CASE REPORT

Implantable cardioverter defibrillators in the context of hypertrophic cardiomyopathy: a lesson in patient autonomy

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SUMMARY

Hypertrophiccardiomyopathy (HCM) is common, whereas the decision not to have an implantable cardioverterdefibrillator (ICD) when probably falling into a 'high-risk' category is not. A solicitor aged 45 years attended the inherited cardiac conditions clinic for review of her HCM and discussion about ICD implantation for primary prevention of sudden cardiac death (SCD). Despite a predicted 7% risk of SCD within the next 5 years, according to the European Society of Cardiology endorsed HCM Risk-SCD risk stratification tool, the patient opted against implantation of an ICD and comprehensively justifies her decision. This report discusses ethical aspects of a consultation offering ICD protection against SCD in the context of HCM and emphasises the clinicians' role in respecting patient autonomy.

BACKGROUND

Hypertrophic cardiomyopathy (HCM) is a common genetic condition which affects around 0.2% of adults.[1] Patients with HCM are at a higher risk of mortality from several causes including: stroke, heart failure and sudden cardiac death (SCD).[2] Irrespective of whether the patient is symptomatic or not, risk stratification for SCD should be performed for all patients with HCM. SCD tends to occur on average at around 43 years of age (±SD of 15 years). Early detection in the young and ICD intervention has markedly reduced the mortality rate of modern cohorts to <1%,[3 4] six times less than historical rates of up to 6%.[5] Recent guidelines from the European Society of Cardiology (ESC) advocates use of a risk calculator (HCM Risk-SCD) to work out an individual's risk of SCD in the next 5 years.[6] The risk calculator has been externally validated with mixed reviews regarding the accuracy of the estimated risk produced (reviewed and compared with comparable guidelines by Trivedi and Knight).[7] In particular, critics have called into question the sensitivity of the tool, in other words some patients categorised as low risk may in fact have a higher risk than the ESC risk stratification tool anticipates.[4] The predictive clinical variables used by the stratification tool include: age, maximum left ventricle wall thickness, left atrial size, maximum left ventricular outflow gradient, family history of SCD, non-sustained VT and unexplained syncope. However, this risk stratification is not perfect as there is mounting evidence to suggest that another risk factor: late gadolinium enhancement (LGE) on cardiac MRI is strongly associated with SCD, but is not included in the HCM Risk-SCD criteria.[8 9] The ESC guidelines define high risk as a ≥6% risk of SCD in the following 5 years, intermediate risk as ≥4% to <6%/5 years and low risk as <4%/5 years. It is advised that for a patient who falls into the high-risk category an 'ICD should be considered', for intermediate risk an 'ICD may be considered' and for low risk an 'ICD (is) generally not indicated'.[10] The ESC guideline for high-risk individuals is a class IIa recommendation, which is defined as where the 'weight of evidence/opinion is in favour of usefulness/efficacy'. Using these criteria, multiple studies have demonstrated that the majority (c. 78%) of patients are categorised as having a low risk, whereas the minority (c. 12%) are categorised as high risk according to the HCM Risk-SCD model.[4 11] These studies report that it is uncommon for patients within the high-risk category to opt against ICD implantation, accounting for between 13% and 35% of high-risk patients or 2% and 3% of patients with HCM.

CASE PRESENTATION

The patient is a fit and active female aged 45 years who works as a solicitor and was reviewed at the inherited cardiac conditions clinic, during which her risk of SCD due to her HCM was discussed. This entailed discussing the risk of SCD and the potential risks and benefits of undergoing implantation of an ICD. This issue had been discussed previously. Her HCM has been closely monitored since being diagnosed at the age of 31, following the diagnosis of her father. During this period, she has reported frequent self-limiting isolated palpitations, occurring up to once every 2 days and the odd infrequent run of a racing heartbeat. This can last for several minutes but does not make her feel presyncopal. She has been treated with 2.5 mg bisoprolol. Nevertheless, she exercises regularly and has not experienced any breathlessness or change in her exercise tolerance. However, she has on occasion experienced syncope on exertion and presyncope at rest in the past. She has a strong family history of HCM as both her father and uncle also have the condition. Indeed, her father had an ICD fitted and suffered a device infection and this may have influenced her decision not to have an

