CASE REPORT

Eccentric apical hypertrophic cardiomyopathy unmasked by multimodality imaging: an uncommon but missed cause of out of hospital cardiac arrest

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SUMMARY

A woman in her late 50s experienced a witnessed, sudden out of hospital cardiac arrest. Initial workup included coronary angiography, transthoracic echocardiogram and a CT scan of the chest to rule out pulmonary embolus. She was subsequently discharged home without an implantable cardioverter defibrillator (ICD) or a life vest. On follow-up at another facility, an ICD was placed and a Holter monitor showed no ventricular ectopy. Further transthoracic echocardiographic images were obtained, which were suggestive of apical hypertrophic cardiomyopathy. A limited transthoracic echocardiogram with contrast was performed, which did not elucidate the hypertrophy. However, eccentric left ventricular apical wall hypertrophy was visualised by a coronary CT scan.

BACKGROUND

Hypertrophic cardiomyopathy (HCM) is the most commonly inherited cardiomyopathy. Apical HCM is a morphological variant of HCM, with left ventricular (LV) hypertrophy confined to the cardiac apex, accounting for approximately 10% of patients with HCM.¹

TTE is the initial diagnostic tool in the evaluation in HCM, however, recognition of structural alterations at the apex can be challenging. Thus, a multimodality imaging approach (CT and cardiac MRI) may be warranted if there is an index of suspicion for the diagnosis of apical HCM.^{2–3} American College of Cardiology/American Heart Association (ACC/AHA) guidelines recommend use of an intravenous echocardiographic contrast agent if the diagnosis of apical HCM or severity of hypertrophy is in doubt (ACC/AHA class IIa indication, Level of Evidence: C).⁴

Echocardiographic features of apical HCM consist of thickened apical segments producing a 'spade-shaped', small apical cavity. Electrocardiographic signs include negative T-waves in the precordial leads (found in 93% of patients), followed by LV hypertrophy (65% of patients).⁵ Since LV hypertrophy in apical HCM predominantly affects the apex as opposed to the base, it is not associated with LV outflow tract obstruction. Hence, symptoms in apical HCM relate to diastolic dysfunction as opposed to obstructive physiology. Many patients with apical HCM are asymptomatic or manifest only minimal symptoms, resulting in diagnosis often being an incidental finding,⁶ and sudden cardiac arrest is rare in apical HCM.⁵ Given the low rates of cardiovascular

mortality, implantable cardioverter defibrillator (ICD) placement is utilised infrequently in apical HCM and is recommended for patients with HCM with prior documented cardiac arrest, ventricular fibrillation (VF) or haemodynamically significant ventricular tachycardia (Class I Level of Evidence: B).⁴

This is a rare presentation of apical HCM, and outlines the appropriate investigative strategy and treatment for patients with sudden out of hospital cardiac arrest (OHCA).

CASE PRESENTATION

An asymptomatic 56-year-old woman with no cardiac history or relevant family history experienced a witnessed, sudden collapse while taking an afternoon walk. Cardiopulmonary resuscitation was promptly initiated by a neighbour. The initial rhythm on paramedic arrival was VF and the patient required multiple shocks to return her to sinus rhythm. She was transferred to an outside facility for hypothermia protocol.

INVESTIGATIONS

The patient's subsequent hospital course was unremarkable. Workup for the cause of arrest included coronary angiography, a TTE and a CT scan of the chest to rule out pulmonary embolus, all of which were reported as normal. The patient was discharged after a 2-week hospital stay. She was evaluated at another facility for follow-up. A stress TTE was reported as normal and no evidence of HCM was identified. Repeat Holter monitoring showed occasional episodes of supraventricular tachycardia (SVT) and a 14 beat run of nonsustained ventricular tachycardia. An exercise treadmill stress test was reported as normal.

