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Motivation to Pursue Genetic Testing in Individuals with a Personal or Family History of Cardiac Events or Sudden Cardiac Death

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Abstract

Genetic testing is becoming increasingly available for cardiac channelopathies, such as long QT syndrome and Brugada syndrome, which can lead to sudden cardiac death. Test results can be used to shape an individual's medical management and to identify at-risk family members. In our qualitative study, all participants had a personal or family history of a diagnosed cardiac arrhythmia syndrome or sudden cardiac death. Open-ended interviews were conducted individually and in focus groups. Interviews were audio recorded, transcribed verbatim, and analyzed using a qualitative grounded-theory approach. Of 50 participants, 37 described their motivations for pursuing genetic testing for long QT syndrome or another cardiac channelopathy. Participants' motivations included: to find an explanation for a family member's sudden death, to relieve uncertainty regarding a diagnosis, to guide future medical management, to allay concern about children or other family members, and to comply with recommendations of physicians or family members. Perceived reasons not to pursue genetic testing included denial, fear, and lack of information. The genetic counseling and informed consent process can be enhanced by understanding and addressing an individual's internal and external motivations either for or against pursuing genetic testing.

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Keywords

genetic testing; sudden cardiac death; long QT syndrome; motivation; channelopathy; genetic counseling

INTRODUCTION

Cardiac channelopathies with genetic etiologies, such as long QT syndrome (LQTS), Brugada syndrome (BS), and catecholaminergic polymorphic ventricular tachycardia (CPVT), present a unique challenge to clinicians because of the cardiac, genetic, and familial implications associated with a diagnosis. Individuals who have been found to carry deleterious mutations are often tested because of a personal or family history of life-threatening cardiac events or sudden death, which is often classified as sudden infant death syndrome (SIDS) or sudden unexplained death syndrome (SUDS). Through obtaining ECGs, personal and family histories, and now genetic testing results, physicians and other health professionals can effectively manage the medical care of an individual and determine the implications for his or her family members.

To explore attitudes toward and motivations for genetic testing for hereditary arrhythmias, we focused on LQTS as an example of a familial cardiac disorder. LQTS is an autosomal dominant cardiac channelopathy with variable expressivity that affects approximately one in 3000 to one in 7000 individuals (GeneReviews). Of note, one form of LQTS, Jervell and Lange-Nielsen syndrome, is autosomal recessive and is accompanied by congenital deafness, however, this study focuses primarily on the autosomal dominant forms of LQTS. Affected individuals may be completely asymptomatic and be detected during pre-op ECGs, sports physicals, or other routine medical visits. Others may present with symptoms ranging from lightheadedness and syncopal episodes to serious arrhythmias or sudden cardiac death (Roden, 2008). Individuals are diagnosed based on family history, clinical findings, comprehensive cardiac evaluation, and increasingly through molecular testing, which detects mutations in known genes involved in the functioning of cardiac ion channels. Treatment options may include lifestyle modifications, avoidance of certain medications, use of a beta blocker, and an implantable cardioverter defibrillator (ICD) (Goldenberg & Moss, 2008; Wilde & Bezzina, 2005). Individuals with a cause of death classified as SIDS or SUDS may also be diagnosed posthumously via molecular autopsy (Schwartz et al., 2000; Wang et al. 2007).

With current technology, commercial genetic testing detects a deleterious mutation in approximately 70 to 80% of individuals with a clinical diagnosis of LQTS (GeneReviews, Familion, GeneDX). Based on the specific gene in which a mutation is identified, the subtype of LQTS can be identified. The three most common subtypes have different profiles of stimuli which trigger cardiac episodes: exercise in LQT1, emotion or startle response in LQT2, and sleep in LQT3 (Wilde & Bezzina, 2005; Schwartz et al., 2001). While genetic testing proves informative for refining the diagnosis of LQTS, diagnosis and treatment are still largely based upon clinical findings. Nevertheless, genetic testing results have direct

implications for at-risk family members in terms of improving risk stratification and guiding preventive interventions.

As genetic testing for LQTS becomes more widely available, a better understanding of the motivations of individuals pursuing genetic testing is critical for genetic counseling and the informed consent process. To date, there is limited literature exploring individuals' motivations for genetic testing in the context of cardiac channelopathies and hypertrophic cardiomyopathy (HCM). Andersen et al. (2008) interviewed 7 individuals regarding the psychosocial aspects of having LQTS. Many of their participants felt relieved to know their status and viewed genetic testing as a necessary piece of information in order to effectively manage their own care as well as maintain the health of their relatives. Similarly, Charron et al. (2002) followed 29 individuals who presented for presymptomatic genetic testing for HCM. After obtaining additional information regarding the test, 19 out of 29 pursued testing. This group found that many of their participants opted to pursue genetic testing because they found uncertainty to be a burden and were concerned about the risks for their children. Additionally, Smart (2010) interviewed 27 individuals undergoing genetic testing for either LQTS or HCM, most who reported doing so in order to reduce uncertainty regarding their diagnosis. Some individuals who pursued testing, however, obtained negative results. In cases of both definite and uncertain clinical diagnoses, these results only served to increase their uncertainty and caused significant frustration.

