Published in final edited form as:

J Genet Couns. 2015 August ; 24(4): 608–615. doi:10.1007/s10897-014-9783-7.

Disclosing Genetic Information to Family Members about Inherited Cardiac Arrhythmias: An Obligation or a Choice?

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Abstract

Inherited cardiac arrhythmias such as long QT syndrome and Brugada syndrome, present clinical as well as ethical, legal, and social challenges. Many individuals who carry a deleterious mutation are largely asymptomatic and therefore may not be diagnosed until after the occurrence of a personal or family member's cardiac event. The familial nature of inherited genetic information raises numerous ethical, legal, and social issues regarding the sharing of genetic information, particularly when an individual found to carry a deleterious mutation refuses to disclose his or her results to at-risk family members who could benefit from life-saving treatments. This qualitative study sought to understand the experiences with genetic testing for individuals ($n = 50$) with a personal or family history of cardiac events or sudden death. Unstructured in-person focus groups or interviews were conducted for each participant in the study. The recordings of these interviews were transcribed verbatim and subsequently analyzed and coded. Participants' comments regarding sharing of genetic information centered around four main themes: (1) motivation to

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All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

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Isha Kalia, M.S., CGC declares that she has no conflict of interest.

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Siobhan M. Dolan, M.D., MPH declares that she has no conflict of interest.

disclose; (2) extent of disclosure; (3) effect of disclosure on family dynamics; and (4) reasons for not sharing genetic information. The majority of individuals believed that affected individuals are obligated to disclose genetic information to family members. In the era of personalized medicine, the disclosure of genetic information provides individuals the opportunities to learn about the genetics, disease characteristics, and treatment options in order to reduce morbidity and mortality in themselves and their family members. Further research is necessary to identify and explore the barriers to sharing genetic information with at-risk family members.

Keywords

disclosing genetic information; cardiac arrhythmia; long QT syndrome; ethical; legal; and social issues; genetic counseling; family communication

In the United States, more than 450,000 people die each year of sudden cardiac death (Ingles & Semsarian, 2007). This high prevalence has raised significant public awareness and encouraged the medical community to attempt to identify the underlying causes of these sudden deaths. Of the etiologies known to be associated with sudden cardiac deaths, inherited cardiac diseases, such as long QT syndrome (LQTS), short QT syndrome, Brugada syndrome, Progressive Cardiac Conduction Disease (PCCD), Hypertrophic Cardiomyopathy (HCM), Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), and Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), represent an important group. These conditions typically have an autosomal dominant mode of transmission and demonstrate both incomplete penetrance and markedly variable expressivity, even within families (Cummings & Priori, 2011; Wilde & Bezzina, 2005). As a result, many individuals who carry a deleterious mutation do not exhibit obvious symptoms of the disease and may not be diagnosed until after an arrhythmic event. These events can be fatal and are often classified as sudden infant death syndrome (SIDS; in children under one year of age) or sudden arrhythmic death syndrome (SADS) (Limelette et al., 2013). In some cases, investigation of a death can lead to a diagnosis of an inherited cardiac disease posthumously (Schwartz et al., 2007).

Commercial genetic testing is currently available for the aforementioned inherited cardiac conditions. If a deleterious mutation is identified in the proband, genetic testing for all first degree relatives is indicated, as each has a 50% chance of carrying the same inherited mutation. In the case of LQTS in particular, gene-specific treatments and specialized interventions can be initiated pre-symptomatically (Wilde & Bezzina, 2005). These interventions may include lifestyle changes, the use of medications, and/or implantable cardioverter defibrillators (ICDs); treatment recommendations vary depending on the subtype of LQTS (see Figure 1). Prophylaxis for individuals at risk of inherited cardiac arrhythmias can be very effective in preventing adverse cardiac events. Management of these cardiogenetic disorders illustrates how personalized medicine can be used to create gene-specific treatments for at-risk families (Beauchamp & Childress, 2001; Schwartz et al., 2000b; Schwartz et al., 2001).