Given an unclear aetiology of sudden cardiac death, the patient was referred to Mayo Clinic's comprehensive sudden cardiac death programme for further evaluation and for recommendations regarding family screening. Her physical examination was unremarkable. Holter monitoring showed nine 3–10 beat runs of SVT but no ventricular ectopy. ST segment depression was noted to 2.1 mm in leads II and V4–V6 (figure 1). A treadmill exercise stress test revealed 2 mm ST segment depression. A signal average ECG was negative for late potentials. In a comprehensive, non-contrast TTE, apical hypertrophy was suspected, so a limited TTE with intravenous contrast was performed but did not elucidate the hypertrophy (video 1). Apical HCM was diagnosed during

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Figure 1 ECG with mild ST-segment depression.

coronary CT angiography, which was performed to exclude anomalous coronary artery origins given unavailability of outside coronary angiogram at the time of evaluation. Coronary artery assessment was normal. However, eccentric LV apical wall hypertrophy was visualised, consistent with atypical apical HCM (figure 2, video 2). While late enhanced multi-slice CT has been considered for detection of fibrosis in a small series of patients with HCM, it is not routinely used clinically and was not performed in this patient's care.⁷

TREATMENT

Despite her VF-documented out-of-hospital cardiac arrest, the patient was initially discharged after a 2-week hospital stay on β -blocker therapy but without an ICD or even a life vest. On follow-up at a separate outside facility, she eventually received an ICD.

OUTCOME AND FOLLOW-UP

Coronary CT scan identified 18 mm eccentric wall thickness of the apical portion of the left ventricle, consistent with atypical apical HCM.

The patient remains active without subsequent ICD discharges or cardiac events.

DISCUSSION

Apical HCM was described initially in Japan by Sakamoto *et al*⁸ in 1976. Of all the morphological subtypes, apical HCM is considered to have a more benign prognosis, with a lower rate of morbidity and mortality. Studies looking at long-term outcomes in these patients showed that the annual rate of cardiovascular mortality is <0.1%.⁵⁻⁹ Although considered a less severe form of HCM, recent studies have shown that the 20-year survival is

Figure 2 CT image of apical measurement.

less than expected (47% vs 60%, $p=0.001$) when compared to age-matched controls.¹⁰

The development of arrhythmias, most likely atrial, is the most common complication that arises in apical HCM, occurring in approximately 10% of patients.⁵ Sickle cell disease (SCD) has been reported to occur as a result of ventricular arrhythmias but remains a rare occurrence in patients with apical HCM. Previous studies have shown that the relative risk for patients with all types of HCM to suffer from SCD is 1.2% per year.¹¹

Although massive hypertrophy (thickness >30 mm) was not present, the patient suffered an OHCA. Patients with OHCA present a diagnostic challenge, and require detailed history and a thorough evaluation. Although the current approach is heterogeneous, it is recognised that all patients should have assessment of electrolytes, cardiac enzymes and a toxicology screen. Imaging studies, such as a chest X-ray, allow for evaluation of heart size and assessment of pulmonary vasculature. A two-dimensional (2D) echocardiogram with Doppler interrogation is essential to rule out congenital and structural heart disease, including the presence of HCM, as well as to assess for presence of regional wall motion abnormalities. Cardiac angiography is needed for

Video 1 Transthoracic echocardiogram with contrast showing no apical hypertrophy.

Video 2 CT angiography showing eccentric left ventricular apical wall hypertrophy.

Table 1 Components of a sudden cardiac death evaluation

Evaluation	Helps in assessment/identification of:
Laboratory	<ul style="list-style-type: none"> ▶ Electrolyte abnormalities ▶ Endocrinopathies
ECG	<ul style="list-style-type: none"> ▶ Brugada syndrome ▶ Cardiomyopathies (ARVC/hypertrophic) ▶ Conduction disturbances (bundle branch blocks, atrio-ventricular blocks) ▶ Ischaemia ▶ Long QT syndrome ▶ Prior silent myocardial infarction ▶ Wolff-Parkinson-White syndrome
Ambulatory Holter monitoring	<ul style="list-style-type: none"> ▶ Recurrent sustained or nonsustained arrhythmias
Transthoracic echocardiography	<ul style="list-style-type: none"> ▶ Cardiomyopathies (ARVC/dilated/hypertrophic) ▶ Coronary artery disease (regional wall motion abnormalities, left ventricular dysfunction) ▶ Congenital structural heart disease ▶ Valvular pathologies (aortic stenosis)
Cardiac MRI	<ul style="list-style-type: none"> ▶ Cardiomyopathies (ARVC, dilated, hypertrophic, infiltrative) ▶ Congenital structural heart disease ▶ Myocardial scar/fibrosis ▶ Myocarditis
Exercise stress testing	<ul style="list-style-type: none"> ▶ Catecholaminergic polymorphic ventricular tachycardia ▶ Long QT syndrome
Genetic testing	<ul style="list-style-type: none"> ▶ Genetically identifiable diseases (Brugada, long QT syndrome, ARVC, HCM)
Coronary angiography	<ul style="list-style-type: none"> ▶ Anomalous coronary artery origin
Electrophysiological study	<ul style="list-style-type: none"> ▶ Accessory pathway (Wolff-Parkinson-White syndrome) ▶ Atrioventricular conduction abnormalities ▶ Inducible ventricular arrhythmias

