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CASE REPORT

Apical Hypertrophic Cardiomyopathy With Aneurysm Formation: Potential For Underdiagnosis And Value Of Contrast Echocardiography

Vinayak Vadgaonkar[1], Suddharsan Subbramaniyam[2], Nooraldaem Yousif[3], Sadananda Shivappa[4], Fuad Abdulqader[5], Leena Sulaibikh[6*]

[1]Chief resident, Department of Cardiology, Mohammed Bin Khalifa Cardiac Centre (MKCC) Riffa, Kingdom of Bahrain.

[2]Senior resident, Department of Cardiology, Mohammed Bin Khalifa Cardiac Centre (MKCC), Riffa, Kingdom of Bahrain.

[3]Consultant, Department of Cardiology, Mohammed Bin Khalifa Cardiac Centre (MKCC)Riffa, Kingdom of Bahrain.

[4]Consultant, Department of Cardiology, Mohammed Bin Khalifa Cardiac Centre (MKCC)Riffa, Kingdom of Bahrain.

[5]Consultant, Department of Cardiology, Mohammed Bin Khalifa Cardiac Centre (MKCC)Riffa, Kingdom of Bahrain.

[6]Consultant, Department of Cardiology, Mohammed Bin Khalifa Cardiac Centre (MKCC)Riffa, Kingdom of Bahrain.

***Corresponding author:**
Dr. Leena Sulaibikh FRCP, Consultant Cardiologist, Head of Non-invasive Cardiology Department, Mohammad Bin Khalifa Cardiac Centre, Bahrain. Email: drsulaibikh@gmail.com

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Abstract

Apical hypertrophic cardiomyopathy (Apical HCM) with apical aneurysm formation is a relatively rare variant of hypertrophic cardiomyopathy. In this case report, the authors share their experience in the diagnosis of this challenging case, in view of the rarity of such peculiar clinical condition accompanied with a variable presentation and clinical course that render the diagnosis of this high-risk HCM phenotype commonly delayed or missed.

Keywords: Cardiomyopathy, Contrast Media, Echocardiography, Heart Aneurysm, Hypertrophic

Introduction

Hypertrophic cardiomyopathy (HCM) is a spectrum of heterogeneous heart muscle disease that is characterized by left ventricular hypertrophy (LVH), which is unexplained by cardiac afterload condition.[1]The disease is commonly inherited in autosomal dominant fashion secondary to mutation in sarcomeric protein genes with several patterns of LVH being described, namely: asymmetric septal, concentric, reverse septal, neutral, apical and some other rare variants.[2]

Apical HCM was first described in Japan in 1976 as deep negative precordial T-waves on surface electrocardiogram (ECG) associated with spade-

like shaped contour of LV cavity at end diastole.[3]

Here, the authors present a case of apical hypertrophic cardiomyopathy and LV apical aneurysm, referred to the cardiac Centre as acute coronary syndrome.

Case Presentation

A 67-year-old gentleman, with no known risk factors, was admitted to a general hospital with a two-hour history of retrosternal chest pain. The patient denied alcohol consumption, smoking and illicit drug use. At admission ECG showed deep T wave inversion in anterolateral leads (Figure 1). Two sets of high sensitivity troponins were negative and 2D echocardiogram reported as normal. The patient was treated as unstable angina and subsequently transferred to the Centre for coronary angiogram, which revealed normal epicardial coronaries.

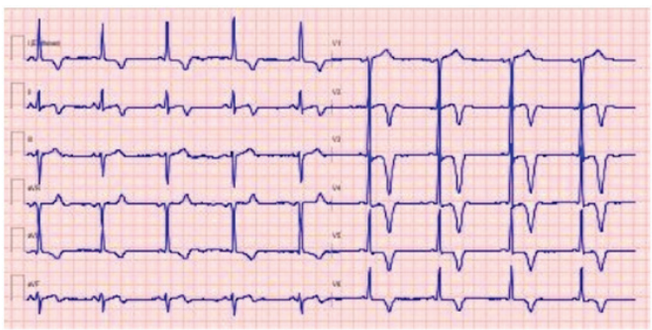

Figure 1: ECG showing widespread T wave inversion in the precordial leads with Left ventricular Hypertrophy by Voltage criteria

A comprehensive non-contrast transthoracic echocardiogram (TTE) was performed in accordance with standard chamber quantification guidelines, showed normal biventricular size and systolic function (Biplane left ventricular (LV) ejection fraction 65%). Left atrium appeared dilated with estimated indexed left atrial volume of 36 ml/m^2. Moderate diastolic dysfunction with high left ventricular filling pressures was noted. Apical views were strongly suspicious of significantly increased wall thickness in apical LV segments confirmed with turbulent flow on color Doppler in apex (Figure 2). Continuous wave doppler signal at rest within apical LV cavity at the point of distal flow obstruction demonstrated mid to late systolic peaking jet followed by void before the second peak of paradoxical early diastolic forward flow and late diastolic run off with peak intraventricular gradients of 33 mm Hg and 31 mm Hg (Figure 3).

