Research Paper

Journal of the Royal Society of Medicine Cardiovascular Disease
6: 1–9

DOI: 10.1177/2048004017698614
journals.sagepub.com/home/cvd

"There are days I wish it wasn't there, and there's days I realize I'm lucky": A qualitative study of psychological sequelae to the implantable cardioverter defibrillator as a treatment for the prevention of sudden cardiac death in arrhythmogenic right ventricular cardiomyopathy

Holly Etchegary[1], Daryl Pullman[2], Sean P Connors[3], Charlene Simmonds[2], Terry-Lynn Young[4] and Kathy A Hodgkinson[1]

Abstract

Objectives: Arrhythmogenic right ventricular cardiomyopathy caused by a TMEM43 p.S358L mutation is a fully penetrant autosomal dominant cause of sudden cardiac death where prophylactic implantable cardioverter defibrillator therapy significantly reduces mortality by returning lethal cardiac rhythms to normal. This qualitative study assessed the psychological ramifications of the implantable cardioverter defibrillator on recipients, their spouses and their mutation negative siblings.

Design: Qualitative interview study.

Participants: Twenty-one individuals (nine mutation positive, eight mutation negative and four spouses) from 15 families completed semi-structured interviews.

Results: No theoretical assumptions about the data were made: inductive sub-coding was accomplished with the constant comparison method and cohesive themes across all respondent interviews were determined. All interviewees had a family history of sudden cardiac death and appropriate implantable cardioverter defibrillator therapy in themselves or family members. Average length of time with an implantable cardioverter defibrillator was 10 years. Major themes included: (1) acceptance and gratitude, (2) grudging acceptance, (3) psychological effects (on emotional and psychological well-being; functioning of the broader family unit; and relationships), and (4) practical concerns (on clothes, travel, loss of driving licence and the effects of an implantable cardioverter defibrillator discharge). These affected all family members, regardless of mutation status.

Conclusions: Despite the survival advantage of implantable cardioverter defibrillator therapy, the intervention carries psychological and practical burdens for family members from kindreds manifesting p.S358L TMEM43 ARVC that does not appear to dissipate with time. A move towards integrating psychology services with the cardiac genetics clinic for the extended family may provide benefit.

[1]Faculty of Medicine, Genetics and Clinical Epidemiology, Memorial University, Canada
[2]Faculty of Medicine, Community Health and Humanities, Memorial University, Canada
[3]Faculty of Medicine, Cardiology, Memorial University, Canada
[4]Faculty of Medicine, Genetics, Memorial University, Canada

Corresponding author:
Holly Etchegary, Faculty of Medicine, Clinical Epidemiology, Craig Dobbin Centre for Genetics, Memorial University, Room 4M210, St. John's A1B 3V6, NL, Canada.
Email: holly.etchegary@med.mun.ca

Keywords
Arrhythmias, sudden cardiac death, cardiomyopathy, implantable cardiac defibrillator, ARVC, psychosocial

Date received: 8 November 2016; revised: 6 January 2017; accepted: 7 February 2017

Introduction

Inherited cardiomyopathies pose psychosocial burdens for affected individuals and their families.[1–4] At-risk individuals face a range of clinical and psychosocial issues, including early diagnosis, risks to family members, restriction of physical or employment activities, lifelong clinical surveillance, invasive treatment modalities, and the potential threat of sudden cardiac death (SCD) or heart failure.[2,5]