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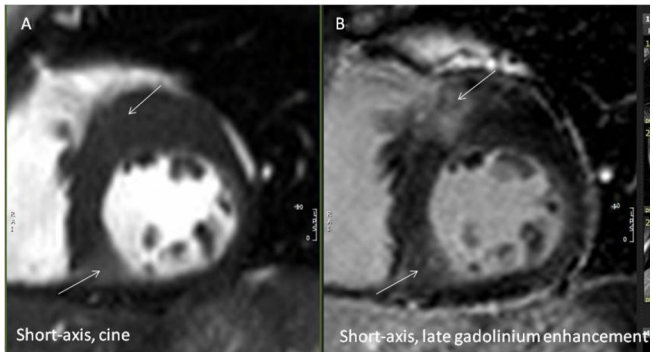

Figure 1 Mid-short axis views (A) cine and (B) corresponding late gadolinium enhancement (LGE) following contrast administration. There is patchy mid-wall LGE in the corresponding areas of pathological myocardial hypertrophy which is also in corresondence of both the inferior and superior insertion points of the right ventricle into the left ventricle (white arrows).

ICD. Yet none of her family member is known to have died from SCD.

On examination, she weighs 60 kg and has a height of 169 cm. On auscultation, there was a grade 2 ejection systolic murmur which is most prominent at the apex. The examination was otherwise normal.

INVESTIGATIONS

In order to estimate her risk of SCD various investigations were performed:

- Both the patient and her father have undergone genetic testing to understand the familial nature of their hypertrophic cardiomyopathy. She and her father were shown to possess two mutations: a recognised pathogenic variation in the MYBPC3 gene and a variant of uncertain significance in MYH7 gene.
- An ECG showed sinus rhythm with left axis deviation, inverted T waves in V1-V3 characteristic of HCM.
- As part of the review process in order to help estimate the risk of dangerous cardiac rhythms, regular Holter monitoring was undertaken. This has demonstrated both supraventricular and ventricular tachycardia.
- An exercise ECG stress test demonstrated a normal blood pressure response following 12 min of The Bruce protocol and showed no ECG changes or rhythm disturbances.
- A transthoracic echo demonstrated severe septal hypertrophy of 27 mm, with associated hypertrophy of the inferior and anterior walls. There was good left ventricular function and no evidence of systolic anterior motion or left ventricular outflow tract obstruction.
- A cardiac MRI scan demonstrated significant LGE (figure 1).

TREATMENT

In addition to taking 2.5 mg bisoprolol to control palpitations, she is monitored every 2 years to assess for ongoing symptoms and to reassess her risk of SCD.

OUTCOME AND FOLLOW-UP

She remains virtually asymptomatic and is not distressed by her infrequent, brief palpitations. To date, she has continues to opt out of having an ICD, and has done for over a year.

DISCUSSION

The patient's decision to opt against ICD implantation will be discussed in the context of both the indications for an ICD and also the potential negative complications, which can arise from ICD implantation. It will be shown that informed patient choice should lead decision making, and that this is justified by recognising patient autonomy: one of the four pillars of principalism.[12] Our patient has capacity and is able to decide on the basis of her own thoughts, and so is autonomous.

Indeed, the patient's understanding of the concept of risk is fundamental to this case. Ineffective presentation of medical statistics can lead to uninformed patients making bad decisions. The probability produced by the HCM Risk-SCD calculator depends on the extent of an individual's risk factors, and so is a conditional probability. There is evidence to suggest that natural frequencies are easier to understand than probability.[13] There are many other tools which can be used to help patients understand risks. These include avoiding only using descriptive terms (such as 'high risk'), using visual aids (such as pie charts) and avoiding numbers (such as not using relative risk).[14] If there is any doubt that the patient does not understand, there is always another way of communicating risk.