The hereditary nature of these cardiac conditions raises challenging ethical, legal, and social issues with respect to sharing genetic information between family members. Ethical

dilemmas arise when probands who have been discovered to carry a deleterious mutation refuse to disclose their results to at-risk family members (Aktan-Collan et al., 2011; Clarke et al., 2005). For clinicians caring for these individuals, this situation challenges the principle of non-maleficence, one of the guiding principles of medicine (Beauchamp & Childress, 2001). The arguments that a physician's primary duty is to protect his or her patient's privacy (Minkoff & Ecker, 2008; Offit, Groeger, Turner, Wadsworth, & Weiser, 2004) and that a physician has a duty to warn at risk relatives (Pelias, 1991) have both been made. The Health Information Portability and Accountability Act (HIPAA) of 1996 nationally protects the privacy of an individual's health information, thus absolving clinicians of the responsibility to warn at-risk family members. Clinicians can only encourage patients to share their genetic testing results, placing the responsibility on the proband to notify family members.

This qualitative study sought to determine the attitudes and beliefs of participants regarding their subjective experience of receiving a cardiogenetic diagnosis, including genetic testing and treatment. We investigated the motivations and goals behind disclosing genetic testing information and the extent to which individuals believe they should share this information with their family.

METHODS

Sample and Recruitment

For inclusion in the study, all participants were required to have a personal or family history of a cardiac event with a clinical diagnosis of a cardiac arrhythmia, or to have a family member whose death was classified as SIDS or SADS. Participants were recruited from three different sources:

- Individuals who received treatment at the Montefiore-Einstein Center for Cardiogenetics (MECC), an interdisciplinary clinic providing evaluation, testing, and treatment;
- Individuals who responded to a newsletter request from the Sudden Unexplained Death in Childhood Program (SUDC);
- Individuals who responded to a newsletter request from the Sudden Arrhythmia Death Syndromes Foundation (SADS).

All participants provided written informed consent prior to participating in this study. Additionally, each participant completed a questionnaire providing demographic information to be used in data analysis. This study was approved by the Institutional Review Board at Albert Einstein College of Medicine/Montefiore Medical Center, and precautions to protect human subjects were followed throughout the study. Additionally, patients at the MECC were informed that declining to participate in the study would not affect their clinical care.

Data Collection and Analysis

Two members of the clinical research psychology team conducted focus groups ($n=7$ groups) and interviews ($n=20$) via phone or in-person. Focus groups and interviews were unstructured and included open ended questions as probes to facilitate discussion. This unstructured technique using general questions allowed for the team to gather “bottom-up” data in which information was organized by the participants of the study. Researchers began the discussions by asking, “Tell me the story of your or your family member’s cardiac episode [or death].” Subsequent probes included “How did you make the decision to undergo genetic testing?” and “Have you decided to have further genetic testing for yourself and/or your children?” As the study progressed, the probe “Do you think that families who know that they have a cardiogenetic problem should be obligated to share that information with their family members?” was included.

Focus groups and interviews were both conducted as they provided an avenue to explore patients’ and families’ perspectives, attitudes, and emotions. Both focus groups and interviews were conducted because previous research has found that some individuals feel comfortable speaking freely in groups that share their personal experiences, while others find the privacy of individual interviews allows them to be more open (Silverstein, Auerbach, & Levant, 2002). Focus groups were defined as two or more unrelated families, while interviews consisted of one or more related individuals. A total of 27 focus groups and interviews were conducted as summarized in Table 2.