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a clinically difficult to diagnose[6] single-gene disorder which can cause SCD due to ventricular arrhythmias and/or heart failure. For those at high risk of ARVC-related SCD, the implantable cardioverter defibrillator (ICD) is an effective therapy,[4,7] which cardioverts a potentially lethal rhythm back to sinus rhythm. These ICD discharges may occur with the patient conscious or following loss of consciousness and result in the revocation of a driving licence for a period of six months.[8] In Newfoundland and Labrador (NL), Canada, (also Denmark and Germany[9]) ARVC may be caused by a founder mutation in *TMEM43* (p.S358L).[10] This is a sex-influenced genetic ARVC subtype, with a median age to death of 41 years in males compared to 71 years in females.[10] This ARVC subtype is fully penetrant over the life span and is autosomal dominant (children of an affected parent are at 50% risk of inheriting the mutation). In the NL *TMEM43* p.S358L population, ICD therapy has significantly improved the survival of affected individuals and is an essential prophylactic (diagnosis based on positive mutation status alone) first-line treatment for males.[11]

A small number of qualitative studies with ARVC families segregating the *TMEM43* p.S358L mutation[12] and other cardiomyopathies[13–15] highlight general psychosocial burdens for at-risk families. Experiential knowledge of loss and death was clear,[12] while interviews with individuals tested for HCM or Long QT syndrome revealed parents' concern about their children's futures.[13,14] Relatives of young SCD victims in the Netherlands described ongoing worry and a desire to prevent a subsequent fatal event in remaining children and grandchildren.[15]

While the ICD may be life-saving, it comes with its own psychosocial considerations. Psychological and quality of life (QOL) implications of ICD therapy have been addressed in older patients with heart disease who generally cope well with an ICD, although a subgroup will experience psychological difficulties, including post traumatic stress.[16–18] Younger age at implant (<50 years) and multiple ICD shocks were associated with greater distress.[18] Young patients manifesting genetic heart diseases (including ARVC), revealed increased levels of depression, anxiety and posttraumatic stress, and decreased scores on QOL measures in the short term[1,3,19,20] with participants reporting that their life was not 'normal' and for whom up to a third had lost, or changed jobs following an ICD.[19] In ARVC families, ICD recipients are often young; they face many years with their ICD, exposing them to repeated procedures, discharges and life-long psychosocial burdens,[3,4] including body image issues.[3,21]

This study details a qualitative analysis of selected family members from the homogeneous ARVC *TMEM43* p.S358L population to capture the experiences of mutation positive individuals, their mutation negative siblings and unrelated spouses through the use of semi-structured interviews. These findings will contribute to evidence-based psychosocial management of families with highly penetrant malignant mutations where an effective SCD prevention treatment regimen exists.

Methods

Sampling and recruitment

The local Health Research Ethics Board approved the project (HREB 12.053). Participants were recruited from the provincial cardiac genetics clinic, which provides care to families affected by heritable cardiac disease causing SCD (currently ~910 individuals). Referred patients receive genetic counseling, cardiac testing, molecular genetic analysis and follow-up clinical management, as do affected family members following cascade screening. For patients with mental health sequalea, referral to psychiatric services occurs.

Clinical, family and molecular data on families are stored in a computer database. Mutation-specific data sets for autosomal dominant cardiac conditions include all persons born at a 50% *a priori* risk in families. For this study, the mutation-specific dataset for individuals from families affected by ARVC caused by *TMEM43* p.S358L was utilised.

We accessed the first 15 (of 26) families.[10] Families recently diagnosed were excluded as we did not want to cause additional distress upon diagnosis and less time had passed to observe and experience potential impacts of ICD therapy. From these, 292 individuals were eligible for this study (alive, between the ages of 15 and 70 years and living in Newfoundland). The focus was the psychosocial burden associated with ICD therapy on all family members. One author (KAH) identified individuals through purposive sampling to represent a broad range of experience with ARVC (mutation positive, mutation negative, and spouses/partners).[22] Five to six individuals from each group were randomly chosen and invited to participate. Purposive sampling is a hallmark of qualitative research to ensure a broad representation of participant experience and to avoid the overrepresentation of any one group's views[23–25] (e.g. carriers). Study invitations were sent to 73 individuals who could contact the investigators. Clinical and demographic information was obtained from family pedigrees and the *TMEM43* database. The length of time with an ICD and the number of ICD discharges were captured: discharges were categorised as appropriate (the termination of a malignant rhythm) or inappropriate (e.g. a raised normal heart rate).