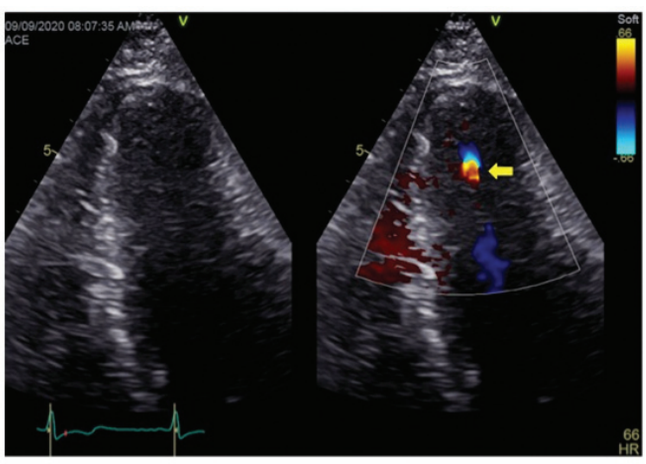

Figure 2: Apical 4 chamber view showing distal flow acceleration in systole (yellow arrow)

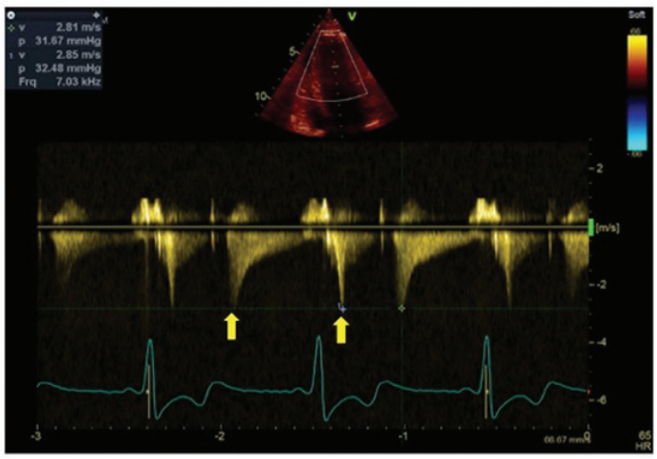

Figure 3: Double peaked CWD signal (yellow arrows) showing paradoxical early diastolic flow suggestive of concealed apical asynergy

In light of significant apical hypertrophy, typical ECG findings and paradoxical diastolic forward flow, which could be, a sign of concealed asynergic apex; it was decided to perform transpulmonary

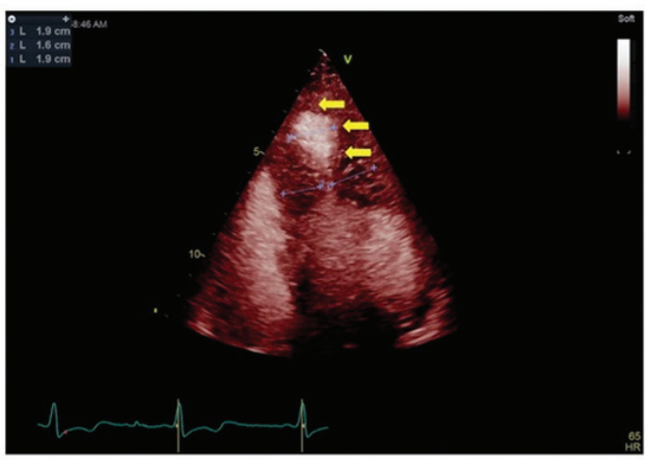

Figure 4: Contrast TTE apical view shows hourglass shaped LV with distal apical aneurysm (yellow arrows)

ultrasonic enhancing echocardiography, also commonly known as contrast TTE. Optison (GE) with intermediate mechanical index of 0.25 was performed. Contrast TTE clearly showed hour-glass shaped configuration of LV in diastole with increased wall thickness below the level of papillary muscles. Maximal wall thickness was noted in apical lateral segment of 19mm. Additionally, thin, discreet akinetic distal apical segment with maximal transverse diameter of 19mm and narrow neck separating proximal and distal apex was observed (Figure 4). Contrast perfused septal myocardium adequately and interestingly linear intramyocardial vessels running from epicardium into LV cavity was noted (figure 5). Speckle tracking echocardiography

