

When Apical Hypertrophy meets Arrhythmia: A case of Yamaguchi Syndrome in an Indian Male

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Introduction

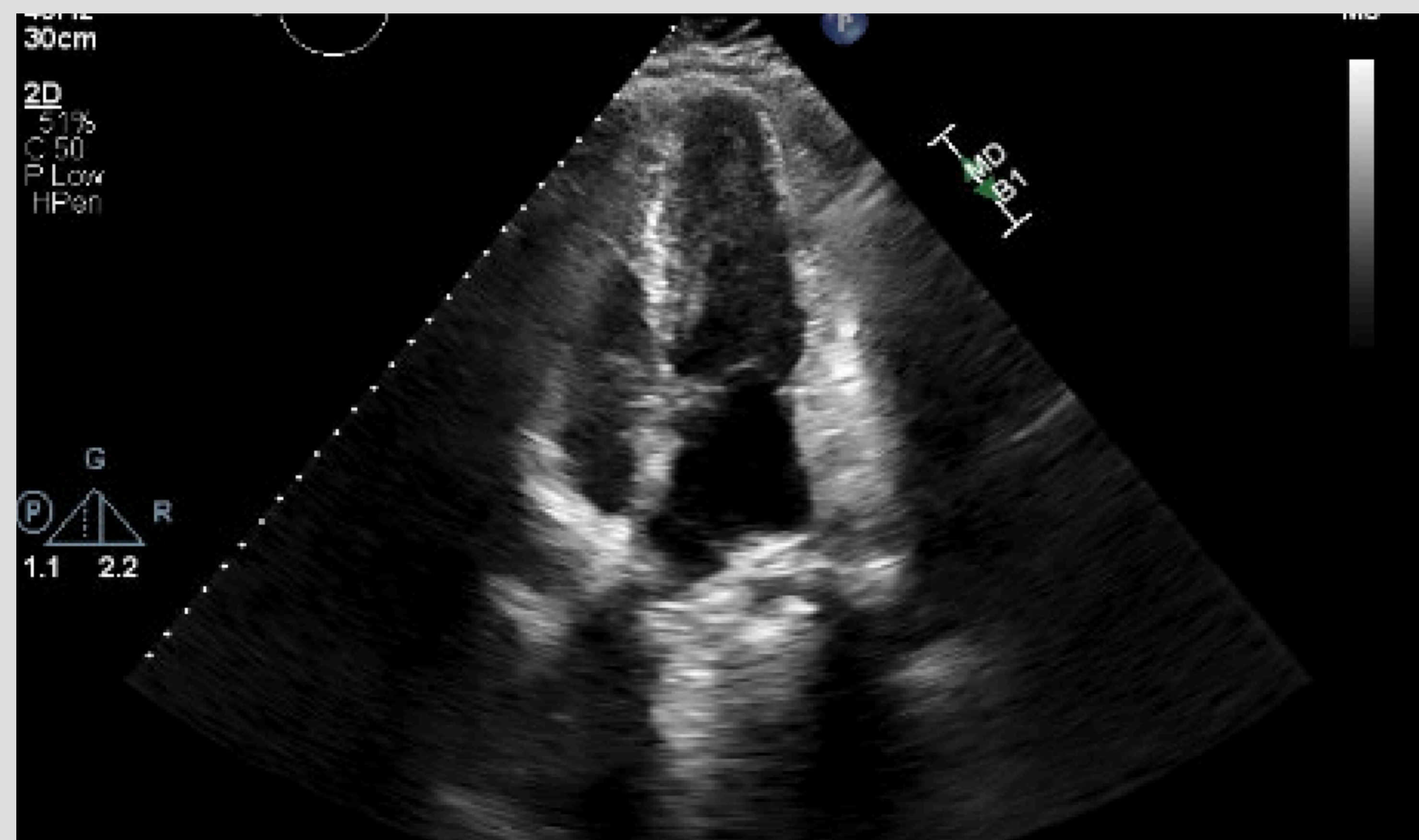
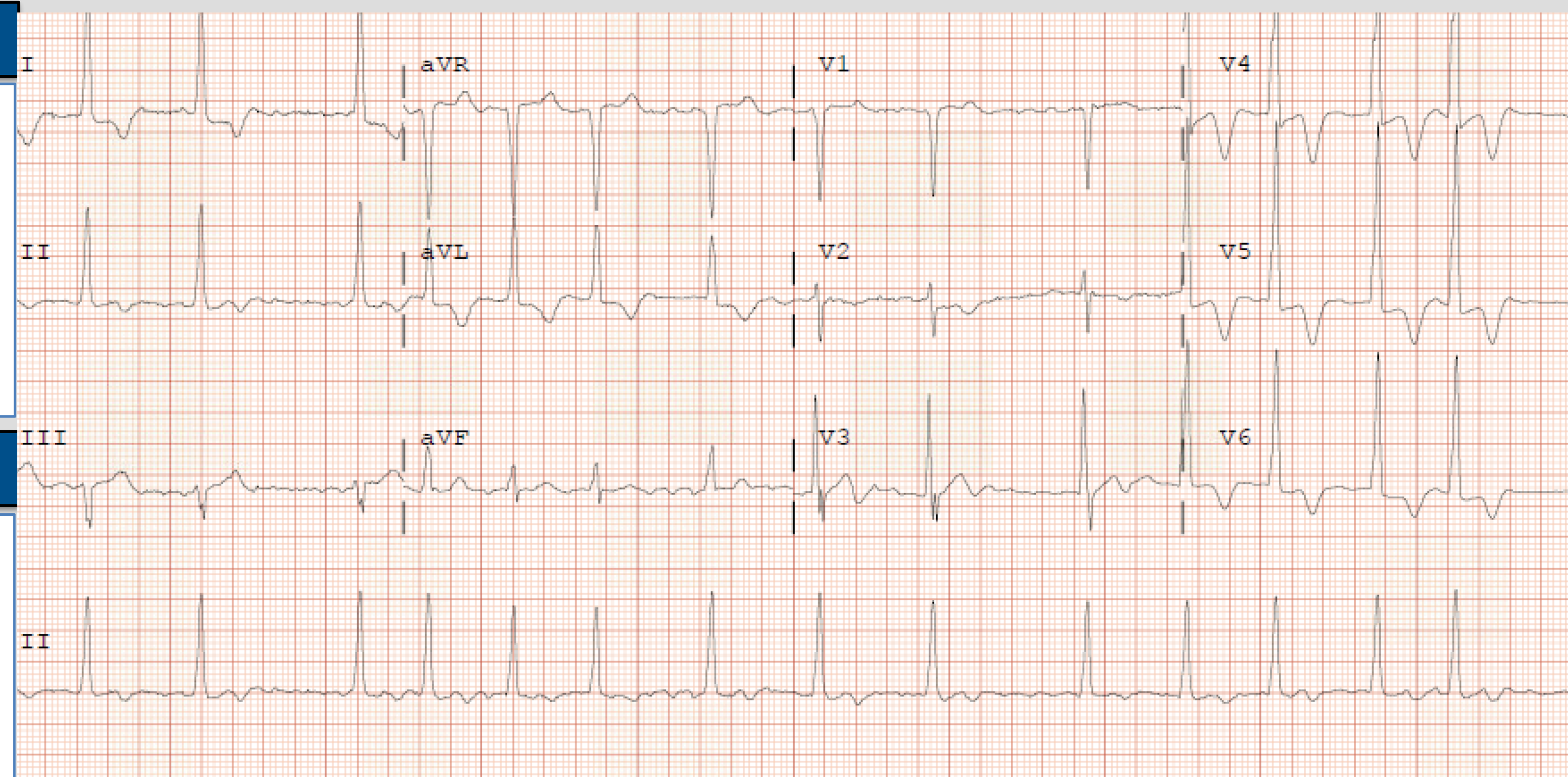
Yamaguchi syndrome is a rare variant of hypertrophic cardiomyopathy (HCM) that is characterized by apical hypertrophy and aneurysm, often associated with a higher risk of arrhythmias and sudden cardiac death. This condition is typically diagnosed using echocardiography and EKG findings. While Yamaguchi syndrome is more commonly reported in Asian populations, its presentation in Indian Bengali patients remain less studied.