In comparison, individuals' motivations for pursuing genetic testing have been extensively examined in the context of other disorders, particularly congenital deafness, hereditary cancer syndromes, and Huntington disease. Boudreault et al. (2010) found that most of their 256 deaf participants indicated that they pursued genetic testing out of curiosity regarding the etiology of their deafness. Similarly, Withrow, Burton, Arnos, Kalfoglou, and Pandya (2008) interviewed 44 individuals, including parents of children with deafness, both hearing and deaf, and single adults with deafness. The most common motivation cited was to discover the cause of deafness, however, other motivations included to alleviate guilt and to provide information for their family and for the future.

In contrast, of 135 at-risk individuals who elected to proceed with predictive testing for Huntington disease, the most common motivations cited included to relieve uncertainty, to make childbearing decisions, and to plan the future (Dufrasne, Roy, Galvez & Rosenblatt, 2011).

The motivations to pursue genetic testing in the context of hereditary cancer predisposition syndromes have also been examined in the literature. In the context of hereditary breast and ovarian cancer syndrome, Clark et al. (2000) found that, among 96 participants who opted to proceed with *BRCA1* and *BRCA2* testing, the three most common motivations for pursuing testing included to provide information for family members and/or children, to protect their own health, and to reduce uncertainty. Similar motivations were also noted when individuals were undergoing genetic testing for Lynch syndrome. Both Esplen et al. (2007) and Claes, Denayer, Evers-Kiebooms, Boogaerts, and Legius (2004) noted the three most common motivations were to provide risks for children, to allow for cancer screening and early detection, and to reduce uncertainty. While not found to be one of the most common reason

for pursuing testing, both groups considering testing for Lynch syndrome had some individuals who reported they were doing so because of external motivators, including family members and medical providers. When individuals were considering testing for Li Fraumeni syndrome, Lammens et al. 2010 noted that the most common common motivations included to reduce certainty, provide risks for children, and to allow for cancer surveillance.

Given the relative lack of study in the area of cardiogenetics, this qualitative study of at-risk families provides preliminary evidence of the range and strength of motivations for and against testing in the context of LQTS and other inherited channelopathies.

METHODS

Participants

A total of 50 individuals representing 32 families participated in the study. All participants had a personal or family history of a cardiac event (including syncopal episodes as well as non-fatal cardiac arrests) with a clinical diagnosis of a cardiac arrhythmia or had a family member who died due to SIDS or SUDS. Participants were recruited from three different sources:

- Patients being treated at the Montefiore-Einstein Center for Cardiogenetics (MECC), an interdisciplinary clinic providing evaluation, testing, and treatment (Erskine et al., 2013);
- Individuals responding to a newsletter invitation from the Sudden Unexplained Death in Childhood Program (SUDC);
- Individuals responding to a newsletter invitation from the Sudden Arrhythmia Death Syndromes Foundation (SADS),

Refer to Table 1 for participant demographics and Table 2 for participant diagnosis, presenting family history, and genetic testing status.

Individuals were recruited from the MECC at the time of their medical appointments or 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 after they had received the results of genetic testing, either while at the clinic for a scheduled medical appointment or at an alternative agreed-upon time. Individuals from SADS and SUDC were recruited through liaisons, who either contacted their members through newsletters or by speaking directly to families. If the families agreed to participate, the liaisons provided the research team with their contact information.

All participants provided written informed consent prior to participating in this study and completed a questionnaire providing demographic information. 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.

Procedures

A total of five focus groups, ranging in size from 2 individuals to 4 individuals, were conducted: one focus group from MECC, one focus group from SADS, and three focus groups from SUDC; each focus group included individuals representing either two or three families. All remaining interviews from MECC included the proband as well as at least one family member. Because participants from SADS lived in widely separated communities, most participants were interviewed individually by phone. No thematic differences were noted between sample groups and interview modality.

At least one member of the research team conducted each focus group and individual interview. Interviews were unstructured and followed a “bottom-up” model. This model yields interviews that are structured by participants. Interviews began with the question “Tell me the story of your or your family member’s cardiac episode [or death]”. Only after this question was fully answered did interviewers introduce subsequent questions, which examined attitudes towards genetic testing and sharing of genetic information with family members. Given the “bottom up” method, not all interviews explored further attitudes towards genetic testing. The interview design was developed by research psychologists and, given the open-ended design of the interviews, a set of standardized questions was not created.

Data Analysis

All interviews were audio-recorded and transcribed verbatim. All identifying information regarding participants was removed from the transcripts. Transcripts were then analyzed by the research team using a grounded-theory method (Auerbach & Silverstein, 2003). In this analysis, repeating ideas, or words and phrases used regularly, were first identified. Subsequently, these repeating ideas were organized into themes, which further identified overriding concepts. After at least two members of the research team individually coded each interview, findings were compared to ensure consistency and were subsequently presented to entire research team; any disagreements in coding were addressed and a consensus reached at that time. Both the coded and original transcripts were then reviewed by the genetic counselor for the MECC. The motivations discussed by participants both for and against pursuing genetic testing were identified and are discussed below.