All interviews and focus groups were audio-recorded and transcribed verbatim. Since the data collected from both interviews and focus groups were textual data, both sets of data were combined, as no differences between the patterns in these two contexts emerged. Each transcript was given a specific number (T1, T2, T3, etc) and individual quotes were denoted P1, P2, P3, etc. Additionally, all identifying information regarding participants was removed from the transcripts. Three members of the team then analyzed and coded the transcripts separately, based on a grounded theory analysis developed by Auerbach and Silverstein (Auerbach, 2003). The methodology of grounded theory analysis begins by identifying text that is pertinent to the research question. These quotations are then organized into repeating words, phrases, or ideas expressed by more than one participant. Subsequently, these words, phrases, or ideas are grouped into a second level of more abstract themes. After each stage of data analysis, these themes were presented to the interdisciplinary research team and the project’s advisory board that provided feedback to assure that the analysis was transparent, communicable, and coherent.

RESULTS

A total of 50 individuals representing 32 families were interviewed: 27 from the MECC, 14 from SADS, and 9 from SUDC. Table 1 summarizes the demographic information obtained from the participants. Of the 50 participants interviewed through focus groups and interviews, 23 expressed clear viewpoints about familial obligations toward sharing genetic information. Of these participants, 20 stated that they believe an individual is obligated to disclose genetic information to his/her family members (Table 3). On the other hand, three believed there is not an obligation for one to disclose information to family members. Of the

27 participants who did not express a viewpoint, 16 had informed or were in the process of informing their family members.

Participants' comments revealed four main ideas about sharing genetic information with family members: (1) motivation to disclose, (2) extent of disclosure, (3) effect of disclosure on family dynamics, and (4) reasons to not disclose.

Motivation to Disclose

The most frequently reported motivation for disclosing genetic information was to inform family members of their risk with the goal of potentially saving someone's life. Two participants in particular expressed their opinions regarding the importance of sharing family medical information for this reason:

"Yeah, because it can save a life. You should never keep medical/health history out of your family. [People] have a right to know. You need to know your family history. And they need to know how important it is." (T27:BM)

"I think it's extremely important to let people know if they [have] a deathly disease... That's like knowing you have HIV and having sex with someone. Like that's like a death wish for someone. And if you know that and you withhold that information from a family member... and don't tell them, that's selfish. ...you're withholding [the information] when it can help someone!" (T9:E)

Many participants alluded to the simple nature of sharing genetic information in comparison to the far-reaching ramifications it can have in preventing another family member from dying. One participant expressed this sentiment with regards to how buying a relatively inexpensive medication could prevent a cardiac event:

"I feel obligated to say because what if... you didn't say, and somebody died needlessly, right, and a \$10 medicine at Wal-Mart would have prevented it...if you have got something like [a genetic condition], it should be something that needs to be shared with the family." (T20:MH)

Additional motivations surfaced with regard to long-term implications. Not only were numerous participants concerned with the health of their living relatives, but they were also concerned about saving lives in future generations:

"I think it's a good [thing to share genetic information]. Because my daughter may not have it, but because there's all this research going on, I want to inform her of it. Because when it comes time for her to have her family, she can be informed of it, she can know what to look for in her children." (T8:UP)

"I'm educating them now ...I look at it as it's great that we know what they have because ... it'll help the next generation and stuff. I'm looking at the bigger picture." (T14:PE)

Other participants stressed the importance of disclosing information to family members to assure that their relatives have all the information possible, in order to make informed decisions for their health:

“A lot of people have reservations, but I know if you don’t share [your genetic testing results]... then you’re missing a piece of the puzzle and your puzzle is never complete. So you have to be as open and honest as you can.” (T23:HK)

“I feel like you have at least an obligation to inform [your family members] and what they choose to do or not do is, you know, not really up to you nor should it be.” (T28:JM)

Extent of Disclosure

The decision as to whom participants informed and how much information they shared varied significantly among participants. Additionally, the extent of disclosure was influenced by both the age of the family member and a desire to protect others from worry. Some parents reported being selective when disclosing details of the condition to their children. One mother shared information based on her assessment of her 7 year old daughter’s readiness and interest:

“Well, she knows that it’s related to her father dying... I just said to her, ‘Your heart takes a little bit longer to restart than most people. So they want to give you [a defibrillator] so that if something happens you’ll be ok until someone can get to you, a paramedic, or until they can get you to the doctor.’” (T6:YC)

In contrast, another mother limited the details she disclosed to her 16-year-old daughter due to fear of worrying her daughter:

“Well, I was limited because I didn’t want to scare [my daughter]... I just wanted to cushion her and make her feel like everyone else... I don’t know how to explain it to her- I want to be so open with her because I want her to know, but I don’t want to scare her.” (T23:HK)

Disclosure Altering Family Dynamics

Participants reported that sharing genetic information with family members brought about positive and negative effects to both the individual and the family unit. In some situations, disclosure and dealing with a life-threatening situation brought families closer together due to an increased level of communication:

“I think [our family] got closer than what we were. And we check up on each other all the time.” (T2:MR)

“It’s been really kind of sweet, because of this information, like I don’t even really know [my half-brother] that well, but this information has kind of bonded us... We have kind of been connected and we have been in touch on Facebook with him and his wife...it’s kind of brought us together.” (T28:JM)

On the other hand, several participants reported that disclosure further separated their family due to feelings of animosity and blame, bringing about social tensions and ill effects within the family unit:

“It totally changed our lives. It put a very, very big strain on our family. They always say that life-threatening things bring you closer together, I felt it threw our family apart.” (T24: JWS)

“[My brother-in-law] said something to me six months later like ‘Well, you know I am just, I am so scared I just wish I had never known this’... he was kind of angry at me, that’s what it seemed like.” (T26: DH)

Many participants expressed strong feelings of isolation as well as feelings of frustration toward their family members who refused to pursue testing, even after reaching out to them to disclose their personal genetic information:

“I’ve talked to some of them, and some of them are like extremely stubborn, some are scared, and then there’s others, I just look at them like, ‘You know what? You don’t wanna test yourself, that’s on you, but you got your little kids, so at least test them or test yourself so you know if there’s a possibility you could have gave it to them.’ Now, for my aunt that didn’t do it, I mean, ‘You’re 40, you’re 50, you’re older than I am, I told you, that’s all I could do’.” (T3: MK)

“I sent out the e-mail where you know it turns out [my daughter] tested positive for long QT and...not one person of my family...responded to my e-mail....I had a paragraph in there and said ‘Unfortunately this means that it’s in our genetic makeup... you might consider getting tested,’ you know... several of my siblings have children of course, not surprisingly, and so anyway no one responded [at] all which was really hurtful for me.” (T26: DH)

Several participants expressed their frustration with family members who do not discuss their genetic information with other members of the family.

“My frustration, especially with my father in terms of my half brother and his daughter, is that my dad never told him about my heart condition or what this thing is that’s running in our family... I had a lot of anger at my father for not sharing this information because he put [my half brother]’s health at risk basically by not communicating that.” (T28: JM)

“...her dad and I see it so different. He’s more like ‘We don’t talk about it’ - the pink elephant in the room-he ignores it.” (T23: HK)

Reasons Not to Disclose

The four participants who felt that individuals did not have an obligation to disclose genetic information to family members provided interesting thoughts about the topic of disclosure. Several participants explained their reluctance to disclose because this information inevitably generates worry about health issues in the future. Others attributed their reluctance to disclose genetic information to the fact that they found it difficult to explain the complexities of the disorders effectively:

“It’s not easy to talk about it because even if I talk to my father, or my brother or my sisters, and I try to explain what my daughters have, it’s kind of hard for me to explain because questions that they ask, I don’t know the answer.” (T8: MR)

“It would have been helpful [to have additional printed information] because on top of that too, your family asks you ten million questions... and if you want to ever explain it to someone, you wanna get it right.” (T3: MK)

Other participants expressed feelings of apprehension, in that they did not want to relay difficult news to their family members:

“I would have felt like shit. Excuse my language. It would have been awful to be the bearer of bad news.” (T22: TC)

“I don’t think they should [have an obligation to disclose]. I think that that would be a personal thing between like you and the family members.” (T29: ME)

“No. ...You know I don’t know that I would just put this all out there...we’ll go through this or go have that, or this is what happened in her case.” (T29: FN)

DISCUSSION

Our qualitative research suggests that the prevailing attitude of individuals in families with inherited cardiac arrhythmias is that they should share genetic information with family members. Our model suggests that the primary motivation to disclose is that genetic information could be life saving and thus its benefits far outweigh its risks. However, the extent of disclosure to children varied most widely based on the age of the child and the perceived psychological risk of knowing the information. Our model finds that there are potentially adverse effects on family dynamics, but some families reported positive effects on family dynamics. Lastly, our model revealed a variety of reasons that might deter individuals from sharing genetic information, all of which have important implications for genetic counseling and education of individuals undergoing genetic testing.

The barriers that arise in disclosing genetic information for inherited cardiac disorders are similar to those in various fields of genetics (Aktan-Collan et al., 2011; Clarke et al., 2005). However, inherited cardiac arrhythmias are often diagnosed by sudden cardiac death within a family. This means of diagnosis is a distinguishing feature for the field of cardiogenetics. While many genetic conditions manifest phenotypically, inherited cardiac arrhythmias often remain hidden until a sudden cardiac event. Thus, genetic testing for these conditions allows for direct preventive care for affected individuals and family members.

The most commonly reported motivation for disclosing genetic information was to save someone’s life, especially in the setting of the sudden and often fatal onset of symptoms associated with inherited cardiac arrhythmias. This sentiment is echoed in existing cancer literature, in which the act of disclosing results could save the life of a loved one and affect the lives of generations to come (Aktan-Collan et al., 2000; Stoffel et al., 2008). The time course of the onset of familial cancer syndromes often leaves ample time for deliberation about genetic testing and disclosing genetic information to family members. In the case of inherited cardiac arrhythmias, the acute nature of the onset of the condition can accelerate this timeline. Often times, the diagnosis of inherited cardiac disorders occurs simultaneously with a traumatic event. Thus, it would be beneficial to have a grief counselor, social worker, or psychologist assist genetic counselors and patients in the process of disclosure and

provide services for individual and family counseling. Supporting patients and families with ongoing and continual follow-up is critical. Additionally, families should be given written information in the form of pamphlets and brochures, which they can take home to assist in providing information to other family members.

Even when a decision is made to disclose genetic information, how much information is shared varies depending on the age of onset, severity, and inheritance of the condition (Peterson et al., 2003; Plantinga et al., 2003). Due to the acute nature of cardiac arrhythmias, we found that disclosure to children and family members needed to occur soon after a diagnosis was made in order to begin prophylactic cardiac monitoring or surgery. Since many of these cardiac conditions have a genetic basis, disclosure often invoked a strong sense of guilt for passing on the condition to family members. Management of cardiac arrhythmias often includes restriction from strenuous physical activity, leading to parental perception that their child's quality of life is diminished (Hallowell et al, 2003; Smets et al., 2008). Thus, genetic counseling that emphasizes the clinical utility of the genetic information and the steps that can be taken to save a child's life, might offset the guilt regarding restrictions that a parent or family member might feel.

Our research suggests that educational information that explained these complex inherited cardiac arrhythmias, as well as their mode of inheritance and treatment options, might greatly assist family members in disclosing genetic information to family members. In addition, such information would be particularly useful if it was available in various forms, including materials that are age-appropriate for use with children at various times in their lives (Gallo, Angst, Knafl, Hadley, & Smith, 2005) as well as being culturally and linguistically appropriate.

