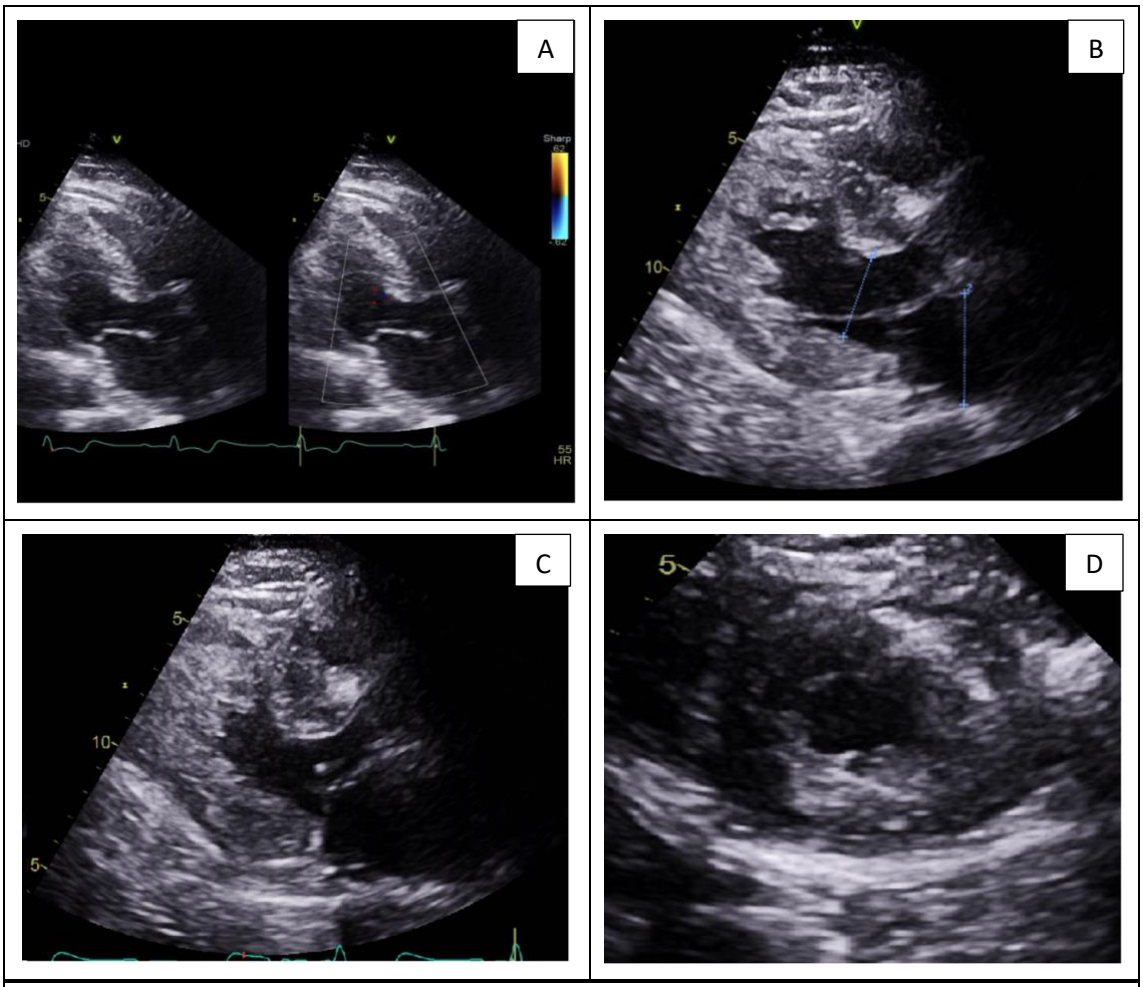

Figure 3A: No evidence of a left ventricular aneurysm and a left ventricle spade-like appearance; **(3B and 3C):** Apical thickening parasternal long axis view; **(3D):** Apical thickening in parasternal short axis view.

