Psychological Issues in Managing Families with Inherited Cardiovascular Diseases

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The field of cardiovascular genetic counseling has evolved dramatically in recent years largely to manage the unique psychological needs of the inherited cardiovascular disease patient population. For many, there can be difficulty in coming to terms with a diagnosis, whether it be adjusting to lifestyle recommendations such as exclusion from competitive sports or living with a small but remarkable risk of sudden cardiac death. For those considered at risk of life-threatening ventricular arrhythmias, the decision to have an implantable cardioverter defibrillator can be difficult. Living with the device, especially for those who are young and those who receive multiple shocks, can precipitate psychological distress and poor adaptation to the device. Family members who experience a sudden cardiac death of a young relative have a significant risk of poor psychological outcomes. The roles of the cardiac genetic counselor in facilitating patients' adaptation to their diagnoses and management and recognizing when additional support from a clinical psychologist is needed are key to ensuring families receive the best possible care.

Inherited cardiovascular diseases can affect all aspects of the heart. They include cardiomyopathies, arrhythmia syndromes, and metabolic and connective tissue diseases, as well as numerous other genetic syndromes that can present with cardiac manifestations (Fig. 1). As a result, the field of cardiac genetic counseling has rapidly emerged as a growing subspecialty. Sudden cardiac death is a rare but tragic outcome among those with inherited cardiovascular diseases. Managing the risk of a future sudden cardiac death can add layers of complexity to more traditional genetic counseling roles. For example,

eliciting a family history from a patient newly diagnosed and told to consider an implantable cardioverter defibrillator by their cardiologist for prevention of fatal arrhythmias will be challenging. Likewise, explaining inheritance risks and undertaking pretest genetic counseling will not be straightforward for a family that have recently come to medical attention because of the sudden cardiac death of a young and otherwise healthy relative. In many cases, the pressing concern is making sense of the situation that confronts them. Providing information about their cardiovascular disease and management,

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J. Ingles

Figure 1. Inherited cardiovascular diseases.

and heritability, in combination with psychotherapeutic counseling are key skills for a cardiac genetic counselor. Counselors need to be as comfortable discussing the psychological aspects of adjusting to an implantable cardioverter defibrillator as they would be discussing inheritance risks. Counseling skills to sensitively convey important information while attending to patients' and families' psychological responses are roles that define cardiac genetic counselors as key partners in a specialized multidisciplinary team. This review aims to describe the psychological issues frequently encountered by patients and family members with inherited cardiovascular diseases, specifically inherited cardiomyopathies and arrhythmia syndromes, and to demonstrate the vital role of cardiac genetic counseling in patient care.

ASSIMILATING DIAGNOSIS OF AN INHERITED CARDIOVASCULAR DISEASE

Cardiovascular disease remains the leading cause of death in the United States (Benjamin et al. 2019), often occurring in the elderly population, with environmental factors that include lifestyle choices that contribute to disease risk and development (McGovern et al. 1996). In contrast, patients with inherited cardiovascular diseases often present with symptoms in childhood, adolescence, and early adulthood, and the cause is largely due to a gene variant that is inherited in an autosomal-dominant pattern.

Many patients with inherited cardiovascular diseases have only minimal cardiac symptoms and yet are required to make significant lifestyle adjustments to live with an elevated risk of developing the most serious outcomes: heart failure and sudden cardiac death. As such, adaptation to the diagnosis of a hereditary cardiovascular disease can be a challenging long-term process for patients and their families.

Collectively, hereditary cardiovascular diseases present similar genetic counseling issues. Adaptation to a diagnosis of any of these conditions at a young age, and specifically in cases in which there is little, if any, experience living with the disease in the family, can be challenging in many ways. There is a need to make sense of this new health threat, considering the prognostic and management information provided by the clinical team, which may require commencement of medications despite feeling well. Periodic follow-up with a cardiologist is indicated, and because of the risk of sudden cardiac death, some patients undergo prophylactic implantable cardioverter defibrillator (ICD) therapy. Other complications more frequently encountered are development of atrial fibrillation and progression to heart failure, in some instances requiring cardiac transplant. Cardiac symptoms frequently reported by patients include chest pain, shortness of breath on exertion, dizziness (presyncope), blackouts (syncope), and palpitations. For many patients with one of these conditions, there will be few if any cardiac symptoms. Clinicians' ability to predict who will experience poor clinical outcomes is limited. Even within a family, it is common to see the full phenotypic range expressed among relatives. In most cases, having a severely affected family member does not mean that other relatives will be similarly affected. For patients, direct experience with the disease will significantly affect perceptions about prognosis (Ormondroyd et al. 2014; Connors et al. 2015). As such, learning of this variability in the clinical course and uncertainty about the progression of disease can be difficult.