The interviews

In person interviews occurred with 21 individuals, with a minority ($n = 3$) by phone. A trained interviewer (CS) completed semi-structured interviews (questions were not ordered, and participants were encouraged to raise additional issues) outside of clinic appointments; these lasted about an hour, were recorded and transcribed verbatim. ICD recipients were asked directly about their experiences with the device. This was part of a larger sub-study on ARVC (due to *TMEM43* p.S358L) related psychosocial effects. Interviews covered family experiences of SCD, genetic testing decisions,[22] and the impact of ARVC on psychological well-being, behaviour, and economics.[5] Initial questions were drafted by HE and input from team members resulted in minor changes.

Data analysis

Qualitative description[23] was used to summarise the ICD therapy data. No philosophical or theoretical assumptions about the data were made. Data were presented in the language of participants. Transcripts were read several times by HE to identify emerging themes. When possible, interviews proceeded until data saturation was reached (the point at which no new ideas emerged). Interview data pertaining to the ICD were isolated and used to identify categories for the current analysis.

Two investigators (HE, KAH,) independently read the ICD data. Inductive sub coding was accomplished with the method of constant comparison[24,25] (data were compared between and within transcripts). Investigators independently identified categories and themes, and agreement on the basic ideas was high.

Results

Twenty-one individuals completed an interview (five males). Non-responders included those with heart transplants and those who declined genetic testing. Detailed clinical and demographic information is provided in Table 1. All participants had a history of SCD in their families. Of the 17 family participants, eight had an ICD for an average duration of 10.25 years. All ICDs were provided for prophylaxis. Life saving discharges occurred in 63% ($n = 5$) of ICD recipients, the remainder had relatives whose lives had been saved. Of the individuals negative for the p.S358L *TMEM43* mutation, 88% ($n = 7$) had experience of appropriate ICD therapy in relatives, as did 75% ($n = 3$) spouses. Mean age of participants was 46 years (SD = 15) and all participants (except the adolescents) were married with children. Quantitative data were not collected on mean incomes; however, the experience of investigators suggests that all participants are from middle-income families.

Thematic analysis

Respondents noted several psychosocial issues related to the ICD. Four major themes emerged: (1) acceptance and gratitude, (2) grudging acceptance, (3) psychological effects, and (4) practical concerns. Psychological and practical effects were not confined to ICD recipients, and the effects were not independent. Supportive quotations for each theme are presented. The unabridged data (all quotations) are given in Supplemental Table 2.

Acceptance and gratitude

Across all groups, it was clear that the survival benefit of the ICD was understood, particularly for families where young people had died, and for those who recognised that a loved one was alive because of their ICD. This was apparent from spousal accounts, with partners who would have died in the absence of their device.

The research has done so much…it has given people an opportunity…it has saved two of my relatives at least twice. (Negative female)

The ICD was the life saver…my husband otherwise would have been dead at 27. (Female spouse)

Table 1. Clinical and demographic characteristics of participants.

