CASE REPORT

Aborted sudden cardiac death (SCD) in a patient with hypertrophic cardiomyopathy (HCM) with low-risk factors for SCD

Sashank Kaushik,¹ Stalin Ramakrishnan Subramanian,¹ Shahrokh Rafii,² Ricardo Castillo²

¹Department of Internal Medicine, Brookdale University Hospital and Medical Center, Brooklyn, New York, USA

²Department of Cardiovascular Medicine, Brookdale University Hospital and Medical Center, Brooklyn, New York, USA

Correspondence to
Dr Sashank Kaushik,
dr.skaushik@gmail.com

SUMMARY

Remarkable advancements have been made in understanding the pathophysiology of hypertrophic cardiomyopathy (HCM), since the first implantable cardioverter defibrillator (ICD) was placed in a human, more than 25 years ago to prevent sudden cardiac death (SCD). ICD has become the cornerstone in the management of HCM, with an ability to change the natural course of this complex disease. American College of Cardiology/American Heart Association guidelines have been charted out to help risk stratify asymptomatic or minimally symptomatic adults with HCM, in order to prevent SCD in such individuals. Our patient with left ventricular outflow tract gradient <30 mm Hg, left ventricular wall thickness < 30 mm, negative medical history of syncope and no family history of SCD, would have been at low risk for SCD.

BACKGROUND

Although there is a universal agreement that hypertrophic cardiomyopathy (HCM) patients are to be offered with secondary prevention after cardiac arrest or a documented ventricular tachycardia (VT) or ventricular fibrillation (VF), there are no clear-cut guidelines for implantable cardioverter defibrillator (ICD) placement as a primary prevention for HCM patients. Here, we present a patient with HCM who had no conventional risk factors, yet suffered a cardiac arrest and was successfully resuscitated.

CASE PRESENTATION

A 40-year-old woman was brought to the emergency department following a collapse while cooking. She was found pulseless by her cousin (who also happened to be a cardiac critical care nurse) and cardiopulmonary resuscitation was performed for approximately 2 min before emergency medical services (EMS) arrived. Initial rhythm was VF and she was shocked once with 200 J with successful conversion to sinus tachycardia. She was intubated in the field by EMS, but was combative after the shock and self-extubated, before reaching the hospital. On arrival, she was haemodynamically stable, maintained her respiration and remained in sinus rhythm. Medical history, family history was non-contributory and negative for sudden cardiac death (SCD). A review of the systems was negative. She had never undergone an echocardiogram, cardiac stress testing or Holter monitoring prior to this

event, which would have identified HCM and/or risk factors for SCD. Laboratory tests revealed elevated cardiac enzyme levels.

INVESTIGATIONS

ECG demonstrated sinus rhythm with left atrial (LA) enlargement and left ventricular (LV) hypertrophy. Chest radiography revealed pulmonary congestion. Transthoracic echocardiography showed asymmetrical septal hypertrophy, LA enlargement (4.2 cm) and systolic anterior motion of mitral valve without leaflet septal contact or significant left ventricular outflow tract (LVOT) obstruction. Pulmonary artery pressure was 48 mm Hg and ejection fraction was normal. Cardiac catheterisation showed no significant coronary artery disease, but has showed significant diastolic dysfunction. There was HCM with mid-cavitary obstruction with an average intracavity gradient of 27 mm Hg. Brockenhough-Braunwald-Morrow sign¹ as well as the spike and dome configuration of the postobstructive systolic pressures were noted (figure 1A,B). There was no significant LVOT gradient and maximum LV wall thickness was 2.36 cm.

DIFFERENTIAL DIAGNOSIS

Athlete's heart—LV hypertrophy associated with athletic training.

TREATMENT

She had a dual-chamber ICD placed for secondary prevention and was discharged on medical therapy with β -blockers.

OUTCOME AND FOLLOW-UP

After 16 months of follow-up with complete physical examination and periodical ICD interrogation, she is healthy and has not received any therapy from the ICD.

DISCUSSION

HCM is a particularly heterogeneous and unpredictable disease with respect to its clinical expression and natural history. SCD continues to be the most devastating complication of HCM. However, there were virtually no effective strategies for SCD prevention until recently, when HCM entered the ICD era, creating an enhanced focus on risk stratification and reliable identification of high-risk patients.²

To cite: Kaushik S, Subramanian SR, Rafii S, et al. *BMJ Case Reports* Published online: [please include Day Month Year] doi:10.1136/bcr-2012-006459

Figure 1 (A) Decrease in arterial pressure after a premature ventricular contraction (PVC) due to increased dynamic LV outflow tract obstruction. (B) Increased left ventricular pressure post-PVC from 145 to 190 mm Hg due to dynamic LV outflow tract obstruction in turn due to post-extrasystolic potentiation.

Risk factors for SCD in HCM have been derived from multiple observational studies and registries. A consensus document on HCM from the American College of Cardiology Foundation and American Heart Association, categorised known risk factors for SCD as ‘major’ and ‘possible’ in individual patients.^{3–6} The major risk factors include prior cardiac arrest, spontaneous sustained VT, spontaneous or exercise-induced non-sustained VT, family history of SCD, unexplained syncope, LV thickness ≥ 30 mm, and an abnormal blood pressure response to exercise. This consensus document also noted *possible risk factors*, which include LVOT obstruction (resting gradient >30 mm Hg), high-risk mutations, LV apical aneurysm and late gadolinium enhancement (LGE) on cardiovascular MRI (CMRI). The severity of other symptoms, such as dyspnoea, chest pain and effort intolerance have not been correlated with an increased risk of SCD. In HCM patients, with no discernible risk factors, SCD occurs at a rate of just less than 1% per year. Multiple risk factors in a patient would intuitively suggest a greater risk for SCD, however, the vast majority of patients with ≥ 1 risk factor will not experience SCD and simple addition of the risk factors would be imprecise, because of uncertainty of the relative weight of each individual risk factor. Having said that, even the presence of a single risk factor may be enough to warrant an ICD implantation based on patient’s age, strength of the risk factor and the risk-benefit of lifelong ICD therapy. Patients with >3 risk factors appear to have event rates that approach 5% per year, and survivors of prior SCD have a rate of subsequent SCD of around 10% per year.

