CLINICAL VIGNETTE

A Case of Severe Hypertrophic Cardiomyopathy: Defibrillator Consideration

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Abstract

Hypertrophic cardiomyopathy is an inherited heart condition that carries a 1% risk of sudden cardiac death. ICD placement is indicated in patients with 2 or more major risk factors. This case illustrates the management for a patient with HCM that presented with mild symptoms but only one major risk factor, a severe thickening of the left and right ventricles. Consideration for ICD placement requires specific risk stratification criteria; however, current evidence does not show a significant benefit of ICD placement in patients with only ventricular hypertrophy as a risk factor. In summary, the benefits that ICD placement can provide need to be carefully weighed against its risks in patients with only one major risk factor and who are mildly symptomatic.

Background

Hypertrophic cardiomyopathy (HCM) is a condition with complex pathophysiology that occurs in 1 in 500 patients in the general population. While many people with HCM remain asymptomatic, sudden cardiac death (SCD) is a severe consequence of HCM that may lead to consideration of ICD placement. Consideration for ICD is indicated for patients with high risk factors, but specific risk stratification criteria are unclear for patients with intermediate risk factors, as presented in this case.

Case Presentation

A 34 year-old male with no significant past medical history presented to an outside physician with worsening shortness of breath with exertion. Throughout his teenage years and young adulthood, he became short of breath when he exercised heavily and was unable to run long distances, but generally had no limitations with routine activities. In the previous 3 years, he became more short of breath when playing basketball and noticed a decrease in exercise tolerance, with two episodes of lightheadedness without loss of consciousness. These symptoms resolved with rest. The patient denied ever having syncope, palpitations, shortness of breath at rest, orthopnea, or paroxysmal nocturnal dyspnea.

On initial physical exam, he was found to have a 2/6 systolic murmur that began after S1 and was most prominent at the mid-left sternal border. The murmur decreased with Valsalva maneuver, and there was no S3 or S4. No other murmurs were appreciated. The echocardiogram (ECG) showed normal left ventricular size with pronounced left ventricular hypertrophy (LVH) (Figure 1), and a peak left ventricular outflow tract (LVOT) gradient of 6 mmHg with and without Valsalva. The right ventricle showed hypertrophy with preserved systolic function and normal pulmonary artery pressure. Both atria were normal sized. Stress echocardiogram was negative by EKG and echocardiographic criteria. There was no increase in LVOT gradient across with stress.

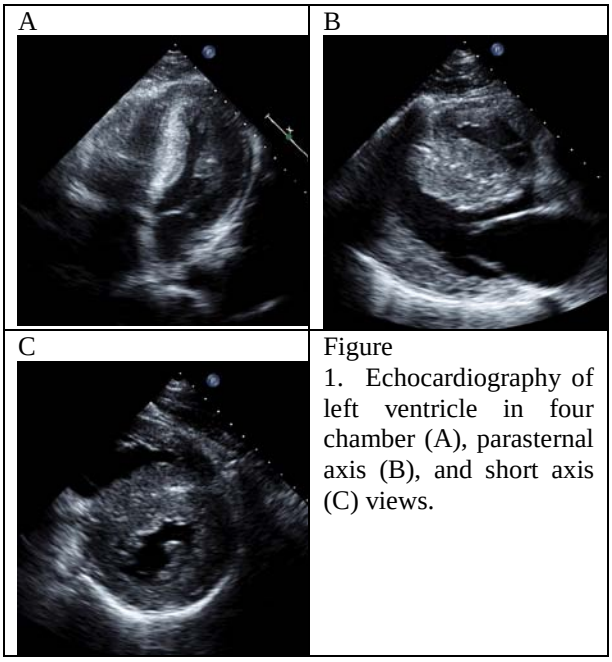

Figure 1. Echocardiography of left ventricle in four chamber (A), parasternal axis (B), and short axis (C) views.

The patient was diagnosed with hypertrophic cardiomyopathy. There was no evidence of cardiovascular obstruction with severe exercise and no family history of cardiovascular disease, syncope, arrhythmias, or sudden cardiac death. ICD placement was offered for prevention of SCD. Patient declined placement of ICD and followed up routinely.