Sex	ID	Family ID[a]	Age[b]	Family history of SCD and ICDs	*TMEM43* p.S358L Mutation status	ICD	# ICD discharges	Years with ICD
F	P1	AR2	41	One 3° relative with SCD at 39 years (post ICD and HT at 29 years) One 1° relative: HT at 55 years (post ICD) Five 3° relatives with ICD's	Negative	No	N/A	N/A
F	P2	AR1	68	One 1° relative with SCD at 29 years Two 2° relatives SCD aged 35 and 69 years Five 3° relatives SCD at 23, 47, 44, 42 and 40 years One 1° relative ICD Three 3° relatives with ICD	Negative	No	N/A	N/A
F	P3	AR1	56	Two 1° relatives with SCD at 32 and 29 years Three 2° relatives with SCD aged 42, 44 and 40 years One 1° relative with ICD	Positive	Yes	None	14
F	P4	AR10	63	Two 1° relatives with SCD at 34 and 40 years One 1 relative with ICD at 27 years and HT at 34 years	Positive	Yes	7 appropriate 5 inappropriate	13
F	P5	AR8	60	One 1° relative with SCD at 40 years One 3° relative with SCD at 19 years One 1° relative with ICD One 2° relative with ICD Five 3° relatives with ICD	Positive	Yes	4 appropriate 2 inappropriate	14
F	P6	AR2	47	One 3° relative with SCD at 39 years One 1° relative with ICD Two 2° relatives with ICD	Positive	Yes	1 appropriate	7
F	P7	AR8	64	Two 1° relatives with SCD at 19 and 72 years Two 1° relatives with ICD Two 2° relatives with ICD Five 3° relatives with ICD	Positive	Yes	None	14
M	P8	AR6	48	One 1° relative SCD at 44 years One 1° relative with ICD One 2° relative with ICD One 3° relative with ICD	Negative	No	N/A	N/A
F	P9	AR2	47	One 3° relative SCD at 39 years One 1° relative with ICD Two 2° relatives with ICD	Negative	No	N/A	N/A
F	P10	AR12	27	Two 2° relatives with SCD at 30 and 47 years One 1° relative HT aged 40 years One 1° relative with ICD	Negative	No	N/A	N/A

(continued)

Table 1. Continued

Sex	ID	Family ID[a]	Age[b]	Family history of SCD and ICDs	*TMEM43* p.S358L Mutation status	ICD	# ICD discharges	Years with ICD
				One 2° relative with ICD One 3° relative with ICD				
F	P11	AR2	17	Two 1° relatives with ICD One 2° relative with ICD One 2° relative with HT	Positive	No	N/A	N/A
M	P12	AR2	20	One 1° relative with ICD One 2° relative with ICD One 2° relative with HT	Positive	Yes	1 inappropriate	3
M	P13	AR2	45	One 3° relative with SCD at 39 years (post ICD and HT at 29 years) One 1° relative: HT at 55 years (post ICD) Five 3° relatives with ICD	Negative	No	N/A	N/A
F	P14	AR1	65	Two 1° relative with SCD at 26 years and 35 years Five 3° relatives with SCD at 30, 44,42, 40 and 23 years One 3° relative with ICD	Negative	No	N/A	N/A
M	P15	AR2	48	One 2° relative HT at 55 years (post ICD) One 3° relative SCD 38 years One 1° relative ICD	Positive	Yes	8 appropriate 1 inappropriate	8
F	P16	AR2	61	One 1° relative SCD 30 years One 2° relative SCD 39 years One 3° relative SCD 27 years Five 2° relatives with ICD Two 3° relatives with ICD	Negative	No	N/A	N/A
M	P17	AR15	38	Two 2° relatives SCD at 35 and 47 years One 1° relative HT at 39 years Two 2° relatives with ICD One 3° relative with ICD	Positive	Yes	No discharges	9

Spouses of affected persons

Sex	ID	Family	Age	No. of ICD discharges in spouse	Extended family history of the Spouses' family
F	P18	AR2	48	Eight appropriate One inappropriate	Spouse (P15) with ICD One 2° relative HT at 55 years (post ICD) One 3° relative SCD 38 years One 1° relative ICD

(continued)

Table 1. Continued

Spouses of affected persons					
Sex	ID	Family	Age	No. of ICD discharges in spouse	Extended family history of the Spouses' family
F	P19	AR15	39	No discharges	Spouse (P17) with ICD One 1° relative HT at 39 years Two 2° relatives with ICDs One 3° relative with ICD
F	P20	AR11	43	Fifteen appropriate	Spouse with ICD for 14 years (did not interview) Two 2° relatives SCD 30 and 50 years Two 1° relatives with ICD
F	P21	AR15	38	One appropriate	Spouse with ICD for 13 years (did not interview) Two 2° relatives SCD 52 and 75 years Two 1° relatives with ICDs

HT: Heart transplant N/A not applicable; shaded = urban; non-shaded = rural.
[a]Pedigree structures are presented in a previous publication.[10]
[b]Age at interview.