Reasons for having an ICD

When the ESC-approved criteria are entered into the HCM Risk-SCD calculator this produces an estimated risk of 7.31%/5 years, for which the ESC recommendation is that an 'ICD should be considered'.[10] This risk may in fact be higher given the patchy LGE found on cardiac MRI shown in figure 1.[8 9] An ICD is an effective intervention in preventing SCD and has therefore been used in primary prevention, and therefore could be argued to be in the patient's best interests.[15] Interestingly, our patient referred to hoping that her doctor would tell her that she needed an ICD if it were essential. Although it is impossible to know if an ICD is essential, this helps justify why in this case ICD implantation was thoroughly encouraged. One of our patient's main concerns was a decrease in her quality of life as a result of ICD implantation. However, in representative samples having an ICD has been associated with only a short-term decrease in quality of life,[16] and even an increase after 6 months.[17] It is important to inform the patient of the risk of inappropriate shocks (4.6%/year) and implant-related complications (5.1%/year). Despite these device complications being unusual, our patient was concerned by these risks as her father had experienced a pocket infection in the past, and this likely added to her reservations.[18] ICDs prevent out-of-hospital cardiac arrests which have a poor survival rate,[19] and are also expensive in the long term.[18] Ethically, it is also important that respect for autonomy does not jeopardise delivery of a publicly funded health system with finite resources as the cost-effectiveness of ICDs has been demonstrated.[20–23]

Reasons against having an ICD: an individual patient's decision

However, there is an increasing appreciation that the benefit of having an ICD must be balanced with psychosocial distress related to ICD implantation. At the time of implantation, the prevalence of anxiety is thought to vary from 13% to 46%, and similarly the prevalence of depression has been documented as varying from 24% to 46%.[24] Anxiety related to sexual function is common and can be overlooked despite posing a threat to psychosocial well-being and relationships.[25] The incidence of post-traumatic stress disorder (PTSD) in patients who have received ICDs has been reported at around 26% at over 3 months of treatment,

Patient's perspective

At my latest check-up when my consultant mentioned that he thought I should consider the possibility of having a defibrillator fitted I was shocked and completely unprepared for the discussion that ensued. I felt very much the same way I did when I was first diagnosed with hypertrophic cardiomyopathy about 15 years ago. In that instance, I was just being screened as my dad had recently been diagnosed, having been very unwell for some time. Although I had a 50:50 chance of inheriting the condition, as I was fit and healthy, I do not think I had really given the matter much thought. My dad has had a defibrillator for the last 8 years or so and before that a pace maker. He however has also had period of very poor health as a result of his condition. In theory, I have accepted that I too will probably have a defibrillator or pacemaker too at some stage, but in my mind it has always been something that would happen when I am probably in retirement—like my dad.

The issue for me is that far as I am concerned I am perfectly healthy. I lead an active life and apart from the occasional inconvenience of having to attend annual check-ups and to take a beta-blocker once a day, my 'condition' does not affect my life. I do not have to think about it on a day-to-day basis, and most of the time I do not. It has not stopped me doing anything that I want to do (apart from giving blood!). For this reason, it is very difficult to even begin to consider having what in my mind is an intrusive and also currently in my view, an unnecessary procedure. This is particularly as I know that it is likely to affect my mental health far more than my heart condition has ever affected my physical health to date.

After putting a few pieces of information about my condition into an app, my consultant suggested that I had a 5% chance of having a fatal heart rhythm in the next 5 years. I do not consider myself to be a risk taker, but in any event in my view, 5% is a pretty low risk. That was my initial, I guess, emotional reaction to the suggestion, without giving it much real thought. Having now had time to think, my view has not changed at all. In making this decision, I take into account:

- There is no history in my family of anyone dropping dead in their prime;
- Both my uncle and dad, who both have the same condition, are in their 70s, and have both lived very active lives, are still alive and in fairly good health;
- My uncle ran the London marathon in his 40s unaware of this heart condition;
- Both my dad and my uncle were not diagnosed until they retired. I am 45 and have had the benefit of about 15 years of excellent medical care which I hope means that my long-term prognosis could be even better than theirs;
- Once I have the defibrillator there is no going back and I know it will have a negative emotional impact on me and finally
- The way it will make me look.