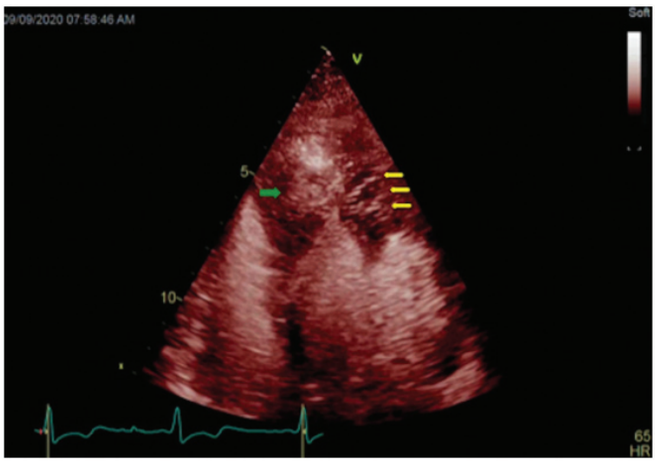

Figure 5: Contrast TTE apical view shows normal perfusion of myocardium (green arrow) along with distinct intramyocardial linear vessels (yellow arrows)

(STE) demonstrated significantly reduced average global longitudinal peak systolic strain (GLS) of -12.3% with systolic lengthening of apical segments reflecting akinetic/dyskinetic apex. Peak strain dispersion (PSD) was noted to be very high at 212 milliseconds (figure 6).

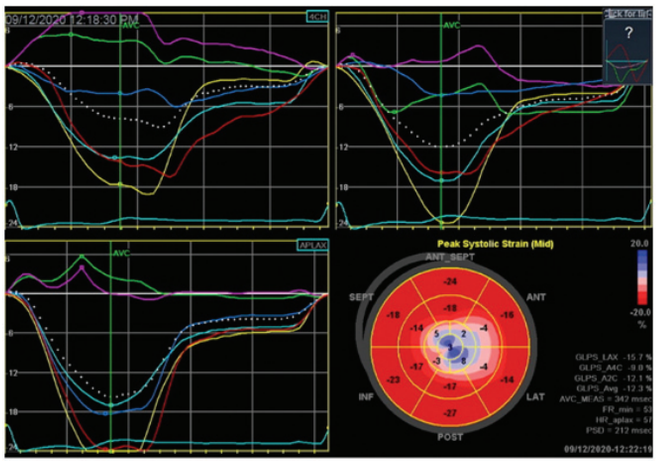

Figure 6: STE demonstrated GLS of -12.3% with paradoxical blue color strain in apex suggestive of apical aneurysm. Very high PSD of 212 ms

Discussion

In general population, the average presentation age of apical HCM is 41 ± 15 with male to female ratio of 1.6 to 2.8. Maron, *et al*. found that out of 28 patients with HCM and LV apical aneurysm only 5% were recognized by echocardiography, remaining patients needed cardiac magnetic resonance imaging (CMR) to prove or refute the diagnosis.[4]

Cavity obliteration in Apical HCM causes ischemia possibly secondary to microvascular obstruction and small vessel disease.[5] The persistence of apical contraction into mid diastole, results in dynamic small vessel obstruction in apical segments, myocardial perfusion defects and chest pain and often associated with paradoxical mid-cavity diastolic flow jet indicating the presence of apical aneurysm.[6]

Apical aneurysms are distinct, thin walled, akinetic/dyskinetic segment of the most distal portion of LV with a relatively wide communication to the main LV cavity in diastole. They are found in 2% of patients with HCM and 13-15% with Apical HCM.[7]

The patient had moderate diastolic dysfunction, evidence of paradoxical diastolic forward flow with distal apical aneurysm, GLS reduced to -12.3% despite preserved LVEF and significantly high PSD of 212ms. Based on these TTE findings, the patient falls in the high-risk category of this relatively benign HCM spectrum in which 25% of patients may experience major complications including: sudden cardiac death, progressive heart failure, thromboembolic events and ventricular arrhythmias necessitating ICD implantation in view of the presence of scarred myocardium at the junction of viable and abnormal tissue where the re-entry circuit occurs, acting as a primary arrhythmogenic focus generating malignant ventricular arrhythmias independent of aneurysm size.[8] Rowin, *et al*. reported that apical HCM patients with LV apical aneurysm experienced major adverse event rate, which is 3 times greater than that of their HCM counterparts.[9]

Management of patients with HCM involves assessment of symptoms, risk stratification and family screening. Treatment options include medical and electrophysiological intervention.[10] Since LV outflow obstruction is absent in apical HCM, therapeutic benefit may be lower than in classic HCM and myomectomy like approaches are not routinely indicated in this group of patients.[11]

Conclusion

This report outlines a clinical presentation of apical HCM with aneurysm formation as identified by contrast TTE. Reduced LV systolic function was better identified by GLS and high PSD predisposing him to malignant ventricular arrythmias. Clinicians should be aware of the manifestations of this entity to avoid misdiagnosis or underdiagnosis in view of associated higher risk of morbidity and mortality.

Author Contribution

All authors share equal effort contribution towards (1) substantial contributions to conception and design, analysis, and interpretation of data; (2) drafting the article and revising it critically for important intellectual content; and (3) final approval of the manuscript version to be published. Yes.

Consent for publication

The authors confirm that consent for submission and publication of this case report including images and associated text has been obtained from the patient.

Potential Conflicts of Interest

None

Competing Interest

None

Sponsorship

None

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