When Apical Hypertrophy Meets Arrhythmia: A Case of Yamaguchi Syndrome in an Indian Male

Authors

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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 electrocardiographic findings. While Yamaguchi syndrome is more commonly reported in Asian populations, its presentation in Indian Bengali patients remains less studied.

Case Presentation

This is a 54-year-old male with past medical history of CAD with chronic occlusion of right coronary artery (RCA) with collaterals, type 2 diabetes mellitus, apical variant hypertrophic cardiomyopathy with apical aneurysm on warfarin, hypertension, dyslipidemia, mild intermittent asthma who presented to emergency department with shortness of breath. He was noted to be in atrial fibrillation (A-fib) during the emergency department visit which could have been contributing to patient's shortness of breath. He has no family history of hypertrophic cardiomyopathy, premature sudden death, or syncope. Echocardiogram revealed mild concentric left ventricular hypertrophy(LVH), apical hypertrophy with apical aneurysm, and a poorly visualized clot type Doppler of flow out of the aneurysm, heavy apical trabeculations raising suspicion for noncompaction,and stage I

diastolic dysfunction. EKG revealed sinus rhythm, LVH, deep T wave inversions in V2 to V6 and in lateral leads. Holter monitor was done which revealed 11 beat nonsustained VT on Holter monitor which was monomorphic in nature. Patient was referred for EP study and possible ICD placement for prevention. EP study during that time could not induce any sustained ventricular tachycardia or V-fib's. As patient has hypertrophic cardiomyopathy with nonsustained VT, the patient was recommended ICD placement for primary prevention of sudden cardiac death which patient did not get done. Due to diagnosis of A-fib likely contributing to recent hospitalization with shortness of breath, he was referred to electrophysiology for cardioversion to restore sinus rhythm. Patient is already on warfarin for hypertrophic cardiomyopathy with apical aneurysm. He was recommended to follow closely with EP for possible ICD placement for primary prevention as well.

Discussion

Hypertrophic cardiomyopathy (HCM) is a genetic disorder marked by abnormal myocardial thickening that can cause outflow obstruction, arrhythmias, and sudden cardiac death. The apical variant, or Yamaguchi syndrome, features hypertrophy confined to the left-ventricular apex and may include apical aneurysm formation. Although most common in East Asian populations, it occurs across various ethnic groups. In this patient, echocardiographic evidence of apical hypertrophy and an apical aneurysm, along with deep T-wave inversions on EKG, supports the diagnosis of Yamaguchi syndrome (1). The presence of nonsustained ventricular tachycardia increases the risk of sudden cardiac death (2). Although the EP study did not induce sustained VT, the patient remains high risk, and ICD placement for primary prevention is guideline-supported. ACC/AHA 2024 guidelines recommend considering an ICD when at least one major risk factor is present, including apical aneurysm with scar, nonsustained Ventricular tachycardia, family history of sudden

cardiac death, LV wall thickness >30 mm, unexplained syncope, or LV systolic dysfunction (EF <50%) (3). Atrial fibrillation, common in HCM, likely contributed to this patient's shortness of breath and symptom worsening.

References

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