Hereditary arrhythmia syndromes often present in childhood and adolescence, with sudden onset of arrhythmias leading to symptoms

like palpitations, dizziness, and syncope (Priori et al. 2015). They present with a greater risk of ventricular arrhythmias that can result in sudden cardiac death. Taking a family history from patients with hereditary arrhythmia syndromes will often reveal significant cardiac events among relatives, such as frequent “fainters,” sudden infant death syndrome (SIDS), and epilepsy diagnoses, and, importantly, fatal ventricular arrhythmias can be misattributed to other causes if they occur while driving (motor vehicle accident) or swimming (drowning). Attention to the details of any young deaths in the family is critical to determining whether there may have been an underlying cardiac cause. Eliciting this information from the family can stir up feelings of grief and loss, and it can be evident that there is significant anxiety about the risk to other relatives, especially children (Ormondroyd et al. 2014; Burns et al. 2016). In contrast, hereditary cardiomyopathies typically present later in life, from teenage years through to adulthood, and will often result in cardiac-limiting symptoms, such as chest pain and shortness of breath at some point during life (Watkins et al. 2011). The impact on physical health-related quality of life is thus an important consideration for this group. Historical physical activity recommendations have further compounded this issue, with many patients ceasing to do any form of physical activity because of the perceived risk of sudden cardiac death (Sweeting et al. 2016).

RESEARCH FINDINGS

Numerous studies have assessed self-reported psychological outcomes or explored perceptions following diagnosis of a hereditary cardiovascular disease. Research that has generated evidence to help shape the field is presented in Table 1. In one of the largest quantitative studies, health-related quality of life was assessed in more than 400 individuals with inherited cardiovascular diseases, including both affected patients and at-risk relatives (Ingles et al. 2013b). Among those with inherited cardiomyopathies such as hypertrophic cardiomyopathy (HCM) and dilated cardiomyopathy (DCM), there was lower

physical quality of life, whereas mental quality of life was generally maintained. In those participants with long QT syndrome (LQTS), psychological quality of life was reduced compared to rates in the general population. These significant differences between hereditary cardiomyopathies and hereditary arrhythmia syndromes have been reported by others, and likely relate to the differences between the disease profile among the two groups (Table 1).

Medications are recommended for many patients with inherited cardiovascular diseases, especially for those with arrhythmia syndromes. For young patients, adherence to β -blocker medication can be strongly influenced by psychological and social factors (Ingles et al. 2015; O'Donovan et al. 2018). In the setting of LQTS, β -blocker therapy is shown to reduce the likelihood of cardiac arrhythmias and is an important first-tier treatment (Chockalingam et al. 2012). Arrhythmias while taking a β -blocker can signal the need for additional therapy such as an ICD. In the medical literature, studies of adherence to any prescribed therapy indicate that psychological factors, including illness perception, beliefs about the medication, self-efficacy (i.e., their confidence in their ability to take the medication), and psychosocial well-being, are important determinants of medication adherence (Jin et al. 2008). One study examining adherence to β -blocker therapy among patients with LQTS found 16% were not adherent (O'Donovan et al. 2018). Those who were younger and had an inherited arrhythmia syndrome reported lower self-efficacy, greater concerns, low necessity beliefs, and poorer understanding of their disease. Importantly, they were also less likely to take their prescribed β -blocker medication. Another study investigating self-reported medication adherence among patients with HCM found that 30% were nonadherent and more likely to be from a minority ethnicity with self-reported anxiety symptoms (Ingles et al. 2015). These studies provide evidence of the need for genetic counseling interventions to uphold adherence to prescribed medical therapy. This may be an achievable goal of genetic counseling. Targets of the interventions may be affirming beliefs about the cardioprotective role of the medica-

J. Ingles

Table 1. Key studies investigating the psychological and social impact of living with an inherited cardiovascular disease

Study	Disease setting	Study design	Psychosocial measures	Key findings
Bates et al. 2019	SCD in the young	N = 33 first-degree relatives following a SCD of a young relative and no premorbid diagnosis	Hospital anxiety and depression scale (HADS), psychological adaptation to genetic information scale (PAGIS), impact of events scale-revised (IES-R), multidimensional scale of perceived social support (MSPSS)	More than one-third reported poor adaptation to genetic information following the postmortem molecular autopsy. Those with poor adaptation were more likely to have posttraumatic stress symptoms, depression, lower perceived social support, and support from significant others, family members, and friends.
Ingles et al. 2016	SCD in the young	N = 103 first-degree relatives following a SCD of a young relative and no premorbid diagnosis	Short Form Health Survey (SF-36), depression anxiety stress scale (DASS-21), IES-R, prolonged grief disorder scale	Prolonged grief was reported by 21% (36% of mothers, 42% of those who witnessed the death) and posttraumatic stress symptoms reported by 44% (59% of mothers, 67% of those who witnessed the death). Overall almost 50% of relatives reported symptoms requiring intervention with a clinical psychologist, on average 6 yr after the death.
van der Werf et al. 2014	SCD in the young	N = 9 first-degree relatives or spouses; semistructured interviews, analyzed thematically	Qualitative thematic analysis of interview transcripts	There was a strong need to understand the cause of the death and wanting to prevent future deaths in their family. The current multidisciplinary clinic model was appreciated, although ways to improve information processes and better support of decision-making were deemed important.

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Psychological Issues with Inherited Cardiovascular Diseases

Table 1. Continued

Study	Disease setting	Study design	Psychosocial measures	Key findings
Ingles et al. 2013b	HCM, DCM, ARVC, LQTS, CPVT	N = 409 participants (affected and at-risk relatives); cross-sectional self-report survey	SF-36	Physical component scores were impaired for patients with HCM, DCM, and CPVT. Mental component scores were impaired in those with LQTS. In HCM, predictors of poor physical component scores were female gender, comorbidities, and higher NYHA functional class.
James et al. 2012	ARVC	N = 86 adults with ARVC and an ICD	Florida shock anxiety scale (FSAS), Florida patient acceptance survey (FPAS), Duke activity status index, HADS	Overall device acceptance was normative; however, there were marked body image concerns, particularly among younger patients. Younger patients also had greater device-specific anxiety. Those who showed poor adjustment to their device were more likely to have anxiety and depression.
Hamang et al. 2010	HCM, LQTS	N = 127 (affected and at-risk relatives); cross-sectional self-report survey	SF-36	All participants had impaired general health scores compared to the general population. Employment, higher education, and referral to genetic counseling through a family member were associated with improved health status.
Christiaans et al. 2009	HCM	N = 228 (gene-positive relatives, affected and unaffected)	SF-36, HADS, revised version of Illness Perception Questionnaire (IPQ-R)	Quality of life and distress were worse in those with manifest HCM compared to those without. Cardiac symptoms and a stronger belief in consequences of

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J. Ingles

Table 1. *Continued*

Study	Disease setting	Study design	Psychosocial measures	Key findings
				carriership were associated with worse quality of life. Illness and risk perception were major determinants of quality of life and distress.
Farnsworth et al. 2006	LQTS	N = 31 parents; qualitative phenomenological study; parents of children with LQTS	Explore fear of death and quality of life	Parents reported fear of their child dying and strategies they used to manage their fear. There was frustration about the lack of LQTS knowledge from health-care professionals.
Hendriks et al. 2005	LQTS	N = 36 parents; longitudinal surveys and one semistructured home interview; parents of gene carrier children	Impact of events scale (IES), Spielberger State Anxiety Inventory (STAI-s), and Beck Depression Inventory (BDI)	Parents of gene carrier children reported greater distress than parents of noncarriers. Up to one-third reported clinically relevant distress. More distress was reported among those with sudden death in the family.

(SCD) Sudden cardiac death, (HCM) hypertrophic cardiomyopathy, (DCM) dilated cardiomyopathy, (ARVC) arrhythmogenic right ventricular cardiomyopathy, (LQTS) long QT syndrome, (CPVT) catecholaminergic polymorphic ventricular cardiomyopathy, (NYHA) New York Heart Association, (ICD) implantable cardioverter defibrillator.

tion and their confidence to take it on a regular basis. Challenging unproductive illness and medication perceptions and acknowledging the emotional barriers to adherence have been shown to be beneficial in other settings (Petrie et al. 2012) and may be applied and studied in the care of hereditary cardiovascular syndromes.

RISK OF SUDDEN CARDIAC DEATH

Adapting to living with a risk of sudden cardiac death affects all patients. Discussing strategies to clinically manage this risk often takes precedence over all other topics in clinic visits, and being considered at increased risk of a ventricular arrhythmia is associated with heightened

anxiety and poorer psychological well-being. Patients may be pragmatic about their own risk of sudden cardiac death, although in some cases, there is high anxiety and worry (Ormondroyd et al. 2014). For many, the risk to their children is forefront (Farnsworth et al. 2006). A Dutch study reported parental distress in up to one-third of parents with children who inherited a pathogenic variant for LQTS (Hendriks et al. 2005). Among those with distress, being familiar with the disease for a longer time, experiencing a sudden cardiac death in the family, having a lower formal education, and expressing dissatisfaction with the information provided were each shown to be associated with psychological distress. A qualitative study explored fear

of sudden cardiac death among parents of children with LQTS (Farnsworth et al. 2006). Parents described coping mechanisms they employed to manage their fear, which included providing children with phones to allow them to check in regularly, using a baby monitor in the child's room at night, and keeping an automatic external defibrillator nearby. Many were involved in increasing awareness of LQTS and ensuring resuscitation action plans were in place in the child's community. Another qualitative study demonstrated challenges associated with a diagnosis of LQTS and risk of sudden cardiac death (Andersen et al. 2008). Those with early and gradual experience with LQTS reported less worry, but their main concern was fear for their children or grandchildren. Further, there was frustration with the limited understanding and knowledge among health professionals, in some cases providing erroneous treatment advice.

DECISION-MAKING REGARDING IMPLANTABLE CARDIOVERTER DEFIBRILLATOR THERAPY

Current knowledge about risk factors predisposing to sudden cardiac death is incomplete. Considering that the incidence of sudden cardiac death is very low, the number of patients who undergo implantation of an ICD is proportionately large. Managing risk of sudden cardiac death presents novel issues, particularly among those with heritable cardiovascular disease in which patients are often young. Given that, in most cases, prolonged ventricular arrhythmias are fatal, with little likelihood of a second chance to reconsider the management strategy, the stakes for making the correct clinical decision are high. Inherently, sudden cardiac death is an unpredictable event, often described as a moment in time. Although a patient may have been a regular runner for most of his or her life, why on one particular early morning run he or she suffered a sudden cardiac arrest is most often unknown.

The decision to undergo ICD implantation is often one the patient feels they have little control over. For example, some patients will experience an unexplained syncope and find themselves in the hospital with the decision to

have an ICD seeming like an urgent and necessary next step in their management. They may later question whether the ICD was needed (Ingles et al. 2013a). Facilitating patients' efforts to weigh the benefits and potential harms of this therapy is an important component of cardiac genetic counseling (Ingles et al. 2011). Prevention of sudden cardiac death is the goal; however, being aware of the potential for low psychological well-being, inappropriate shocks, regular and ongoing device checks, and complications such as infection and device issues including lead fractures are important risks for patients to give their full consideration.

Shared decision-making as a model for deliberation over ICD recommendations has gained favor in the literature (Lewis et al. 2018). Shared decision-making in this setting includes ensuring patient values and preferences are explored and considered. One qualitative study assessed decision-making processes of patients receiving an ICD for any cardiac condition (Carroll et al. 2013). The decision-making process about the ICD was strongly influenced by the clinician's recommendation and a new awareness of the risk of sudden cardiac death. Another qualitative interview study with cardiologists and patients on their perceptions of the ICD decision-making process found that cardiologists consistently emphasized the benefits of an ICD, whereas discussion of risks was variable (Matlock et al. 2011). In contrast, many patients agreed to the ICD on the advice of their clinician without questioning the risks. Those who declined an ICD were unconvinced the ICD was necessary or believed the risks outweighed the likelihood of sudden cardiac death. These studies highlight the critical need for accurate communication of the uncertain and incremental risk of sudden cardiac death balanced with the potential risks of having an ICD.

LIVING WITH AN IMPLANTABLE CARDIOVERTER DEFIBRILLATOR

For those who choose to receive an ICD, overwhelmingly the psychological and social outcomes are favorable (Sweeting et al. 2017; Maron et al. 2018). Yet, there are certain patients who

J. Ingles

do not fare as well and would benefit from psychological counseling. In the broader cardiovascular disease literature, patients who receive an ICD at a younger age and who receive more shocks from their device are at greater risk of poor psychological outcomes (Sears and Conti 2002). The cardiac genetic counselors play an important role in this setting, in collaboration with clinical psychologists when greater support is needed (Ingles et al. 2011; Caleshu et al. 2016).

For those with inherited cardiovascular diseases, adjusting to life with an ICD can pose additional challenges. James et al. (2012) reported the experiences of patients with a diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC) after a mean follow-up of 3 yr, finding healthy adaptation to the device overall. Despite this, body image concerns were markedly elevated and many reported device-related psychological distress—in particular, the younger patients. Among those with ARVC, having an ICD shock, younger age, poor functional capacity, and shorter time since implant all predicted greater device-specific distress, and poor adjustment was associated with greater reported anxiety and depressive symptoms (James et al. 2012). In a cohort of patients with catecholaminergic polymorphic ventricular tachycardia (CPVT) who had an ICD, those who were younger were more likely to report device-related distress and shock anxiety and even greater body image concerns than the ARVC cohort described above (Richardson et al. 2018).

Anxiety about experiencing a shock is an important consideration, as more shocks have consistently been shown to be associated with lower psychological functioning (Perini et al. 2017). In one study, one-third of patients with hereditary cardiovascular disease who received a shock from their ICD reported symptoms of posttraumatic stress (Ingles et al. 2013a). The most endorsed items on the Impact of Events Scale-Revised (IES-R) were “I tried not to think about it” and “I avoided letting myself get upset when I thought about it or was reminded of it,” both of which reflect avoidance. Among the female patients, approximately half reported clin-

ically relevant posttraumatic stress symptoms. Another study showed disparity in psychological functioning by sex, with female ARVC patients more likely to report anxiety relating to shocks (Rhodes et al. 2017).

In cardiac genetic counseling, there is an appreciation of the potential psychological consequences of an ICD that is critical to empathizing with patients and providing psychotherapeutic counseling. In a clinical setting in which busy clinicians are working to manage clinical symptoms, experienced cardiac genetic counselors who explore ICD adjustment and fears of sudden cardiac death with patients and encourage discussion of any perceived psychological distress are likely to engage in a therapeutic relationship in which psychological well-being is shared and assessed. When marked anxiety or posttraumatic stress are suspected, intervention with a clinical psychologist to manage these responses is timely and important and highlights the health benefits of established multidisciplinary networks (Caleshu et al. 2016).

SPORTS RESTRICTIONS

Physical inactivity is a major determinant of poor health outcomes, recognized as the fourth leading cause of death worldwide (Kohl et al. 2012). Among inherited cardiovascular diseases, because of the observed high incidence of cardiac events occurring during exercise (Maron et al. 2014), exclusion from high-level or competitive sports applies for essentially every patient. Two unfavorable outcomes tend to follow this guidance. The first is that the patient overinterprets this recommendation and subsequently engages in little or no physical activity, increasing risk of weight gain, risk of developing other chronic diseases, and risk of lower quality of life. Alternatively, they may disregard the recommendation and continue high-level competitive sports; this occurs most often when the patient was an avid athlete prior to diagnosis.

Managing the former situation is perhaps a more common occurrence in the inherited cardiovascular clinic and likely a reflection of fear of precipitating ventricular arrhythmias and an

overall reluctance to meet recommended physical activity guidelines among the general population. Physical inactivity has been reported in >50% of patients with HCM (Sweeting et al. 2016), and HCM patients are less active overall than their general population counterparts (Reineck et al. 2013). Factors associated with physical inactivity include perceived barriers such as pain and injury/disability and older age (Sweeting et al. 2016). Most patients report purposeful reduction in physical activity due to their diagnosis of HCM, and less time in physical activity is associated with a negative impact on psychological well-being (Reineck et al. 2013). Interest in interventions to encourage safe increases in exercise capacity and increase overall time spent being physically active were motivation for two recently published trials. One randomized controlled trial investigated moderate-intensity training on peak oxygen consumption in HCM patients, showing overall improvement in exercise capacity following a 16-week training program, with no adverse events, such as arrhythmias, reported (Saber et al. 2017). Another study used a control-theory approach to increase physical activity in HCM patients, incorporating motivational interviewing (Sweeting et al. 2018). Although the primary outcome of an increase in purposeful physical activity was not demonstrated, secondary end points relating to psychosocial outcomes such as health-related quality of life, self-efficacy, and perceived barriers to exercise were significantly improved.

For athletes who are diagnosed with an inherited cardiovascular disease, adopting the recommended lifestyle changes and subsequent adjustment to a new life can be profoundly challenging. Many individuals have built their identity around their athletic ability, and for some it has become a career. As such, the sudden loss of rigorous exercise results in grief. Just as patient acceptance and adherence to prescribed medical therapy relies on person-centered factors, such as understanding the necessity of the recommendation and perceived beliefs, discussion with athletes regarding sports participation may benefit from a similar approach. One study investigated athletes diagnosed with HCM and

reported most found it difficult to adapt to the exercise restrictions (Luiten et al. 2016). Those who reported greater negative psychological impact had a history of elite or competitive sports participation, identified as an athlete, and spent less time exercising since their diagnosis. Having friends and family participate with them in lower-intensity exercise was identified as something that helped them cope with the restrictions. A qualitative study explored perspectives of individuals with HCM and LQTS who continue to exercise against current recommendations and found many did make important modifications to their exercise program over time (Subas et al. 2019). The value of a shared decision-making model in which the athlete's preferences are considered may be beneficial to addressing exercise restrictions. Genetic counselors can be integral in facilitating decision-making and adaptation about sports participation and exercise restrictions and in supporting and motivating physically inactive patients to meet recommended exercise targets.

GRIEF AFTER SUDDEN CARDIAC DEATH

In the inherited cardiovascular clinic, the families who experience the sudden and unexpected death of a previously healthy young relative present the greatest challenges. Sudden cardiac death in the young (<35 yr) occurs at an incidence of ~1.3 per 100,000 persons, and 40% of cases will remain unexplained even after comprehensive postmortem examination (Bagnall et al. 2016). Inherited cardiomyopathies will often be identifiable at postmortem examination, because they result in structural disease. However, inherited arrhythmia syndromes, primarily diagnosed on electrocardiogram in living patients, will leave little evidence of pathophysiological changes at postmortem (Semsarian et al. 2015). In these cases, the presenting symptom can be sudden death, with no premorbid diagnosis, and those remaining unexplained at postmortem are presumed due to an inherited cardiac arrhythmia.

The grief experienced by families in this setting can be profound (Ingles and James 2017). Grief is a normal response to a loss and a process

J. Ingles

of coming to terms with a new reality. It is experienced differently among individuals, but can include disbelief, yearning, anger, sadness, and eventually acceptance (Maciejewski et al. 2007). Kübler-Ross and Kessler (2005) wrote: “The reality is that you will grieve forever. You will not ‘get over’ the loss of a loved one; you will learn to live with it. You will heal and you will rebuild yourself around the loss you have suffered. You will be whole again but you will not be the same. Nor should you be the same, nor would you want to.” In ~7% of bereaved individuals, grief does not progress as expected, essentially resulting in the individual becoming “stuck.” This is known as prolonged or complicated grief, characterized by intense yearning and an inability to imagine a new life without their loved one more than 6–12 mo after the death (Simon 2013). Prolonged grief requires intervention by a clinical psychologist or psychiatrist and poses greater risk of adverse health outcomes, suicide, and comorbid psychological conditions such as depression and posttraumatic stress disorder (Prigerson et al. 1997). Factors related to experiencing prolonged grief include being female, the nature of the relationship to the deceased, sudden and unexpected deaths, and poor understanding of the circumstances of the death, among other factors (Fujisawa et al. 2010).

The largest study to quantify psychological difficulties among first-degree relatives of a decedent in which the death was considered due to an inherited cardiovascular disease found almost 50% reported clinically significant posttraumatic stress symptoms or prolonged grief (Ingles et al. 2016). Specifically, 21% reported prolonged grief, including 36% of mothers of the decedent and 42% of those who witnessed the death. Posttraumatic stress symptoms were reported by 44% overall, including 59% of mothers of the decedent and 67% of those who witnessed the death (Fig. 2A). Witnessing the death or discovering the decedent's body was associated with a four to five times greater risk of prolonged grief or posttraumatic stress symptoms. These findings are in contrast to a growing body of literature around family-witnessed resuscitation efforts by emergency medical services,

which suggest this may provide positive psychological benefits (Robinson et al. 1998; Jabre et al. 2013). Although witnessing resuscitation may be beneficial for families when an older relative suffers a cardiac arrest, when the relative is a young person, witnessing the resuscitation efforts and discovery of the body are the highest risk factors for poor psychological outcomes.

Another study investigating psychological well-being in families after a young sudden cardiac death showed that >50% reported clinically significant symptoms of anxiety, on average 4 yr after the death (Yeates et al. 2013). A qualitative analysis in the same study reported numerous factors that helped or hindered participants' ability to cope after the death (Fig. 2B). Experiences of relatives attending a specialized multidisciplinary clinic in the Netherlands identified key reasons for attending the clinic included a need to understand the cause of death and to prevent another sudden cardiac death occurring in the family (van der Werf et al. 2014). Indeed, these study outcomes reflect the overwhelming concerns expressed by relatives attending following a sudden death.

Practically, for a cardiac genetic counselor seeing families after a young sudden cardiac death, ensuring the family has the opportunity for appropriate clinical management, genetic investigations, and psychological counseling is critical. In the current clinic models for cardiovascular genetics, there can be a tendency to focus on the clinical and genetic concerns; however, based on the evidence and experiences of specialized clinics, psychological counseling should be offered to all family members (Caleshu et al. 2016; Ingles et al. 2016; Ingles and James 2017). Recognizing that in many cases these families are unlikely to encounter other health professionals following the death, the cardiologist and genetic counselor must be strong advocates for addressing psychological needs and feel comfortable discussing the family members' well-being, community support, and coping effectiveness. With growing interest in the postmortem “molecular autopsy” (genetic testing of postmortem DNA), cardiac genetic counselors must also work to ensure we limit potential harms of this technology in this vul-

Psychological Issues with Inherited Cardiovascular Diseases

Figure 2. Psychological consequences for family members following the sudden cardiac death of a young relative. (A) There was a greater risk of posttraumatic stress symptoms among first-degree relatives who witnessed the death or discovered the decedent's body. This was true across the three subdomains of the impact of events scale (IES-R) (Ingles et al. 2016). (B) Key quotes from a qualitative study illustrating common difficulties reported by family members, including genetic guilt, shock, disbelief, and fear for remaining children (Yeates et al. 2013).

nerable patient group. This includes pretest genetic counseling that sets realistic expectations about the likelihood of a conclusive result and careful review of variants to avoid misclassification. A variant of uncertain significance will be identified in >40% of postmortem cases, and a causative variant identified in only ~10% (Lahrouchi et al. 2017). How a family facing profound psychological challenges can understand and adapt to the high degree of uncertainty this testing often yields is not well understood, but it has the potential to inflict harm. A small 2018 study showed that more than one-third of first-degree relatives of a young victim of sudden

cardiac death showed difficulty adapting psychologically to the postmortem genetic test information (Bates et al. 2019). Further research to establish an effective method to integrate psychotherapeutic counseling into the cardiac clinic setting is needed.

CONCLUSION

Many challenges exist for patients with inherited cardiovascular diseases and their at-risk relatives. Research-focused genetic counselors have already played an important role in understanding the issues faced by this patient

J. Ingles

population, which offers evidence to inform development of more effective management approaches. Although this review has focused on those aspects of care relating to a diagnosis, managing sudden cardiac death risk, ICD therapy, and sudden cardiac death, numerous other challenges remain, particularly related to genetic testing. For the genetic counselor, always being mindful and aware of psychological challenges that diminish our patient's quality of life is paramount and will ultimately help to improve overall management and care of families with heritable cardiovascular diseases.

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J. Ingles

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