I cannot deny that part of my decision boils down to vanity, but it is more than that. Up until now, I have been able to say that I have a heart condition, but I am fine, I only know I have it because I was screened, it has never affected my life. As soon as I have a defibrillator this will change. It will be something to be explained and suddenly it is an issue. While among friends this would not be a problem for me, I have particular concerns about how it might affect my children. They of course both have a 50:50 chance of inheriting this condition from me (something that I struggle with sometimes). I have been told that it seems likely my son, a very sporty and talented 9 year, probably has inherited this condition. At the moment, I have downplayed things to them both. Explaining we are lucky to receive these extra check-ups and mum is fine. It will be hard however to explain away mum having a device attached to her heart, and they are both bright kids who will then work out this could be what happens to them. My son will probably never be a premier league footballer, but I do not want to shatter that dream for him right now. I know how I feel when I look at my dad, but I can deal with this because it is something that is going to happen (in my head at least) when I am much older, when perhaps it will matter less to me (although over time I am realising it will probably always matter, a lot).

As my consultant suggested, I have discussed my decision with my husband and parents and at this stage they agree with my decision, and accept it is mine to make. I know this discussion will be ongoing but I am prepared to hold my ground until I am presented with a higher risk than I have currently been presented with.

I also know that I am not my dad or my uncle, that my mother's genes could mean that my prognosis is not the same as theirs, but I guess on this point I am prepared to take a risk, so maybe I am a risk taker after all.

substantially higher than the general population. Preimplantation anxiety and ICD concerns are intricately linked with PTSD, and have been shown to be the best predictors of PTSD development.[26] Therefore, the patients who are more reluctant to have an ICD are at higher risk of PTSD, and so pressuring these individuals would not be to act with beneficence. As one may expect, the strongest determinant of PTSD development was previous experience of ICD shock.[26] Experiencing multiple shocks, or even a repeated shock episode known as an ICD storm, can be particularly damaging to a patients' psychosocial well-being. The importance of accounting for a patient's psychological well-being is also highlighted by evidence to suggest that elevated PTSD scores increase mortality risk.[27 28]

There are several reasons against ICD implantation in this case. Our patient felt that as her condition was not affecting her life at the current moment, implantation would act as a physical reminder that she had HCM. She was also concerned about how an ICD may affect her 'mental health', which as previously mentioned puts her into a higher risk group for PTSD.[29] In addition, young patients are often concerned about how their device may interfere with socialisation and sexual function.[30] There is further evidence to suggest that women are particularly at risk of psychological distress following ICD implantation.[31] She also made an interesting point that as a mother she is concerned about how having an ICD may affect her children who have a chance of having inherited the condition themselves. As the patient expressed in her patient's perspective, she feels strongly that this is '(her) decision' to make. Nevertheless, she is willing to reconsider an ICD, should her health status change. These risks must be taken into account as currently the patient is almost asymptomatic and is relatively not concerned about the risk of SCD, therefore these risks pose a threat to her quality of life.

A qualitative semi-structured interview-based study of several individuals opting out of ICD implantation has many parallels

with this case.[32] It is common for patients who opt out of ICD intervention to focus on their health status at the present rather than their potential future health status. Another thought shared by this study and our case was that of taking responsibility to make an independent decision given the information that the physician provides. Above all, the most emotive feeling across this group and this case is the autonomous idea that it is the physicians' job to protect their patients, but it is the individuals' choice to make. Moreover, legally there is an emphasis on the importance of patient autonomy. This is particularly apparent following the Montgomery case, which introduced the legal test of '*materiality*' for obtaining patient consent.[33 34] Although there is a cost to this autonomy as there is substantial evidence showing that leaving patients without ICD protection is more expensive,[20 22] perhaps this is the price for optimising patient autonomy.

