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Multimodality Imaging Demonstrating an Apical Variant Hypertrophic Cardiomyopathy in an Uncommon Pentad

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Abstract

A 79-year-old man was admitted for a transcatheter aortic valve replacement due to severe aortic stenosis. A preoperative chest computed tomography with angiography revealed an apical variant hypertrophic cardiomyopathy with a prominent apical pouch. In addition, there was near-complete obliteration of the left ventricle in the mid to apical aspect during systole suggesting a midventricular gradient. Postoperative transthoracic echocardiography confirmed the apical variant hypertrophic cardiomyopathy with an apical aneurysm and a gradient with a peak velocity of 2 m/s, and mid-cavitary gradient with a peak velocity of 3 m/s. It also revealed a fusiform aneurysmal dilatation of the ascending aorta.

Keywords

apical hypertrophic cardiomyopathy, apical aneurysm, mid-cavitary obstruction, transthoracic echocardiography, computed tomography, CT

Case Presentation

A 79-year-old man presented with dyspnea on exertion due to severe aortic stenosis. The patient has a past medical history significant for diabetes mellitus, ascending aortic aneurysm (45-49 mm), and apical hypertrophic cardiomyopathy (ApHCM) with left ventricle (LV) mid-cavitary obliteration, apical aneurysm (pouch), and apical and mid-cavitary gradient. The LV wall thickness (16-20 mm), apical aneurysm, and cavitary gradient have been stable without symptoms for more than 35 years. Additionally, he has a history of aortic stenosis that developed rapidly over a few years. The aortic valve previously showed a normal mean pressure gradient across the valve with no evidence of stenosis on transthoracic echocardiography (TTE) done 9 years prior to admission. Aortic valve stenosis was first detected by TTE 3 years prior to the admission with a mean pressure gradient across the valve of 33 mm Hg and a valve area of 1.02 cm^2. The patient does not have any family history of hypertrophic cardiomyopathy, arrhythmia, or sudden cardiac death.

On admission, a chest computed tomography with angiography was performed and findings are demonstrated in Figure 1. A TTE was done and showed a mean pressure gradient across the aortic valve of 44 mm Hg with a valve area of 0.92 cm^2 indicating surgical aortic valve replacement (AVR).[1] It also showed a stable ascending aortic aneurysm with a diameter of 47 mm. All surgical approaches were discussed with the patient who opted for transcatheter AVR (TAVR) and deferring treatment of the aortic aneurysm. Subsequently, the patient underwent transesophageal echocardiography-guided right transfemoral TAVR with a 23 mm Edwards Lifesciences pericardial tissue valve.

The postoperative TTE (Figures 2 and 3) showed a successfully implanted aortic valve prosthesis with a mean gradient of 20 mm Hg across the valve and mild regurgitation. It