Grudging acceptance

Despite the acknowledgment of the survival benefit of the ICD (it was commonly described as "a safety net," "an insurance policy," or "a security blanket"), accepting the ICD took time. The saved life of a sibling was the impetus required for one female to accept an ICD, but a sense of resignation remained.

You learn to live with it, but it took a long time to come to terms with it. (Positive female)

Psychological impact

These effects were related to: (a) emotional and psychological well-being; (b) functioning of the broader family unit; and (c) romantic and social relationships.

Emotional and psychological well-being

Mutation carriers described a variety of negative emotional impacts ranging from constant awareness of the ICD to the presence of recurring intrusive thoughts. These issues affected partners in a similar way. Mental health sequalae (anxiety, depression, and fear) were present in all respondent groups.

I ended up on an anti-depressant.... I can feel my pacemaker kicking in... you're constantly reminded. (Positive female)

I used to sit up at night and watch him breathe... it brought on severe anxiety and panic attacks, so I'm on medication for that. (Female spouse)

Functioning of the broader family unit

All accounts (particularly spouses) highlight a range of negative emotions such as fear and guilt (including survivor guilt). Psychological effects on children are illustrated by a spouse describing the effect on her young offspring, and from a negative adult female recalling 'scary' childhood experiences.

I knew I should have been happy [about testing negative], but I wasn't. (Negative Female)

I still feel guilty because I was given a second chance and [name] wasn't. (Positive female)

Relationships

Marriages were affected, ranging from dealing with anger, to how to talk to children about the family risk, and the nature of the romantic relationship. The resulting burden on the unaffected spouse was upsetting for some, while dating issues were problematic for younger respondents. Adolescents described disquiet over views held by friends and teachers, and spouses detailed the nervousness of friends following an ICD discharge or the secrecy that some participants felt the

need to maintain, leading to issues of isolation. Social relationships with friends and co-workers were affected.

His friends are a bit nervous around him. (Female spouse)

Teachers seem to be quite negative about this kind of stuff. (Positive male)

Practical concerns

Practical considerations ranged from clothing choices (reflecting the position of the scar and their ICD 'bump'), travel choices (the availability of appropriate medical facilities and cell phone reception), the ability to continue driving and social and recreational activities (continuing to play a loved sport). The loss of a driving license was the most difficult for those with an ICD, a loss particularly inconvenient for the many ICD recipients and their families living in rural areas of Newfoundland and Labrador (the majority of this cohort). Respondents noted how their families' (and their own) social lives were affected. Practical considerations included the physical effects of an ICD discharge and the stress of potential loss of consciousness.

. . .after my defibrillator went off, I didn't want to go anywhere. (Positive Male)

He won't go anywhere without cell phone reception. . ..(or). . .. unless he researches medical facilities. . .. (Female Spouse)

Discussion

This study found numerous psychosocial burdens associated with ICD therapy in families affected by ARVC caused by a homogeneous p.S358L mutation in *TMEM43*. Whether there are major differences between the heritable cardiomyopathies with a risk of SCD (e.g. ARVC, DCM and HCM) in the type and severity of psychological effects is not known. As the natural history varies between diseases (and between different mutations causing the same disease), the same might be true for psychological issues. This study population differs in several respects from previous studies on different heritable diseases or on genetically heterogeneous groups of ARVC individuals. Here the families are large; the disease-causing mutation is fully penetrant (and both necessary and sufficient to cause disease), untreated disease results in early SCD; extended family history documents multiple young SCDs, most individuals have direct experience of young deaths and the provincial effort to ascertain all at-risk individuals has resulted in effective management and treatment leading to significant improvements in survival, especially for individuals provided with an ICD for prophylaxis.[11]