Learning points

- Maintaining patient autonomy is a crucial part of any consultation. This is especially true in the context of whether to have an ICD because of the potential for extensive psychological harm.
- Risk is only an estimate and the true probability of an outcome can be higher or lower than anticipated. Therefore, risk should not be considered as all-important and should be balanced with patient's wishes.
- The physician may be surprised by a patient's decision, but he/she cannot let this influence their important role in delivering objective information.

Contributors JJHB is responsible for writing the manuscript, obtaining consent and reviewing the literature surrounding the case. CBD provided help with figure acquisition. GS is the Consultant Cardiologist who consulted the patient about the decision of whether to have an ICD or not, was responsible for conceptualising the case report and thoroughly reviewed the introductory and background literature. All authors edited and reviewed the manuscript, and have approved the final version.

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REFERENCES

1 Maron BJ, Gardin JM, Flack JM, *et al*. Prevalence of hypertrophic cardiomyopathy in a general population of young adults : echocardiographic analysis of 4111 subjects in the cardia study. *Circulation* 1995;92:785–9.
2 Maron BJ, Olivotto I, Spirito P, *et al*. Epidemiology of hypertrophic cardiomyopathy-related death: revisited in a large non-referral-based patient population. *Circulation* 2000;102:858–64.
3 Elliott PM, Gimeno JR, Thaman R, *et al*. Historical trends in reported survival rates in patients with hypertrophic cardiomyopathy. *Heart* 2006;92:785–91.
4 Maron BJ, Casey SA, Chan RH, *et al*. Independent assessment of the European Society of cardiology sudden death risk model for hypertrophic cardiomyopathy. *Am J Cardiol* 2015;116:757–64.
5 Hardarson T, De la Calzada CS, Curiel R, *et al*. Prognosis and mortality of hypertrophic obstructive cardiomyopathy. *Lancet* 1973;2:1462–7.
6 O'Mahony C, Jichi F, Pavlou M, *et al*. A novel clinical risk prediction model for sudden cardiac death in hypertrophic cardiomyopathy (HCM risk-SCD). *Eur Heart J* 2014;35:2010–20.
7 Trivedi A, Knight BP. ICD therapy for primary prevention in hypertrophic cardiomyopathy. *Arrhythm Electrophysiol Rev* 2016;5:188–96.
8 Chan RH, Maron BJ, Olivotto I, *et al*. Prognostic value of quantitative contrast-enhanced cardiovascular magnetic resonance for the evaluation of sudden death risk in patients with hypertrophic cardiomyopathy. *Circulation* 2014;130:484–95.
9 Weng Z, Yao J, Chan RH, *et al*. Prognostic value of LGE-CMR in HCM: a meta-analysis. *JACC Cardiovasc Imaging* 2016;9:1392–402.
10 Elliott PM, Anastasakis A, Borger MA, *et al*. 2014 ESC guidelines on diagnosis and management of hypertrophic cardiomyopathy: the Task Force for the Diagnosis and Management of Hypertrophic Cardiomyopathy of the European Society of Cardiology (ESC). *Eur Heart J* 2014;35:2733–79.
11 Fernández A, Quiroga A, Ochoa JP, *et al*. Validation of the 2014 European Society of Cardiology sudden cardiac death risk prediction model in hypertrophic cardiomyopathy in a reference center in South America. *Am J Cardiol* 2016;118:121–6.
12 Beauchamp T, Childress J. *Principles of biomedical ethics*. 7th edn. Oxford: Oxford University Press, 2012.
13 Hoffrage U, Gigerenzer G. Using natural frequencies to improve diagnostic inferences. *Acad Med* 1998;73:538–40.
14 Paling J. Strategies to help patients understand risks. *BMJ* 2003;327:745–8.