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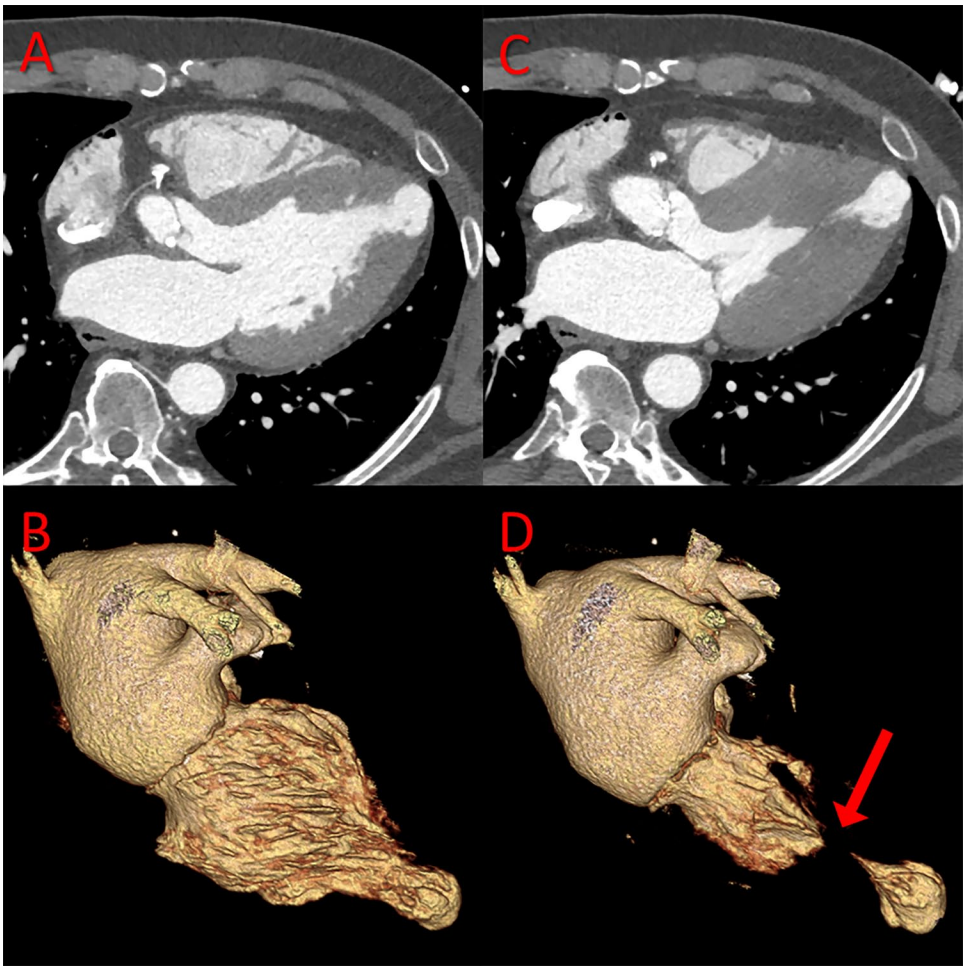

Figure 1. Three-chamber view reconstructions from diastolic (A) and systolic (C) phases of an electrocardiogram-gated contrast-enhanced cardiac computed tomography demonstrate apical-variant hypertrophic cardiomyopathy with apical pouch-aneurysm. Volume-rendered reconstructions of the left ventricular blood pool during diastole (B) and systole (D) demonstrate obliteration of the mid-ventricular cavity during systole (arrow).

also confirmed the pentad: ApHCM, apical aneurysm (pouch), mid-cavitary obliteration with apical and mid-cavitary pressure gradient, ascending aortic aneurysm, and AVR due to severe aortic stenosis (supplementary materials; Videos 1 and 2). The patient has been asymptomatic after the procedure and his postoperative course was free of complications.

Follow-up TEEs done 3 and 11 months postoperatively showed stable ApHCM with stable apical and mid-cavitary gradients and stable apical pouch (Figures 4 and 5). A follow-up computed tomography done 11 months postoperatively showed stable ascending aortic aneurysm and confirmed the pentad stability (Figure 6; Supplementary material Videos 3 and 4).

Discussion

Apical hypertrophic cardiomyopathy is a rare genetic disease characterized by thickening of the LV apex. The condition is frequently sporadic without family history as in our case; however, autosomal dominant inheritance has also been reported.[2] Diagnosis of this condition relies mainly on imaging modalities demonstrating asymmetric hypertrophy of LV mainly at the apex with a wall thickness of 15 mm or more and an apical to posterior wall thickness ratio of 1.5 mm or more.[3]

ApHCM can occur with or without mid-cavitary obliteration that is associated with a higher risk of heart failure, arrhythmia, stroke, and sudden cardiovascular death.[4] Apical aneurysms occur in 15% of patients with ApHCM as a result of myocardial scarring caused by increased LV wall stress and elevated systolic pressures. The presence of apical aneurysms can double the risk of thromboembolic events.[5]

The hypertrophied apex of the LV in ApHCM limits coronary blood flow reserve and could predispose to myocardial ischemia and subsequent ventricular arrhythmia. Moreover, atrial arrhythmia such as atrial fibrillation