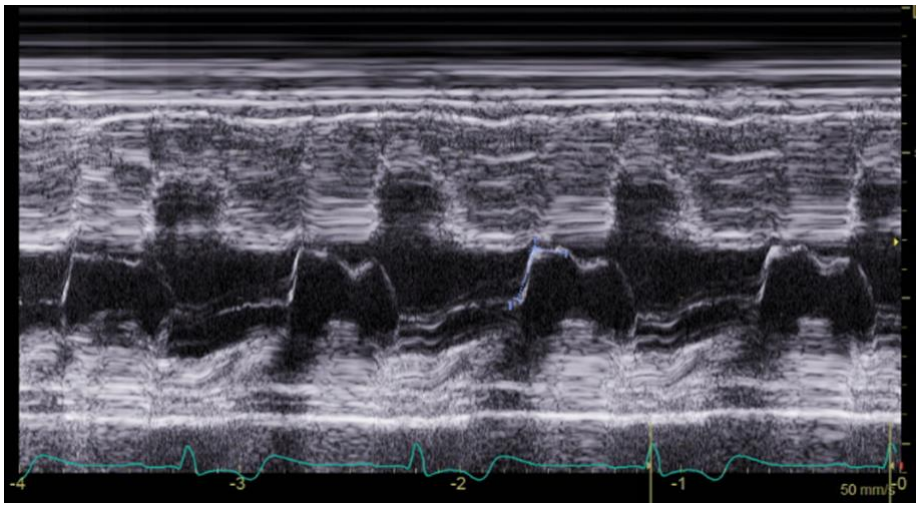

Figure 4: Echocardiography identified systolic anterior motion of the mitral valve.

Discussion

Apical hypertrophic cardiomyopathy (apHCM), or Yamaguchi Disease, is a variant of hypertrophic cardiomyopathy (HCM) that involves pronounced thickening of the apical myocardium of the left ventricle. It has been shown to follow an autosomal dominant inheritance pattern, but also arises secondary to spontaneous mutation. It is often seen in patients of East Asian descent. Given that this variant of HCM is so uncommon outside of this population, this case is rather unique. Cases of apHCM in African Americans have been scarcely documented. This patient's symptoms and demographic presented challenges in regards to coming to the diagnosis of apical hypertrophic cardiomyopathy; a disease seldom considered in patients during cardiac workup. In our case, initial diagnostic measures led us to believe the patient had a left ventricular aneurysm, especially considering the significant cardiac history in this patient. However, enhanced imaging would reveal he had apical hypertrophy, loosely presuming the spade-shape, pointing toward Yamaguchi's, and steering treatment in a different direction. Due to the uniqueness of apical involvement and varying severity, it was important to rule out apical obliteration. The presence of obliteration could be used to predict the risk of significant complications [7->3].

Decision of diagnosis modality presents its own challenges, especially when reviewing which to choose. ApHCM can be more readily detected on cardiac magnetic resonance (CMR) and allows for earlier stages to be seen. When scrutinized, 40% of cases were missed on echocardiography, only to be discovered by CMR. MR studies are more sensitive and are the preferred test of choice, able to identify 25-43% of the apHCM cases that a routine echocardiography had missed [8]. A screening echocardiography is still suggestive and able to find more prominent cases, and it is up to the inclination of the clinician to decide whether a CMR study would be more helpful. To further emphasize this point, CMR provides a unique opportunity when compared to echocardiography. Gadolinium enhancement allows for assessment and quantification of hypertrophy, fibrosis and consequently, ischemia, indicated by a lesser degree of uptake and enhancement of contrast. This decreased uptake is possibly associated with increased risk of mortality with regards to sudden cardiac death or complications such as heart failure [9]. In the case of traditional HCM, there is a considerable risk for events such as sudden cardiac death from ventricular fibrillation, follow-up is recommended on a strict basis, to assess the need for intervention such as AICD placement [5->4]. Patients with traditional HCM have a higher likelihood of developing diastolic dysfunction with an earlier onset of heart failure, most of these patients having a left ventricular outflow obstruction.

A case series of HCM prognosis found that mortality was 0.7% per year for HCM related cause [4->5]. When compared to apHCM, the risks are different, and decreased. A prospective study looking into the long-term outcome and complication development of patients with apHCM found that 15-year survival was 95% with annual cardiovascular mortality recorded at 0.1% [3->6]. Despite apical hypertrophic cardiomyopathy being seen as a more benign disease with a largely unremarkable course, serious cardiac complications are identified in about one-third of cases, with patients experiencing dangerous arrhythmias, atrial fibrillation being the most common, myocardial infarctions, and an increased risk of stroke [3->6]. Although these complications have been recorded in apHCM, they still occur at a decreased rate. In contrast, apical aneurysms have been identified at an increased frequency, about 13-15%, whereas it is only identified in 2% of patients with traditional HCM [1].

Further challenge in diagnostics is observed when noting that apHCM patients less than often describe a family history of similar conditions. A lack of consistent family history, as compared to classical HCM, has led to difficulty in establishing screening modalities and also identifying a strong etiological factor [6->7]. There have been genetic implications in apHCM where a possible autosomal dominant pattern has been identified. In familial cases, a missense mutation in sarcomeric proteins has been identified which can further add to the importance of genetic screening in individuals with specifically apHCM [10]. This case highlights the importance of identification and proper management of apHCM, with close cardiology follow-up for medical management and if comorbidities are present, especially a cardiac history, the necessity for interventional measures to prevent more severe, life-threatening cardiac events is further recommended.

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