In several inherited cardiomyopathies requiring an ICD, higher rates of depression and lower QOL scores have been reported compared to individuals with ICDs for ischemic or valvular disease.[26] Our narratives suggest similar effects and underline the context of uncertainty and fear in families affected by heritable cardiomyopathies.[13–15]

ICD therapy is lived within this broader familial context. Most of our participants were younger and their accounts revealed multiple areas of burden as in previous research.[1,3,19,20] A majority of ICD recipients had experienced life-saving therapy and those with no discharges had close relatives whose lives had been saved. Participants who experienced multiple shocks recounted high levels of psychosocial burden. Practical concerns were manifest. The loss of a driving licence following an ICD discharge was a recurring and upsetting theme. Additional worries, relating to clothing choices, holiday destinations, and sporting activities (previously enjoyed without burden), were enunciated. Romantic and social relationships were affected and may be key factors contributing to elevated anxiety and distress in these families. For those shown to be negative for the mutation, survivor guilt was present.[27]

These practical issues may help explain contradictory quantitative findings. Reduced QOL in young adults with an ICD for the prevention of SCD that returned to normal levels after one year has been reported.[19] Our participants had their ICDs for an average duration of 10 years and although this study was not designed to address quality of life using validated scales, the rich narratives reflect ongoing issues many years post ICD surgery. These issues have clinical relevance, as anticipatory guidance regarding practical and psychological sequalea may be a needed component of pre-implantation counselling: assuring individuals that psychosocial issues are common may lessen some of the post-implantation distress.

Physicians focus on clinical issues during the consent process for ICD therapy, giving less consideration to psychosocial, economic or advance care aspects of living with a device.[28] The severity of this genetic subtype of ARVC lends urgency to treatment as family history shows that for many, their first clinical symptom was their last,[29] so it is less surprising that issues other than the SCD risk have been afforded a lower priority. However, ICD therapy is often situated in the context of a family coping with multiple affected family members, the fear of SCD, feelings of guilt and associated psychological sequalea. These issues point to the need for pre- and post-ICD implantation psychological support for these families. Specialised cardiac genetic clinics have been associated with improved well-being of patients affected by hypertrophic cardiomyopathy,[30] and we are encouraged that the respondents recognised the benefit of early diagnosis and treatment in the saving of young lives,

a sentiment of acceptance and gratitude that crossed all groups. However, the recognition of psychological issues in family members and partners makes it imperative that the physical life-saving benefits are followed by acknowledgment of, and evidence-informed care for, the psychological burdens reflected herein. These data highlight the many psychological and practical burdens for patients and family members. The effects appear not to dissipate with time, likely reflecting the progressive and lethal nature of this ARVC genetic subtype. It is likely that broader education about the burdens experienced by this patient population (with a focus on the extended family), and a move towards integrating psychology services with the cardiac genetics clinic will provide benefit. That the majority of this population resides rurally makes the provision of this type of care difficult, but no less relevant.

Limitations

This is a small group of individuals that limits the generalisability of results. We did not reach data saturation with the groups, particularly carriers who had multiple vs. no discharges or with adolescents living with an ICD. We were unable to recruit those who declined testing (a small group given the severity of the disease) and those who had progressed to heart transplant. We had more female than male respondents (who are more difficult to recruit given that many work off shore, in another province or in the fishery). We also chose not to approach families that had known about their disease for a short time. All participants had undergone genetic testing so they may have specific perceptions that cannot be generalised. However, the narratives are rich, and the themes outlined are repeated across respondent groups. We also note that these data provide the foundation for quantitative study of the psychological impacts of ICD therapy. Future research of the team includes a survey study to measure depression, anxiety and post-traumatic stress across carriers, non-carriers and spouses using validated psychosocial instruments.