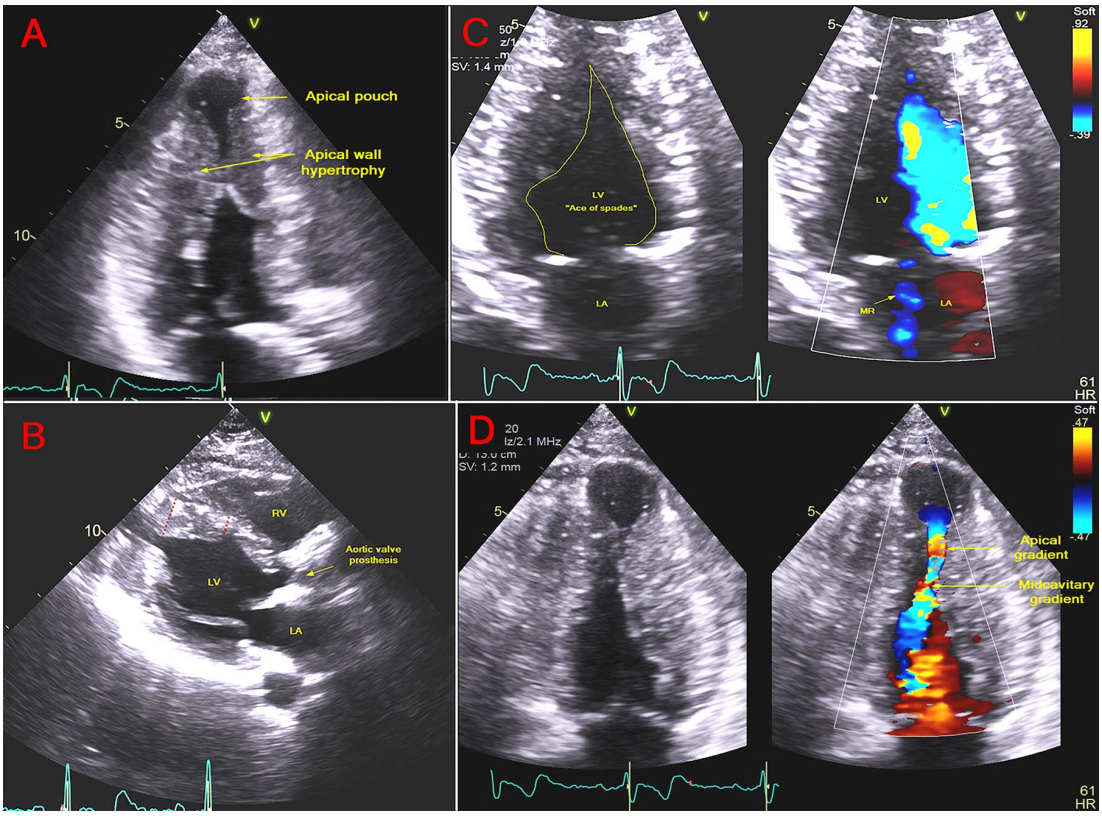

Figure 2. Transthoracic echocardiography: 2-chamber systolic frame (A) showing apical hypertrophy with apical aneurysm (pouch) and mid-cavitary obstruction. Parasternal long axis view (B) shows left ventricle (LV) apical hypertrophy measuring 20 mm. Two-chamber view of LV (C, left) shows "Ace of spades" morphology during systole due to apical hypertrophic cardiomyopathy; color Doppler shows mitral regurgitation (C, right). Two-chamber systolic view (D) shows apical and mid-cavitary pressure gradient with apical and mid-cavitary peak flow of 2, 3 m/s, respectively.