Barriers to information disclosure are not limited to children, but to extended family as well. We found a common motivation behind not disclosing information to extended family was to prevent anxiety. While the majority of participants thought disclosure should include the extended family, many believed that it was best to selectively disclose. Participants were selective about how much information to disclose (purposely avoiding the gravity of the situation), and to whom to disclose. Some participants limited this disclosure because of feelings of guilt or fear of isolation from their families. Others held back information due to a lack of understanding of the condition or wishing to avoid being the bearer of bad news (Etchegary et al., 2009; Wilson et al., 2004). Studies have found that individuals elected not to inform their relatives of their genetic predispositions to respect their family members' right to remain uninformed (Andorno, 2004; Dufrasne, Roy, Galvez, & Rosenblatt, 2011; Forrest et al., 2003; Gaff, 2014; Hallowell et al, 2003), showing a desire to shy away from paternalistic attitudes and to respect the pluralism of individuals' ways of thinking.

Limitations of the study

The participants in this study were self-selected from the MECC or were part of a support group for sudden death (SUDC) or cardiac arrhythmias (SADS). These individuals are therefore already receiving treatment for an arrhythmia or have experienced the death of an affected family member and are seeking emotional support. Thus, these individuals may be more likely to volunteer to participate in a research study. Because individuals are self-

selected, we were unable to obtain the opinions and viewpoints of individuals who declined the genetic testing process or actively chose not to relay genetic information to their relatives. Additionally, the majority of participants in this study were white and female, which may not be representative of other ethnic or racial groups.

Future directions

Based on the limitations of this study, future studies are warranted to assess the attitudes of individuals who have made a conscious choice not to disclose their genetic testing results to family members in order to assess possible barriers to disclosure. Such an understanding is essential in order to provide effective genetic counseling and patient instruction, as to facilitate the disclosure process to at-risk relatives.

Furthermore, because the aim of our overall study was to assess an individual's experience with genetic testing and inherited cardiac arrhythmias, it did not explore participants' views regarding the role of the physician in sharing information with family members. In order to add to the ethical discussion regarding physicians' obligations to warn at-risk family members, probands' attitudes and opinions should be assessed as well.

Acknowledgments

This work was supported by a NIH grant from the National Heart, Lung, and Blood Institute for *The Ethical and Social Implications of Genetic Testing in the Case of Unexpected Deaths: The Translation of Cardiogenetic Knowledge to Clinical Practice for Families Who Have Experienced a Sudden Unexpected Death* Award Number RC1HL100756. We are very grateful to the families and staff of the Montefiore-Einstein Center for Cardiogenetics who participated in this study.

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Figure 1.
Personalized Recommendations for Individuals with Long QT Syndrome

Table 1

Participant Demographic Information

Sex	
Male	11
Female	39
Geographic Region	
Northeast	39
South	3
Midwest	6
West	2
* Geographical regions are consistent with those of the census bureau.	
Ethnicity	
Latino/Hispanic	10
Non-Latino/Hispanic	40
Education	
Less than 9 th grade	1
GED	2
High School	5 (3 still in HS)
Some College	12
College Degree	12
Graduate Degree	15
Unknown	3
Marital Status	
Married	27
Cohabiting	2
Separated	2
Divorced	1
Widowed	3
Single	15
Annual Household Income	
<25,000	6
\$26,000–\$50,000	6
\$51,000–\$80,000	10
>\$80,000	21
Refused	2
Unknown	5
Age	
<20	1
20–30	9
31–40	9

41–50	14
51–60	11
>60	6

Census 2000 geographic definitions: Introduction-geographic presentation of data. 2011

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Table 2

Focus Group and Interview Enrollment

# Focus Groups and Individual Interviews	Number of Participants Per Focus Group/Interview	Total Number of Participants
7	2–4	18
20	1–4	32

Table 3

Is There an Obligation to Share Genetic Information with Family Members?

Overall Response	Number of Participants (n=50)
Yes	20
No	3
Not Discussed/No Response	27