Discussion

Hypertrophic Cardiomyopathy (HCM) is a frequently inherited cardiovascular disease with many known autosomal dominant forms. This disease can appear at any age, and is the most common cause of sudden cardiac death in young people[1]. However, HCM can often present with no symptoms. Clinically, patients with HCM may present with a heart murmur, or with abnormal ECG showing giant T-wave inversions. Diagnosis of HCM is usually by echocardiogram, sometimes facilitated with cardiac magnetic resonance (CMR) imaging to detect for presence of myocardial scarring.

ICD placement can be a cost-effective method for prevention of sudden cardiac death (SCD). Based on observational studies, major risk factors in HCM patients which correlate with SCD are shown in Table 1, and have been used to stratify risk for ICD placement[2]. While the absence of these risk factors has a strong negative predictive value, each factor has only a positive predictive value for SCD of approximately 20%[1]. In patients with fewer than 3 risk factors, there was no association between the number of risk factors and the probability of appropriate ICD discharges[1,3]. Hence, individual risk needs to be considered in the overall clinical scenario for assessment for ICD placement.

Table 1. Risk Factors for Sudden Cardiac Death

Major Risk Factors for Sudden Death in HCM	Possible Risk Factors for Sudden Death in Individuals
Cardiac Arrest (ventricular fibrillation)	Atrial Fibrillation
Spontaneous Sustained Ventricular Tachycardia	Myocardial Ischemia
Family history of premature sudden death	LV outflow obstruction
Unexplained syncope	High-risk mutation
LV thickness greater than or equal to 30 mm	Intense Physical Exertion
Abnormal exercise blood pressure	
Nonsustained ventricular tachycardia (Holter)	

HCM= Hypertrophic Cardiomyopathy, LV= Left Ventricular

Our patient's major risk factor is a LV wall thickness >30mm. While LV thickness correlates with sudden cardiac death, there is no specific threshold that significantly increases the risk of SCD (1). A LV thickness of >30mm has a positive predictive value of 16% for SCD. Moreover, the 5-year survival rate of patients with a thickness >30mm as the only risk factor is not significantly different than patients with wall thickness <30mm, consistent with the data showing that high-risk mutations associated with SCD have only mild hypertrophy[4,5]. Given that this patient had no other risk factors and is young, there is questionable benefit of ICD placement.

The ventricular thickness with otherwise normal heart function and mild symptoms prompted further risk assessment with cardiac magnetic resonance (CMR) imaging. CMR allows for measurement of wall thickness and may be useful when echocardiography results are equivocal. It is also sensitive in detecting a subgroup of HCM patients with apical aneurysms[6]. Late gadolinium enhancement (LGE) on CMR is sensitive for cardiac fibrosis, a marker associated with SCD[7,8].

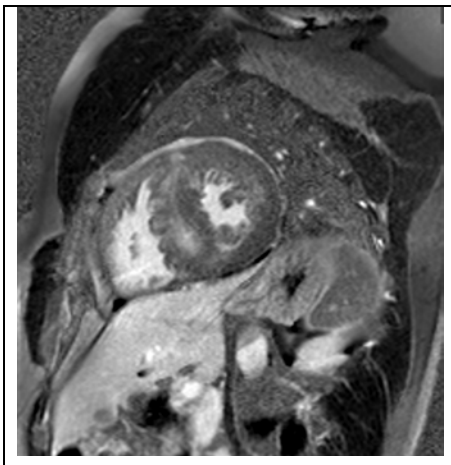

Figure 2. Cardiac magnetic resonance imaging of patient H.C. demonstrating late gadolinium enhancement in the left ventricle.

Our patient's CMR showed enhanced LGE in LV septum with no apical aneurysm (Figure 2). This result, per se, is an insufficient predictor of SCD primarily because LGE is commonly found in HCM but incidence of SCD in HCM patient is low (~1%)[9]. Hence, CMR plays an adjunctive role with echocardiography in consideration ICD placement.

Complications of ICD include inappropriate shocks, infection, and device malfunction[10]. Two studies with patients with ICD demonstrated a rate of inappropriate shocks that range from 23% to 27% and appropriate shocks in 16% to 20%[3,10]. Since the complication rates are significant, it is important to

carefully weigh them against the benefits of ICD placement on an individual basis.

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FIGURE LEGEND

Figure 1A-C: Echocardiography of left ventricle in four chamber (A), parasternal axis (B), and short axis (C) views.

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