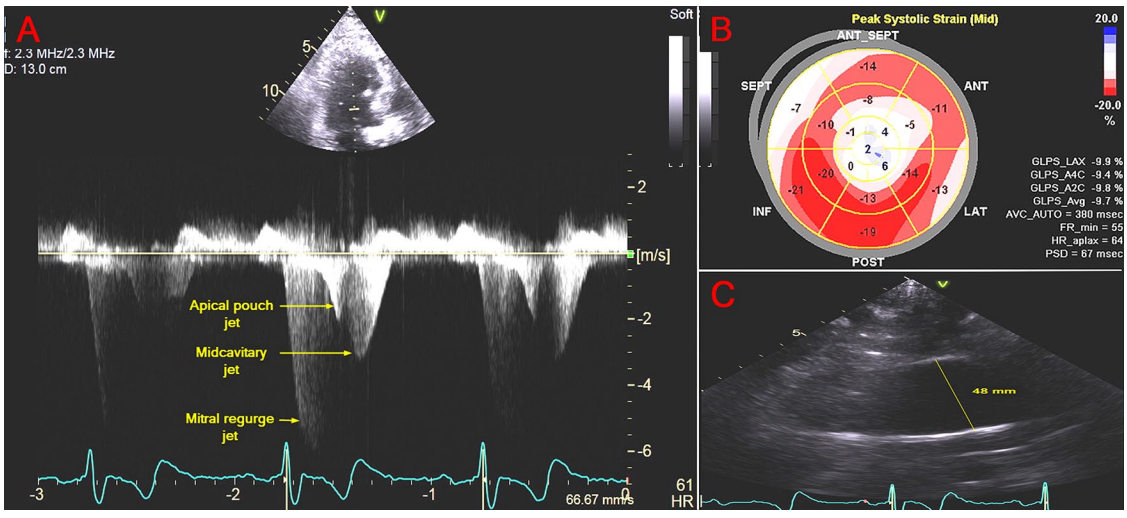

Figure 3. Continuous wave spectral Doppler (A) shows 3 signals: mitral regurgitation peak velocity (6.1 m/s), mid-cavitary peak velocity (3 m/s), and apical left ventricle peak velocity (2 m/s). Polar map of 17 segments strain (B) shows marked reduction (−9.7%) with marked apical and mid-segment involvement. Ascending aortic aneurysm with midlevel diameter of 48 mm (C).

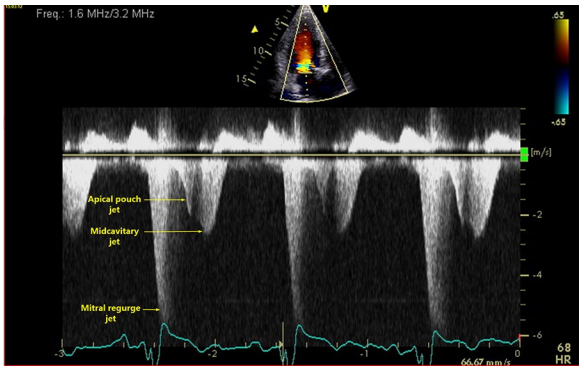

Figure 4. Continuous wave spectral Doppler (3 months postoperatively) shows 3 signals: MR peak velocity (6 m/s), mid-cavitary peak velocity (3 m/s), and apical LV peak velocity (2 m/s).

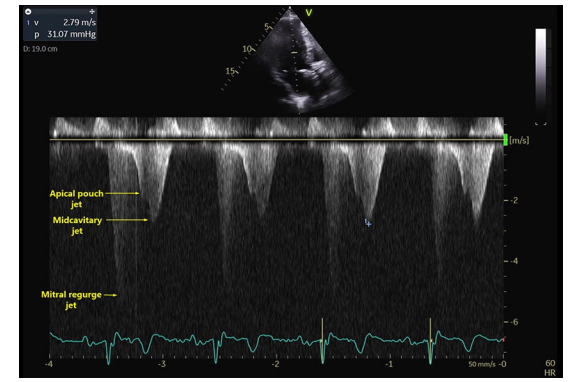

Figure 5. Continuous wave spectral Doppler (11 months postoperatively) shows 3 signals: mitral regurgitation peak velocity (5.9 m/s), mid-cavitary peak velocity (2.8 m/s), and apical left ventricular peak velocity (2 m/s).

occurs frequently in such population and is hypothesized to be a consequence of left atrial enlargement caused by LV diastolic dysfunction.[6] Cardiovascular death in ApHCM can result from any of the complications mentioned above. Recent data suggest up to 4% annual cardiac death associated with ApHCM, approaching those associated with classic HCM.[7]

In our case, the presence of ascending aortic aneurysm and TAVR due to severe aortic stenosis adds more complexity to this condition. The ideal surgical approach in such a case is open AVR with concomitant repair of the aortic aneurysm. However, the patient opted for TAVR and deferred aortic aneurysm repair with close follow-up. The patient was scheduled for regular 6 months follow-up with repeating TTE and computed tomography with angiography. To date, the patient's follow-up course has shown stable aneurysm diameter and stable prosthetic valve with mild regurgitation for a year.