15 Maron BJ, Shen WK, Link MS, *et al*. Efficacy of implantable cardioverter-defibrillators for the prevention of sudden death in patients with hypertrophic cardiomyopathy. *N Engl J Med* 2000;342:365–73.
16 May CD, Smith PR, Murdock CJ, *et al*. The impact of the implantable cardioverter defibrillator on quality-of-life. *Pacing Clin Electrophysiol* 1995;18:1411–8.
17 Pedersen SS, Mastenbroek MH, Carter N, *et al*. A comparison of the quality of life of patients with an entirely subcutaneous implantable defibrillator system versus a transvenous system (from the EFFORTLESS S-ICD Quality of Life Substudy). *Am J Cardiol* 2016;118:520–6.
18 O'Mahony C, Lambiase PD, Quarta G, *et al*. The long-term survival and the risks and benefits of implantable cardioverter defibrillators in patients with hypertrophic cardiomyopathy. *Heart* 2012;98:116–25.
19 Horsted TI, Rasmussen LS, Meyhoff CS, *et al*. Long-term prognosis after out-of-hospital cardiac arrest. *Resuscitation* 2007;72:214–8.
20 Geri G, Fahrenbruch C, Meischke H, *et al*. Effects of bystander CPR following out-of-hospital cardiac arrest on hospital costs and long-term survival. *Resuscitation* 2017;115:129–34.
21 Cassel CK, Guest JA. Choosing wisely: helping physicians and patients make smart decisions about their care. *JAMA* 2012;307:1801–2.
22 Sanders GD, Hlatky MA, Owens DK. Cost-effectiveness of implantable cardioverter-defibrillators. *N Engl J Med* 2005;353:1471–80.
23 Campbell AGG, Jones G. *205 'Justice and health care' medical ethics*. 4th edn. Oxford: Oxford Univeristy Press:254–67.
24 Sears SF, Matchett M, Conti JB. Effective management of ICD patient psychosocial issues and patient critical events. *J Cardiovasc Electrophysiol* 2009;20:1297–304.
25 Vazquez LD, Sears SF, Shea JB, *et al*. Sexual health for patients with an implantable cardioverter defibrillator. *Circulation* 2010;122:e465–67.
26 Versteeg H, Theuns DA, Erdman RA, *et al*. Posttraumatic stress in implantable cardioverter defibrillator patients: the role of pre-implantation distress and shocks. *Int J Cardiol* 2011;146:438–9.
27 Ladwig KH, Baumert J, Marten-Mittag B, *et al*. Posttraumatic stress symptoms and predicted mortality in patients with implantable cardioverter-defibrillators: results from the prospective living with an implanted cardioverter-defibrillator study. *Arch Gen Psychiatry* 2008;65:1324–30.
28 Kikkenborg Berg S, Caspar Thygesen L, Hastrup Svendsen J, *et al*. Anxiety predicts mortality in ICD patients: results from the cross-sectional national CopenHeartICD survey with register follow-up. *Pacing Clin Electrophysiol* 2014;37:1641–50.
29 Sears SF, Todaro JF, Lewis TS, *et al*. Examining the psychosocial impact of implantable cardioverter defibrillators: a literature review. *Clin Cardiol* 1999;22:481–9.
30 Dubin AM, Batsford WP, Lewis RJ, *et al*. Quality-of-life in patients receiving implantable cardioverter defibrillators at or before age 40. *Pacing Clin Electrophysiol* 1996;19:1555–9.
31 Vazquez LD, Conti JB, Sears SF. Female-specific education, management, and lifestyle enhancement for implantable cardioverter defibrillator patients: the FEMALE-ICD study. *Pacing Clin Electrophysiol* 2010;33:1131–40.
32 Ottenberg AL, Mueller PS, Topazian RJ, *et al*. "It's not broke, so let's not try to fix it": why patients decline a cardiovascular implantable electronic device. *Pacing Clin Electrophysiol* 2014;37:1306–14.
33 Montgomery v Lanarkshire Health Board (Scotland). *Hearing before the UK Supreme Court*, 2015.
34 General Medical Council. Making decisions together: the implications of the montgomer judgement. https://gmcuk.wordpress.com/2015/05/29/making-decisions-together-the-implications-of-the-montgomery-judgment/ (accessed 16 Jan 2018).