Future research

Many questions remain to be answered. We need to address (i) how psychological effects change across the lifespan, (ii) the experience of severe disease in close relatives (e.g. cardiac transplantation), (iii) the magnitude of diagnosable mental illness and (iv) which aspects of the psychological burdens seen are related to (a) genetics, (b) the disease process and (c) the treatment.

Acknowledgements

We thank the families of Newfoundland and Labrador affected by ARVC for their continued support of our research.

Declaration of conflicting interests

The authors declare that there is no conflict of interest.

Funding

The author(s) disclosed receipt of the following financial support for the research, authorship, and/or publication of this article: This work was supported by the Atlantic Canda Opportunties Agency, Atlantic Innovation Fund (grant no. 781-16881-197778).

Ethical approval

None.

Guarantor

Dr Holly Etchegary is the guarantor for the work.

Contributorship

HE and KH conceived the study, and all authors contributed to the final choice of study design and methods. DP, TY, and KH are co-leads of the broader project from which this work was funded. CS collected the data; HE, KH, and SC analysed the data. HE drafted the manuscript; all authors read and approved the final version.

References

1. Ingles J, Sarina T, Kasparian N, et al. Psychological well-being and posttraumatic stress associated with implantable cardioverter defibrillator therapy in young adults with genetic heart disease. *Int J Cardiol* 2013; 168: 3779–3784.
2. Ingles J, Yeates L, Hunt L, et al. Health status of cardiac genetic disease patients and their at-risk relatives. *Int J Cardiol* 2013; 165: 448–453.
3. James CA, Tichnell C, Murray B, et al. General and disease-specific psychosocial adjustment in patients with arrhythmogenic right ventricular dysplasia/cardiomyopathy with implantable cardioverter defibrillators: a Large Cohort Study. *Circ Cardiovasc Genet* 2012; 5: 18–24.
4. Schinkel AFL. Implantable cardioverter defibrillators in arrhythmogenic right ventricular dysplasia/cardiomyopathy: patient outcomes, incidence of appropriate and inappropriate interventions, and complications. *Circ Arrhythm Electrophysiol* 2013; 6: 562–568.
5. Etchegary H, Enright G, Audas R, et al. Perceived economic burden associated with an inherited cardiac condition: a qualitative inquiry with families affected by arrhythmogenic right ventricular cardiomyopathy. *Genet Med* 2016; 18: 584–592.
6. Marcus FI, McKenna WJ, Sherrill D, et al. Diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia. *Eur Heart J* 2010; 31: 806–814.