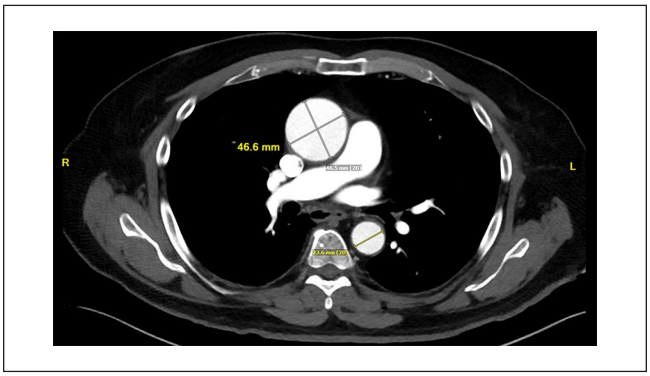

Figure 6. Cross-sectional computed tomography angiography of the chest (11 months postoperatively) showing stable ascending aortic aneurysm with a diameter of 46.6 mm.

In patients with hypertrophic cardiomyopathy, β-blockers, relieving the symptoms and reducing the burden of ventricular arrhythmia, are the first-line medical treatment.[8] Other options for managing ventricular and atrial arrhythmias include amiodarone and procainamide. Implantable cardioverter defibrillator is recommended for patients with high risk including those with history of cardiac arrest, syncope, episodes of sustained or nonsustained ventricular tachycardia on serial Holter monitoring, or family history of sudden cardiac death.[9] Our patient had serial Holter monitoring that showed 99% normal sinus beats with <1% premature ventricular complexes and <1% premature atrial complexes. One episode of ventricular tachycardia for 4 beats was detected only once. In light of the asymptomatic condition of our patient and his negative family history, implantable cardioverter defibrillator was not warranted in his case and the patient continued on chronic β-blockers therapy. Surgical treatment with transapical myectomy is indicated in those with symptomatic LV mid-cavitary obliteration and evidence of heart failure, as it can reduce gradients, increase end-diastolic dimensions, and improve heart failure symptoms.[10]

Declaration of Conflicting Interests

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Ethics Approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed Consent

Verbal informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

Supplemental Material

Supplemental material for this article is available online.

References

1. Nishimura RA, Otto CM, Bonow RO, et al. 2017 AHA/ACC focused update of the 2014 AHA/ACC Guideline for the management of patients with valvular heart disease: a report of the American College of Cardiology/American Heart Association Task Force on clinical practice guidelines. *J Am Coll Cardiol*. 2017;70:252-289.
2. Arad M, Penas-Lado M, Monserrat L, et al. Gene mutations in apical hypertrophic cardiomyopathy. *Circulation*. 2005;112:2805-2811.
3. Eriksson MJ, Sonnenberg B, Woo A, et al. Long-term outcome in patients with apical hypertrophic cardiomyopathy. *J Am Coll Cardiol*. 2002;39:638-645.
4. Kim H, Park JH, Won KB, et al. Significance of apical cavity obliteration in apical hypertrophic cardiomyopathy. *Heart*. 2016;102:1215-1220.
5. Maron MS, Finley JJ, Bos JM, et al. Prevalence, clinical significance, and natural history of left ventricular apical aneurysms in hypertrophic cardiomyopathy. *Circulation*. 2008;118:1541-1549.
6. Patten M, Pecha S, Aydin A. Atrial fibrillation in hypertrophic cardiomyopathy: diagnosis and considerations for management. *J Atr Fibrillation*. 2018;10:1556.
7. Klarich KW, Jost CHA, Binder J, et al. Risk of death in long-term follow-up of patients with apical hypertrophic cardiomyopathy. *Am J Cardiol*. 2013;111:1784-1791.
8. Spoladore R, Maron MS, D'Amato R, Camici PG, Olivotto L. Pharmacological treatment options for hypertrophic cardiomyopathy: high time for evidence. *Eur Heart J*. 2012;33:1724-1733.
9. Maron BJ. Risk stratification and prevention of sudden death in hypertrophic cardiomyopathy. *Cardiol Rev*. 2002;10:173-181.
10. Nguyen A, Schaff HV. Surgical myectomy: subaortic, midventricular, and apical. *Cardiol Clin*. 2019;37:95-104.