One modifiable SCD risk factor is participation in intense activity sports and patients should not undergo ICD implantation in order to allow them to participate in such sports.

Although there have been vast advances in molecular genetics, HCM represents an exceedingly complex biological entity with significant intragenic heterogeneity involving >1400 mutations in at least 8 genes. Majority of mutations seem to effect proteins within or associated with the sarcomere. While in the past, some HCM genes and variants were considered to imply more severe disease consequences than other, there is, now a general consensus that routine screening for specific mutations cannot be designated either ‘benign’ or ‘malignant’ or reliably predict clinical outcomes.^{2,7}

In vivo detection of LV myocardial fibrosis by LGE on CMRI is hypothesised to represent an underlying electrical substrate,

which can cause ventricular tachyarrhythmias. This has created a great interest in this field and LGE on CMRI is now considered as a potential SCD risk modifier.^{2,3,8,9}

Learning points

- ▶ Common first presentations of hypertrophic cardiomyopathy (HCM) are sudden cardiac death (SCD), heart failure, atrial fibrillation with thromboembolic event or stroke.
- ▶ Clinically, HCM is diagnosed with left ventricular (LV) wall thickness ≥ 15 mm with nondilated ventricular chambers in the absence of another cardiac or systemic disease as aetiology of the LV hypertrophy.
- ▶ All patients with HCM should undergo comprehensive SCD risk stratification.
- ▶ The major risk factors include prior cardiac arrest, spontaneous sustained ventricular tachycardia (VT), spontaneous nonsustained VT, family history of SCD, syncope, LV thickness ≥ 30 mm and an abnormal blood pressure response to exercise.
- ▶ The possible risk factors are LV outflow obstruction, double or compounding (>1) mutations, late gadolinium enhancement on cardiovascular MRI, LV apical aneurysm.
- ▶ Any patient with HCM identified, to have one or more major risk factors should be carefully evaluated for implantable cardioverter defibrillator (ICD) implantation for primary prevention of SCD.
- ▶ All HCM patients are to be offered ICD as a secondary prevention after cardiac arrest or a documented ventricular fibrillation or a haemodynamically significant VT.
- ▶ The majority of asymptomatic patients with HCM have a relatively benign course. However, risk stratification needs to be done in each patient diagnosed with HCM.
- ▶ Patients not considered candidates for ICD, should have periodic (12–24 months) SCD risk stratification using ECG, transthoracic echocardiography, Holter monitors, exercise testing and clinical assessments.
- ▶ Screening of first-degree relative of patients with HCM, familial inheritance and genetic counselling is recommended. Genetic testing is recommended in first-degree relatives of index patients, who have been identified to carry a high-risk mutation.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Brockenbrough EC, Braunwald E, Morrow AG. A hemodynamic technic for the detection of hypertrophic subaortic stenosis. *Circulation* 1961;23:189–94.
- 2 Maron BJ. Contemporary insights and strategies for risk stratification and prevention of sudden death in hypertrophic cardiomyopathy. *Circulation* 2010;121:445–56.
- 3 Gersh BJ, Maron BJ, Bonow Ro, et al. 2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy: executive summary A report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines Developed in Collaboration with the American Association for Thoracic Surgery, American Society of Echocardiography, American Society of Nuclear Cardiology, Heart Failure Society of America, Heart Rhythm Society, Society for Cardiovascular Angiography and Interventions, and Society of Thoracic Surgeons. *J Thorac Cardiovasc. Surg.* 2011;142:1303–e1338.
- 4 Ikeda H, Maki S, Yoshida N, et al. Predictors of death from congestive heart failure in hypertrophic cardiomyopathy. *Am J Cardiol* 1999;83:1280–3.

- 5 Maki S, Ikeda H, Muro A, *et al*. Predictors of sudden cardiac death in hypertrophic cardiomyopathy. *Am J Cardiol* 1998;82:774–8.
- 6 Takagi E, Yamakado T, Nakano T. Prognosis of completely asymptomatic adult patients with hypertrophic cardiomyopathy. *J Am Coll Cardiol* 1999;33:206–11.
- 7 Maron JB, Maron MS, Semsarian C. Genetics of hypertrophic cardiomyopathy after 20 years: clinical perspectives. *J Amer Coll Card* 2012;60:705–15.
- 8 Rubinshtein R, Glockner JF, Ommen SR, *et al*. Characteristics and clinical significance of late gadolinium enhancement by contrast-enhanced magnetic resonance imaging in patients with hypertrophic cardiomyopathy. *Circ Heart Fail* 2010;3:51–8.
- 9 O'Hanlon R, Grasso A, Roughton M, *et al*. Prognostic significance of myocardial fibrosis in hypertrophic cardiomyopathy. *J Am Coll Cardiol* 2010;56:867–74.

Copyright 2013 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit

<http://group.bmj.com/group/rights-licensing/permissions>.

BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow