Published in final edited form as:

Per Med. 2014 September 1; 11(7): 631–640. doi:10.2217/PME.14.50.

Psychological stress associated with cardiogenetic conditions

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Abstract

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Published reports that have analyzed different themes from this research team include: Barlevy *et al.*, who addressed reproductive decision-making in the context of genetic predispositions to sudden cardiac death [18]; Cohen *et al.*, who discussed special considerations for genetic testing with adolescents [19]; Erskine *et al.*, who described the advantages of an interdisciplinary approach to personalized medicine [20]; Linder *et al.*, who identified major concerns associated with an implantable cardioverter-defibrillator (ICD) [21]; and Erskine *et al.*, who analyzed motivations to pursue genetic testing in individuals with a personal or family history of cardiac events or sudden cardiac death [22]. The entire data set is available from the corresponding author.

Disclaimer

The content of this article is solely the responsibility of the authors and does not necessarily represent the official views of the National Heart, Lung, and Blood Institute or the NIH.

Financial & competing interests disclosure

The authors have no other relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript apart from those disclosed.

No writing assistance was utilized in the production of this manuscript.

Ethical conduct of research

The authors state that they have obtained appropriate institutional review board approval or have followed the principles outlined in the Declaration of Helsinki for all human or animal experimental investigations. In addition, for investigations involving human subjects, informed consent has been obtained from the participants involved.

Aim—Genetic testing now makes it possible to identify specific mutations that may lead to life-threatening cardiac arrhythmias. This article presents data from a qualitative research study that explored the subjective experiences of individuals and families with cardiogenetic conditions. We focus on describing patients' experiences of psychological stresses associated with having a cardiogenetic condition, illustrating the importance of integrating psychological and medical care. This integration of care is particularly important as personalized genomic medicine continues to evolve and the implications of genetic testing have a profound effect on individuals and families.

Methods—The researchers interviewed 50 participants from 32 families. The research team used a systematic, grounded theory procedure to code and analyze interview and focus group transcripts, incorporating multiple coders at several stages of the data analysis process.

Results—Three major themes emerged: a bereavement trajectory associated with sudden death in the absence of prior symptoms; high anxiety about transmitting a genetic mutation; and resilience reflected in positive lifestyle changes and participation in support groups.

Conclusion—This article identifies patient perspectives on personalized genomic medicine in cardiogenetics that can improve clinical care, including: specialized bereavement counseling; improving education about cardiogenetic conditions for medical professionals; parent guidelines for discussing cardiogenetic conditions with their children; information about support groups; and the routine inclusion of clinical psychologists in interdisciplinary treatment teams. Given recent advances in technology and decreasing costs, whole-genome sequencing is likely to become common practice in the near future. Therefore, these recommendations are likely to be relevant for other genetic conditions, as well as the entire field of personalized genomic medicine.

Keywords

cardiogenetic conditions; genetic testing; interdisciplinary treatment; long QT syndrome; personalized genomic medicine; psychological stress and genetic testing; sudden death

Psychological stress associated with cardiogenetics

This study looked at the psychological impact on patients and family members of cardiogenetic conditions such as long QT syndrome, Brugada syndrome, short QT syndrome and catecholaminergic polymorphic ventricular tachycardia. These cardiac arrhythmia syndromes can be caused by genetic mutations that are inherited, usually in an autosomal dominant pattern (i.e., transmitted from parent to child) [1]. Also known as cardiac channelopathies, these conditions lead to changes in the cardiovascular conduction system, specifically the sodium, potassium and calcium ion channels [2]. Because these abnormalities are electrical and not structural, they often go undetected. Individuals may remain asymptomatic until their initial clinical manifestation of a potentially lethal heart arrhythmia or sudden death.

Many physicians have not been trained in diagnosing inherited cardiac arrhythmias, and ECGs can be difficult to interpret, even by those with specialty training. For example, in a study designed to identify at-risk young athletes, Hill *et al.* found that 32% of abnormal ECGs were never identified, 19% of at-risk patients received no warning and 26% of teenagers with normal ECGs were advised to avoid strenuous exercise [3]. In addition, given

the lack of familiarity with the genetic basis of some abnormal ECG findings, genetic testing is often not requested in response to symptoms or as part of an autopsy following a sudden death. Therefore, families are frequently left with the ambiguous conclusion that a loved one died from 'unknown' causes. This lack of clarity generates additional stress for families.

Ingles and Semsarian pointed out that the sudden and unexpected deaths associated with these cardiogenetic conditions, particularly in young people who have been asymptomatic, leave families at a loss as to why an otherwise healthy person has died [4]. Moreover, these deaths frequently occur without witnesses (e.g., during sleep), and ambulance and police reports typically shed minimal light on the cause of death. In addition to feeling upset about the death of a loved one, family members are further stressed by the suddenness of their loss. Unlike the death of someone who has been sick for some period of time, family members in the context of sudden death have had no time to prepare themselves for the loss.

When genetic testing has been recommended, the process of testing often elicits a wide variety of additional stressors for patients and family members. Individuals might have to make decisions about whether or not to undergo genetic testing shortly after they have lost a family member or experienced their own near-death event. Some individuals refuse genetic testing because they fear that testing positive for a mutation will generate anxiety and interfere with their ability to enjoy life. Others welcome genetic testing because a positive finding may enable them to take specific preventative measures for themselves and/or family members based on the precise genetic variant identified. Nevertheless, even those who elect to have genetic testing struggle with the implication of a positive result, worrying that disclosing a positive test result could have a negative impact on a family member [5].

When receiving an inconclusive genetic test result, sometimes referred to as a variant of uncertain significance [6], most people are relieved at first. An inconclusive finding, however, is not a guarantee of health, because many gene variants associated with life-threatening conditions, including cardiac arrhythmia syndromes, have yet to be identified. Thus, patients who receive inconclusive findings may have an unknown mutation and therefore still be at risk of a genetic disorder, leaving the patient with continuing uncertainty. In summary, the medical community's limited familiarity with these cardiogenetic conditions, the phenomena of sudden death in the absence of prior symptoms and the complexities of genetic testing constitute a high stress context for patients and families.

Research on psychological stress & cardiac conditions

A number of studies have explored the psychological stresses associated with cardiogenetics. Merlevede *et al.* interviewed 74 relatives of 53 individuals who had died suddenly and unexpectedly [7]. Participants complained of being left with unanswered questions regarding the cause and circumstances of the death. Also common were feelings of guilt, self-blame and blaming others, including medical personnel. Vincent reported that family members often feared that sudden death would recur in the family and worried about who would die next [8]. Ormondroyd *et al.* observed that the testing of children was a difficult decision for parents to make, as they worried about coping with the risks that a positive test result might bring [9].

Given this stressful context, several organizations have recommended the inclusion of clinical psychologists in medical treatment teams. The summary document of the 32nd Bethesda Conference for the Care of the Adult with Congenital Heart Disease in 2000 stated that emotional health should be of the utmost importance and that these individuals should have access to screening and referral sources for mental health treatment [10]. The Canadian Consensus document also emphasized psychological support for such patients [11]. In one study, researchers interviewed 14 psychologists and social workers with extensive experience in helping patients deal with the psychological sequelae of a cardiac event [12]. Their findings suggested that, given the stress of cardiac conditions, psychologists should be integral members of cardiac rehabilitation teams.

However, the medical community has not always followed these guidelines. In a study of 22 adults with congenital heart disease who were assumed to be 'well adjusted', more than a third ($n = 8$) were actually experiencing a diagnosable psychiatric disorder with prominent symptoms of anxiety or depression [13]. These results suggest that psychological intervention may be necessary, even for cardiac patients who appear emotionally well adjusted.

Methods

A qualitative design was chosen for the current study because it is particularly well suited for studying phenomena regarding which little is known. Over the past two decades, the number of disorders for which genetic testing is available has increased from approximately ten to over 2000 [14]. Using genetic testing in order to discover cardiac channelopathies is even more recent. Given these rapid changes in medical care, relatively little is known about patients' subjective experiences and the psychological impact of the growing field of personalized genomic medicine. Given this lack of information, a hypothesis-generating, grounded theory research design was used. Grounding the data in the actual words of the participants accurately and evocatively reflects the psychological experiences of patients with cardiogenetic conditions.

Participants & recruitment

The sample population included 50 participants representing 32 families recruited via three methods. Table 1 presents the participants' demographic data. All of the participants had experienced their own cardiac event or had a family member who had experienced a cardiac event or had died suddenly. The Institutional Review Board of the Albert Einstein College of Medicine of Yeshiva University reviewed and approved the design of the study.

Sample 1 included slightly more than half of the total study participants ($n = 27$). These patients and family members were recruited from a cardiogenetics clinic associated with the Albert Einstein College of Medicine/Montefiore Medical Center located in the Bronx in New York City (NY, USA). They were being evaluated and treated for cardiogenetic conditions. The interdisciplinary treatment team included a pediatric and an adult cardiologist, two clinical geneticists, postdoctoral fellows in cardiology and genetics, a nurse practitioner, a genetic counselor, an ethicist and three psychologists.

The psychologists introduced the study to some sample 1 participants when they were present at the clinic for medical appointments and reached out to others through letters and follow up telephone calls. All patients were assured that if they declined to participate, their clinical care would not be affected. If patients or family members agreed to participate, they were interviewed, either while at the clinic for a scheduled medical appointment or at an alternative agreed-upon time. A graduate student who attended the cardiogenetics clinic also volunteered to be interviewed and is included with sample 1.

In order to expand the sample, the research team contacted two advocacy organizations. Sample 2 participants were recruited through the Sudden Arrhythmia Death Syndromes Foundation (SADS), an organization for families of children and young adults who are genetically predisposed to sudden death due to heart rhythm abnormalities. Sample 3 participants were recruited through the Sudden Unexplained Death in Childhood Program (SUDC), an organization for families who had experienced the death of a child over 1 year of age of due to unexplained causes. Liaisons at each organization informed their members about the study, either through newsletters or by speaking directly to families. If the families agreed to participate, the liaisons provided the research team with their contact information.

The three samples contained both similarities and differences. In all of the samples, each family contained at least one individual who had experienced their own cardiac event or had a family member who had died suddenly. In all sample 1 and sample 2 families, at least one person had undergone genetic testing. In sample 3, although many of the children's symptoms fit the criteria for cardiogenetic conditions, physicians and medical examiners had recommended genetic testing in only two of the seven families. Most of these families were told that their children had died from unknown causes.

In the current study, including all three subgroups increased the variation in the data. For example, including participants from sample 2 gave voice to the experiences of participants from multiple geographical areas within the USA and from various medical contexts beyond a large, urban medical center. Sample 3 increased the number of families who had experienced the sudden death of a child due to unknown causes, providing information about specific bereavement trajectories. Moreover, because the participants from samples 2 and 3 were recruited from advocacy organizations, data emerged regarding the importance of social support for many families. Finally, just over half of the participants from sample 1 ($n = 15$) were Latino or of African descent, representing a relatively large proportion of these traditionally under-represented groups. Thus, aggregating the data increased the variation of the total sample, enhancing the probability that emergent patterns would be representative of diverse experiences of patients with cardiogenetic conditions.

Procedure

Before each interview or focus group, participants signed a consent form and completed a demographic questionnaire. Consent forms were provided in both English and Spanish, and were discussed, either in person or on the telephone, with each participant. Seven focus groups were conducted; one for the sample 1 participants, three for the sample 2 participants and three for the sample 3 participants. Each focus group included individuals representing either two or more families. Because participants in all three samples lived in widely

separated communities, it was difficult to arrange the focus groups. Thus, all other data collection involved individual interviews or interviews with multiple individuals from the same family.

The interviews and focus groups ranged from 1 to 2 h in length and were conducted in person for sample 1 and sample 3 participants. Sample 2 interviews were conducted by telephone because these individuals lived outside of the New York tri-state area. These participants were mailed a copy of the consent form at the time of recruitment. The form was then discussed with them at the beginning of the telephone call. Copies of the consent forms and demographic questionnaires were mailed to the researchers after completion of the interview or focus group.

The researchers began with the following general, open-ended question: “Tell me the story of your [or your family’s] experience with a cardiogenetic condition [or sudden death].” Specific probes were included in order to ask about attitudes towards genetic testing and sharing genetic information with family members. The research team audiorecorded all of the focus groups and interviews and then transcribed the audiotapes. The transcriptions served as the data for this study. The transcripts were stored on a password-protected computer located in the principal investigator’s office. The research team accessed the data from this computer.

Data analysis

Each of the three psychology research team members (LB Silverstein, N Hidayatallah and M Stolerman) independently coded and analyzed each transcript based on the systematic approach to coding and analysis developed by Auerbach and Silverstein [15]. As suggested by Rubin and Rubin [16], the research team tried to utilize a procedure that was transparent (i.e., another investigator could check what had been done), communicable (i.e., the categories made sense to other investigators) and coherent (i.e., the categories were internally consistent while reflecting individual differences and inconsistencies in the culture). In order to achieve these aims, the research team incorporated multiple participants at each stage of the data analysis process.

The psychology research team members independently identified relevant text, defined as text that was pertinent to the research question. The research team then met together to organize these text segments into a list of repeating ideas that were expressed by multiple participants. Coding at the repeating ideas level continued throughout data collection. When no new repeating ideas emerged in the interviews, the researchers determined that data saturation had been reached. Saturation was reached after 50 participants and no more participants were recruited into the study. The researchers then grouped the repeating ideas into the more abstract level of themes that represented general concepts implicit in the repeating ideas. When consensus was reached within the research team, the data were then presented to the study’s interdisciplinary team for discussion. Once this team reached a consensus, the entire interdisciplinary team met with the study’s advisory board, which included medical, legal and educational professionals working in the fields of genetic testing, cardiogenetic conditions and public health. The research team then further refined the data in response to the feedback that emerged in this discussion.

Results

The total sample was primarily female, white, middle class and married, although there were 16 single individuals and three adolescents. More than half of sample 1 was Latino or of African descent ($n = 15$), while the majority of the other two samples was white.

Table 2 displays interview data organized into repeating ideas and themes that were relevant to the psychological reactions of participants who had experienced cardiogenetic conditions and the sudden death of a family member.

Theme 1: bereavement

A total of 31 participants reported extended periods of bereavement.

"I was absolutely numb"—All sample 3 participants had experienced the death of a child, and most described complex reactions to the loss, including initial disbelief and despair. "I could have taken one of the steak knives and cut off my leg, and I knew I wouldn't feel it... I was absolutely numb" (sample 3 white woman, 42 years of age).

Other participants said that the death of their child was difficult to process because it contradicted the expectation that parents will die before their children. "I remember it like it was a dream... it's not the normal order of things" (sample 3 white woman, 38 years of age).

Most bereaved participants described their deceased family member as a formerly healthy individual free of medical complaints, leaving family members feeling shocked and confused." We were very puzzled about what could have caused [our son's] death. He was in the Marines; strong, healthy" (sample 2 African-American woman, 52 years of age).

One husband and wife specifically noted the difference between losing a child suddenly versus losing a child to a chronic illness. "You still lose a child in a chronic illness – but it is not unexplained – it is not sudden ... you know the cause ... you probably got to say goodbye to your child. I did not get to do that" (sample 3 white woman, 33 years of age).

"I need to know what happened"—Bereaved participants expressed the importance of clarifying the cause of death of their loved one and discussed their search for an explanation. One mother was desperate for a medical explanation to expel rumors about substance abuse and foul play. "Suspicion was all around what really caused the death ... we wanted a death certificate, and we wanted the medical examiner to be thorough" (sample 2 African-American woman, 52 years of age).

Even when participants were given an explanation of their loved one's death, some were left unconvinced and unsatisfied. "Doctors initially came up with a diagnosis of acute bronchopneumonia, which didn't sit well with my pediatrician ... we didn't think it was right" (sample 3 white woman, 38 years of age). Other participants felt frustrated about being left without an explanation for months and even years. "I went 15 years without knowing why my daughter died" (sample 2 White woman, 49 years of age).

"It haunts you forever"—Some participants had a particularly difficult time accepting the death of their family member. "It haunts you forever, your life is never the same ... I couldn't go to work for months" (sample 1 white woman, 55 years of age). The impact of the death often negatively affected multiple members of the family. "It was very hard to integrate [my husband's death] ... my daughter had symptoms of [post-traumatic stress disorder]" (sample 1 Latina woman, 55 years of age).

Some participants reported that other family members blamed them. "My mother-in-law told me that my daughter's death was my fault ... There's a lot of blame in this family around the death" (sample 2 white woman, 45 years of age). Several participants became significantly more hypervigilant after the death of their child, especially in regards to health concerns about other children. "I am constantly afraid. I was constantly watching my kids sleep" (sample 3 white woman, 33 years of age).

Theme 2: reactive anxiety

"I have a lot of guilt"—Participants consistently expressed worry for the next generation. Some parents felt guilty that they had transmitted a potentially fatal condition. "I didn't do my job as a mom. I am the carrier" (sample 2 white woman, 46 years of age). "I still feel like there is something that I could have done" (sample 3 white man, 37 years of age).

The possibility of passing the mutation on to future generations affected some individuals' plans for childbearing. "My husband and I are looking at preimplantation genetic diagnosis and *in vitro* fertilization in order to have a child without long QT" (sample 1 white woman, 34 years of age).

"I was the only one"—Some participants felt that others could not relate to what they had experienced. "A normal 21-year-old doesn't have four EKGs; they don't have stress tests" (sample 1 Latina woman, 21 years of age). One participant talked about her experience of isolation as a child due to the preventive measures that she was advised to take. "I had to sit out in gym all the time ... I was frustrated that I had to wear a heart monitor to school, and I tried to hide it" (sample 1 African-American young woman, 17 years of age).

"It'll always be in the back of my mind"—Participants who had received negative or inconclusive genetic testing results continued to deal with uncertainty, realizing that negative results did not guarantee health. "Why was I having all these seizures and everything? The negative results don't mean that I don't have it" (sample 1 white woman, 25 years of age). "This is kind of confusing. It says, 'This result does not rule out a diagnosis of long QT syndrome.' It'll always be in the back of my mind" (sample 1 white female adolescent, 17 years of age).

"How do I say [to my child], 'I'm afraid you might die'?"—Anxiety and ambivalence about sharing genetic information occurred in some families with children and adolescents. Parents struggled over when and how much to reveal to their child about the disorder. "How do I say to my daughter, 'I'm afraid you might die'?" (sample 1 African-American woman, 29 years of age). Two sample 1 families were so anxious about telling their adolescent sons about the seriousness of the disease that they kept the presence of a

genetic mutation in the family a secret. The result was that one son did not undergo genetic testing. “We wanted to wait to tell our son until he was 18” (sample 1 white man, 61 years of age).

Theme 3: positive outcomes

Despite the psychological stress that accompanied their experiences with illness and/or sudden death, many individuals and families ultimately experienced positive outcomes from genetic testing.

“It gives me closure”—Many participants had lost a relative due to ‘unknown causes’. When a family member was tested and learned of the presence of a mutation, they felt a sense of relief because they now had an understanding about what had happened. “It gives me closure, knowing the cause of death [of my son]” (sample 1 African–Caribbean woman, 57 years of age).

Positive test results also provided a sense of comfort and empowered some families to see that living relatives could be treated before the disease became fatal. “I’m grateful that I did find out the kids have long QT ... I’m able to help them at a very early age with the medicine” (sample 1 Latino man, 31 years of age).

“Every day is a gift”—Some participants adopted healthier lifestyles and more positive outlooks. One man, whose wife had died and whose children had inherited a genetic mutation, made a decision to focus more broadly on improving his family’s health. “It educated me that health is very important so it’s the kind of food we eat, and what kind of habits my children pick up from me” (sample 1 Latino man, 31 years of age).

Similarly, while waiting for her daughter’s test results, a mother implemented healthy coping strategies. “I have tried to become more religious and find other ways of dealing with my anxieties ... going to a yoga-type class or reading self-help books or doing what I need to be a better person” (sample 1 white woman, 48 years of age).

Other families felt an urgency to live every day to its fullest, and developed a greater appreciation of life. “You realize that every day is a gift...you don’t dwell on the negative part” (sample 2 white woman, 51 years of age).

“I wanted to create awareness”—All of the sample 2 and sample 3 participants were involved with advocacy groups and participated in educating others about cardiogenetic disease. “I started this [support organization] because I thought – one thing we can do is get people together” (sample 3 white woman, 42 years of age). These activities provided social support through a sense of connectedness to others for some participants. “I spent a lot of time speaking and teaching in many different venues” (sample 2 white woman, 47 years of age).

Summary

Three major themes emerged from the study, including: a specific bereavement trajectory associated with sudden death in the absence of prior symptoms; high levels of anxiety

emanating from guilt about transmitting a mutation; and examples of patient resilience reflected in positive lifestyle changes and participation in advocacy and support groups.

Discussion

Our findings replicate some themes in the recent literature on the psychological stress associated with cardiogenetic disorders. Similarly to Merlevede *et al.* [7], the current study found that participants experienced feelings of guilt and complained of being left with unanswered questions regarding the cause and circumstances of the death of a family member. Similarly, participants spoke of multiple family stresses in much the same manner as in Vincent's study [8].

In addition, the current study provided a rich description of the multiple and complex contexts in which psychological stressors were experienced by individuals with cardiogenetic conditions and their families. These findings suggest the need for psychological counseling in order to address bereavement and anxiety for individuals and families who are undergoing genetic testing and personalized genomic medicine. They also suggest that, in order for personalized genomic medicine to be maximally useful, it must be accompanied by improvements in medical education for a wide range of professionals.

One important finding is the existence of resilient reactions in many participants. Genetic testing gave some patients a sense of closure and decreased anxiety by providing an explanation for the cause of death of a family member, as well as information in order to plan for the treatment of other family members. Other participants responded with a determination to live each day to the fullest and to increase self-care in terms of healthy lifestyle decisions. Several participants became involved in advocacy organizations that provided them with social support and a sense of purpose as they focused on educating others.

In each of these areas – clinical counseling, medical education and referrals for social support – psychologists have the education and training to make a significant contribution. Overall, the findings make a strong case for the integration of the psychological and physical treatment of cardiogenetic conditions in order to translate recent advances in genetics and personalized genomic medicine into effective clinical care. McDaniel came to a similar conclusion, recommending that healthcare professionals trained as family therapists should be routinely included on genetic healthcare teams [17].

Conclusion

Given recent advances in technology and decreasing costs, whole-genome sequencing is likely to become common practice in the near future, facilitating the identification of genetic contributions to a wide variety of complex conditions, such as asthma, diabetes and hypertension. Such information will allow clinicians to offer personalized genomic care. However, the psychological stress associated with identifying and managing genetic susceptibility will be experienced by many more families facing diagnoses that are similar to cardiogenetic conditions. Thus, some of the recommendations that emerged in the context of

cardiogenetic conditions are likely to be relevant for individuals and families with other genetic conditions as well.

Limitations & recommendations for future research

This study has several limitations. Although the sample size is relatively large for a qualitative study, it is still limited in size. Moreover, the participants were self-selected in that they all chose to participate in the research. Therefore, these participants may hold views that are different from individuals who would decline to participate in research and/or would not want genetic testing.

Acknowledgments

The authors would like to give special thanks to N DeGroat for her assistance as study coordinator and C Auerbach for his review of earlier drafts of this manuscript.

Award RC1HL100756 from the National Heart, Lung, and Blood Institute supported this research.

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Executive summary

Background

- Advances in genetic testing have made it possible to identify individuals with cardiogenetic conditions, including long QT syndrome, which can result in sudden death.
- Cardiogenetic conditions are often not detected prior to an acute cardiac event.
- When death occurs due to cardiogenetic conditions, families often experience stress due to the sudden nature of the death, particularly if the individual was asymptomatic prior to the lethal arrhythmia.
- Genetic testing can result in psychological stress for patients and family members who may be mourning the loss of a family member or who have just experienced their own near-death event.
- Several organizations have recommended that clinical psychologists should be included in medical treatment teams for heart disease, but these guidelines have not been widely heeded.

Methods

- This study used a qualitative research design.
- Researchers interviewed 50 individuals from 32 families, all of whom had experienced their own cardiac event or had a family member who had experienced a cardiac event or had died suddenly.
- Participants were recruited from a cardiogenetics clinic, where patients were treated by an interdisciplinary team, and two advocacy organizations.
- Interview transcripts were coded and analyzed using a grounded theory approach, yielding relevant text, repeating ideas expressed by multiple participants and themes.

Results

- Participants reported a unique bereavement trajectory after the sudden death of a family member, including initial disbelief, confusion in the absence of prior symptoms, a need for an explanation of the death and difficulty accepting the death.
- Participants expressed anxiety related to the genetic transmission of cardiogenetic conditions, loneliness in their experience, inconclusive genetic testing results and decision-making regarding how to discuss cardiogenetic conditions with children and adolescents.
- Positive outcomes included relief associated with clear genetic testing results, adoption of healthier lifestyles and obtaining social support via advocacy groups.

Discussion & conclusion

- Psychological counseling is recommended in order to help individuals and families cope with bereavement and anxiety associated with cardiogenetic conditions and genetic testing.
- Medical professionals can benefit from additional training in diagnosing cardiogenetic conditions.
- Cardiogenetic treatment teams should routinely provide patients and family members with information regarding support and advocacy groups.
- Psychologists should participate as members of cardiogenetic treatment teams in order to help integrate psychological and medical care for these conditions.
- These recommendations may apply to the treatment of a wide range of genetic conditions.

Table 1

Individual and family characteristics of the sample.

Characteristic	Sample 1 (n = 27 [†])	Sample 2 (n = 14)	Sample 3 (n = 9)	Total (n = 50)
Sex:				
Male	8	1	2	11
Female	19	13	7	39
Race/ethnicity:				
White	12	12	8	32
African–American	4	1	1	6
African–Caribbean	1	0	0	1
Latino/Hispanic	10	0	0	10
Biracial (white Asian)	0	1	0	1
Education:				
Less than ninth grade	1	0	0	1
Currently in high school	2	0	0	2
GED	1	1	0	2
High school	4	1	0	5
Some college	5	5	1	11
College degree	3	5	4	12
Graduate degree	9	3	3	15
Unknown	2	0	0	2
Annual household income:				
<US\$25,000	3	2	1	6
US\$26,000–50,000	4	2	0	6
US\$51,000–80,000	7	3	0	10
>US\$80,000	7	7	8	22
Refused	2	0	0	2
Unknown	4	0	0	4
Age:				
<20 years old	3	0	0	3
20–30 years old	8	2	0	10
31–50 years old	4	7	8	19
51–60 years old	8	3	0	11
>60 years old	2	2	1	5
Unknown	2	0	0	2
Marital status:				
Married	10	11	7	28
Cohabiting	2	0	0	2
Separated	1	1	0	2

Characteristic	Sample 1 (n = 27 [†])	Sample 2 (n = 14)	Sample 3 (n = 9)	Total (n = 50)
Divorced	0	0	1	1
Single	13	2	1	16
Widow	1	0	0	1
Participants with sudden death in immediate family	13	9	9	31
Families with adolescent or child as patient [‡]	6/14	9/11	7/7	22/32
Families in which at least one immediate family member had genetic testing	14/14	11/11	2/7	27/32

[†]The graduate student who volunteered to participate in the study is included in the table under the sample 1 frequencies.

[‡]Data referring to families with children as patients or families who had undergone genetic testing are reported in terms of n = 32.

GED: General Educational Development Test, designed to measure the skills and knowledge equivalent to a high school course of study.

Table 2

Number of participants expressing themes and repeating ideas.

Themes and repeating ideas	Sample 1 (n = 27 [†])	Sample 2 (n = 14)	Sample 3 (n = 9)	Total (n = 50)
Bereavement:				
“I was absolutely numb”	8	7	8	23
“I need to know what happened”	7	5	9	21
“It haunts you forever”	6	1	4	11
Reactive anxiety:				
“I have a lot of guilt”	14	12	7	33
“I was the only one”	4	0	3	7
“It’ll always be in the back of my mind”	9	5	9	23
“How do I say, ‘I’m afraid you might die’?”	6	1	1	8
Positive outcomes:				
“It gives me closure”	7	7	0	14
“Every day is a gift”	4	3	0	7
“I wanted to create awareness”	7	14	9	30

[†]The graduate student who volunteered to participate in the study is included in the table under sample 1 frequencies.