ARVC, arrhythmogenic right ventricular cardiomyopathy; HCM, hypertrophic cardiomyopathy.

the evaluation of coronary artery obstruction and anomalies. Cardiac CT angiography may be performed as well for coronary artery assessment, and to determine cardiac structure and function. MRI is useful for tissue characterisation with delayed hyper-enhancement/late gadolinium enhancement (scar localisation) and should be carried out prior to ICD implantation, as the presence of an ICD is a relative contraindication for a MRI. An ECG will provide valuable information regarding prolongation of the QT interval, the presence of Brugada syndrome, pre-excitation or other evidence of structural or electrical heart disease. In some instances, such as in patients who are found to have HCM, dilated cardiomyopathy, prolong QT syndrome or other inherited cardiac conditions, genetic testing is recommended, not only to confirm the diagnosis but to screen close relatives (table 1).

Previous studies have professed the need for multi-imaging modalities in the diagnosis of HCM, specifically apical HCM, stating that routine echocardiography has limitations when viewing the apex, and may not be sensitive enough to detect hypertrophy and may have led to the underreporting of this condition.¹² In this case, the initial diagnosis was missed following a 2D echocardiogram, as well as in a follow-up 2D echocardiogram with IV contrast. The diagnosis was performed, however, following definitive images from cardiac CT. Cardiac MRI, which would be a preferred modality to assess suspected apical HCM, was not used, given prior ICD implantation. This reinforces the need for a multi-imaging modality, as opposed to echocardiography alone, especially in cases of OHCA where identification of a cause is essential.

Missing the cause of an OHCA may have severe consequences, not only to the patient but also to the patient's family. Specific guidelines have been published regarding the screening of first and second degree relatives of patients with conditions such as, but not limited to, HCM and long QT syndrome. Every effort should be made to exclude these conditions.

The use of an ICD as prevention following survival of an OHCA is well established. The 2011 ACCF/AHA guidelines on the diagnosis and treatment of hypertrophic cardiomyopathy, states that an ICD is recommended for patients with HCM with prior documented cardiac arrest, VF or haemodynamically significant ventricular tachycardia (Class I Level of Evidence: B).¹³ These patients should also have very close and regular follow-up at a centre with expertise in sudden cardiac death.

In conclusion, patients who suffer from an OHCA should be evaluated thoroughly and should almost never be dismissed without an ICD, as in this patient's case. As demonstrated, single modality imaging, such as TTE, may not be sufficient to identify the cause of the arrest. In selected cases, cardiac MRI may better define cardiac anatomy compared to echocardiography; however, we continue to advocate multimodality imaging, when available, given the unique benefits of each modality (tissue characterisation sequences, dynamic Doppler haemodynamic assessment, etc). These patients, per published guidelines, should all receive an ICD prior to hospital dismissal and must have close follow-up at a centre with expertise in this condition. Failure to complete a proper work up and treatment may lead to catastrophic results for the patient and their family.

Learning points

- ▶ Patients who suffer from an out of hospital cardiac arrest (OHCA) should be evaluated thoroughly.
- ▶ Single modality imaging may not be sufficient to identify the cause of sudden OHCA.
- ▶ These patients, per published guidelines, should all receive an implantable cardioverter defibrillator prior to hospital dismissal and must have close follow-up at a centre with expertise in this condition.
- ▶ Failure to complete a proper work up and treatment may lead to catastrophic results for the patient and their family.

Contributors Each author participated in the planning, literature review, conducting, writing and reporting of the work described in this case report. MJA is the guarantor of the article and managed the case.

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