Case Description

A 54 year old male with past medical history of CAD with chronic occlusion of RCA collaterals, type 2 diabetes mellitus, apical variant hypertrophic cardiomyopathy with apical aneurysm on warfarin, hypertension, dyslipidemia, mild intermittent asthma who presented to ED with shortness of breath. He was noted to be in A.fib during the ED visit which could have been contributing to patient's shortness of breath. He does not have any known family history of hypertrophic cardiomyopathy. He denies any family history of premature death in first-degree relatives before age of 40. Denies any history of syncope. Echo revealed mild concentric LVH, apical hypertrophy with apical aneurysm and poorly visualized clot type Doppler flow out of the aneurysm, heavy apical trabeculations raising suspicion for noncompaction, stage I diastolic dysfunction. EKG revealed Atrial flutter/A fib pattern, LVH, deep T wave inversions in V2 to V6 and in lateral leads. Holter monitor revealed 11 beats of non sustained VT which was monomorphic in nature at a rate of 153.

Further Management

EP study during that time could not induce any sustained ventricular tachycardia or V.Fib's. As patient has hypertrophic cardiomyopathy with non sustained VT's, patient was recommended ICD placement for primary prevention of sudden cardiac death which patient did not get done. Due to diagnosis of A.fib contributing to hospitalization, he underwent cardioversion to restore sinus rhythm. He was recommended to follow with EP for AICD.



Discussion

Hypertrophic cardiomyopathy (HCM) is a genetic disorder characterized by abnormal thickening of the heart muscle, which can lead to obstructed blood flow, arrhythmias, and sudden cardiac death. The apical variant of HCM, commonly known as Yamaguchi syndrome, is characterized by hypertrophy confined to the apex of left ventricle and formation of an apical aneurysm. This variant is most commonly observed in East Asian populations but has also been reported in other ethnic groups. Echo findings of apical hypertrophy and apical aneurysm along with EKG finding of deep T wave inversions in precordial leads support the diagnosis of Yamaguchi syndrome. The presence of non sustained VT further increases the risk of sudden cardiac death, a concern in patients with HCM and its variants. As per ACC/AHA 2024 guidelines, ICD placement for primary prevention of sudden cardiac death is reasonable if at least one major risk factor is present which include left ventricular apical aneurysm with transmural scar, non sustained ventricular tachycardia, family history of sudden cardiac death, massive left ventricular hypertrophy (greater than 30mm), recent unexplained syncope, or LV systolic dysfunction (EF <50%). Atrial fibrillation, a common arrhythmia in patients with HCM, may contribute to worsening symptoms such as SOB, as seen in this patient.

Conclusion

This case highlights the complexity of Yamaguchi syndrome particularly in Indian Bengali male, less stated population with multiple comorbidities. This underscores the importance of early diagnosis and management to mitigate the risk of sudden cardiac death in patients with apical variant hypertrophic cardiomyopathy. This also warrants further research and awareness of Yamaguchi syndrome in non-East Asian populations and other diverse ethnic groups.