7. Groeneweg JA, Bhonsale A, James CA, et al. Clinical presentation, long-term follow-up, and outcomes of 1001 arrhythmogenic right ventricular dysplasia/cardiomyopathy patients and family members. *Circ Cardiovasc Genet* 2015; 8: 437–446.
8. Simpson CS, O'Neill BJ, Sholdice MM, et al. Canadian consensus conference 2003: Assessment of the cardiac patient for fitness to drive and fly – executive summary. *Can J Cardiol* 2004; 20: 1313–1323.
9. Milting H, Klauke B, Christensen AH, et al. The TMEM43 Newfoundland Mutation p.S358L causing ARVC5 was imported from Europe and increases the stiffness of the cell nucleus. *E Heart J* 2014; 36: 872–881.
10. Merner ND, Hodgkinson KA, Haywood AF, et al. Arrhythmogenic right ventricular cardiomyopathy type 5 (ARVD5) is a fully penetrant, lethal arrhythmic disorder caused by a missense mutation in the TMEM43 gene. *Am J Hum Genet* 2008; 82: 809–821.
11. Hodgkinson KA, Howes AJ, Boland P, et al. Long-term clinical outcome of arrhythmogenic right ventricular cardiomyopathy in individuals with a p.S358L mutation in TMEM43 following implantable cardioverter defibrillator therapy. *Circ: Arrhythm Electrophysiol* 2016; 9: 1–9.
12. Manuel A and Brunger F. Making the decision to participate in predictive genetic testing for arrhythmogenic right ventricular cardiomyopathy. *J Genet Counsel* 2014; 23: 1045–1055.
13. Ormondroyd E, Oates S, Parker M, et al. Pre-symptomatic genetic testing for inherited cardiac conditions: a qualitative exploration of psychosocial and ethical implications. *Eur J Hum Genet* 2014; 22: 88–93.
14. Smart A. Impediments to DNA testing and cascade screening for hypertrophic cardiomyopathy and long QT syndrome: a qualitative study of patient experiences. *J Genet Counsel* 2010; 19: 630–639.
15. Van der Werf C, Onderwater AT, Van Langen IM, et al. Experiences, considerations and emotions relating to cardiogenetic evaluation in relatives of young sudden cardiac death victims. *Eur J Hum Genet* 2014; 22: 192–196.
16. Borne RT, Varosy PD and Masoudi FA. Implantable cardioverter-defibrillator shocks: epidemiology, outcomes, and therapeutic approaches. *JAMA Intern Med* 2013; 173: 859–865.
17. Sears SF and Conti JB. Quality of life and psychological functioning of ICD patients. *Heart* 2002; 87: 488–493.
18. Von Känel R, Baumert J, Kolb C, et al. Chronic posttraumatic stress and its predictors in patients living with an implantable cardioverter defibrillator. *J Affect Disord* 2011; 131: 344–352.
19. Verkerk AJ, Vermeer AM, Smets EM, et al. Quality of life in young adult patients with a cardiogenetic condition receiving an ICD for primary prevention of sudden cardiac death. *Pacing Clin Electrophysiol* 2015; 38: 870–877.
20. Webster G, Panek KA, Labella M, et al. Psychiatric functioning and quality of life in young patients with cardiac rhythm devices. *Pediatrics* 2014; 133: e964–e972.
21. Pullman D and Hodgkinson K. The curious case of the De-ICD: Negotiating the dynamics of autonomy and paternalism in complex clinical relationships. *Am J Bioeth* 2016; 16: 3–10.
22. Etchegary H, Pullman D, Simmonds C, et al. 'It had to be done': genetic testing decisions for arrhythmogenic right ventricular cardiomyopathy. *Clin Genet* 2015; 88: 344–351.
23. Sandelowski M. Whatever happened to qualitative description? *Res Nurs Health* 2000; 23: 334–340.
24. Glaser B and Strauss A. *The discovery of grounded theory*. Chicago, IL: Adeline, 1967.
25. Pope C, Ziebland S and Mays N. Analysing qualitative data. *BMJ* 2000; 320: 114–116.
26. Jacq F, Foulldrin G, Savouré A, et al. A comparison of anxiety, depression and quality of life between device shock and nonshock groups in implantable cardioverter defibrillator recipients. *Gen Hosp Psychiatry* 2009; 31: 266–273.
27. Graceffa A, Russo M, Vita GL, et al. Psychosocial impact of presymptomatic genetic testing for transthyretin amyloidotic polyneuropathy. *Neuromuscul Disord* 2009; 19: 44–48.
28. Hill L, McIlfatrick S, Taylor BJ, et al. Implantable cardioverter defibrillator (ICD) deactivation discussions: reality versus recommendations. *Eur J Cardiovasc Nurs* 2016; 15: 20–29.
29. Hodgkinson KA, Connors SP, Merner N, et al. The natural history of a genetic subtype of arrhythmogenic right ventricular cardiomyopathy caused by a p.S358L mutation in TMEM43. *Clin Genet* 2013; 83: 321–331.
30. Ingles J, Lind JM, Phongsavan P, et al. Psychosocial impact of specialized cardiac genetic clinics for hypertrophic cardiomyopathy. *Genet Med* 2008; 10: 117–120.