RESULTS

A total of 50 individuals were interviewed: 27 from MECC, 14 from SADS, and 9 from SUDC. Interviews ranged in length from 1.5 to 2 hours. Of the 50 individuals interviewed, 37 specifically discussed their motivations for pursuing genetic testing. A total of five themes and repeating ideas emerged from their responses. Table 3 summarizes the number of participants citing each motivation along with their personal or family history demographics.

Seeking answers for a previously unexplained death or cardiac event

Numerous individuals cited their desire for more knowledge as the primary reason for pursuing genetic testing for themselves or their relatives or for pursuing molecular autopsy

with the medical examiner for a deceased relative. Many of the parents who had experienced the sudden death of a child indicated that they were looking for a reason why their child died, both for a sense of closure and in order to prevent the same thing from happening to their future or surviving children.

“I’ll get a second mortgage on my house if it means doing the right tests to find out what happened to her.” (Female, 42, daughter died of SUDS)

“*There’s some closure now* [that I know what caused the death].” (Female, 57, son and grandson died of SUDS)

“I was very thankful that we did get a diagnosis and an answer. I guess I really didn’t want it to affect at least another young life. I was worried about my daughters but ... it was ... harder to see a very young person have to look forward with something like this hanging over him, and yet it was good to have an answer to what caused [my daughter’s] *death*.” (Female, 67, son died of SUDS)

Providing certainty regarding a diagnosis

Another common theme was seeking information regarding a diagnosis in order to relieve uncertainty. Several participants stated that, even if the results of genetic testing revealed a hereditary arrhythmia syndrome in the family, it was helpful to give a name to the condition and subsequently obtain more information regarding their risks.

“Yeah, it was a relief of just knowing I guess and wondering back and forth what’s going on. It was a relief of knowing.” (Male, 31, wife died of SUDS, children affected with LQTS)

“I think the fuzziness, I would rather have a definite yes or no. Do you think that genetic testing is important the way it comes out, because like I said, to me it is the piece of the puzzle, you need all the facts you can come up with.” (Female, 48, niece and nephew had cardiac arrests, affected with LQTS)

“I really wanted to know, I definitely wanted to know...I already knew I had it, I knew that was caused by some mutation, and all that genetic testing would do would be to, you know, narrow it down and give me more information.” (Female, 34, family history of LQTS)

Some participants expressed disappointment in having a negative test result. While a negative genetic test result was viewed as good news, it did not necessarily provide clarity in terms of diagnosis.

“I’m happy that’s available. And I know that if it had come back the other way, we would have had concrete answers.” (Female, 38, daughter had negative LQTS testing)

“I wouldn’t say it would be nice to have, it’s just more clear of a definition.” (Female, 17, negative LQTS testing following syncopal episode and possible prolonged QT)

Preventive measures and future health management decisions

Many participants stated that they pursued testing in order to take preventive health measures or to aid in future decision making. Many parents who had experienced the death of a child felt empowered in taking preventive measures to avoid a recurrence with another child or family member.

“I’m grateful that I did find out, that they were able to catch it, and did the testing and I’m able to help them at a very early age with the medicine and stuff.” (Male, 31, wife died of SUDS, children affected with LQTS)

“I was happy to hear that she was gonna be tested because I’m saying that an ounce of prevention is better than a pound of cure, and I’m very glad to know that they found out before it was too late.” (Female, 57, son and grandson died of SUDS)

“Possibly give us an answer again because we had the other two girls. You know we were on board for anything.” (Female, 33, daughter died of SUDS)

Other individuals indicated that they chose to pursue genetic testing in order to make future health decisions for themselves and their families, especially with regard to ICD implantation or lifestyle changes.

“Now I will know for sure beyond the shadow of a doubt because... my little boy who was five, we treated him like he had it just because... his numbers were borderline.” (Female, 51, daughter died of SUDS, diagnosed with LQTS)

“It’s scary, you know and you’re always like, ‘Oh my God, can I do this, can I not do that?’ Always with caution, but I’d rather know so that way I can have caution and prevent it from happening instead of not knowing, and all of a sudden you’re just on the floor, you know.” (Female, 21, family history of cardiac arrest and prolonged QT)

“I still think the genetic testing is important because it offers some kind of foundation there, it is just something else to look at... and work with to make a solid decision [about healthcare decisions].” (Female, 48, niece and nephew with cardiac arrest)

Concern about family members and future generations

One of the key motivators for parents to pursue testing was to provide valuable health information for their children.

“It would be all for my kids. That’s the reason I’m sitting here today, not because of me, cause I already had my time with this.” (Female, 30, personal and family history of cardiac arrest)

“To tell... what my chances were of passing it on to my children.” (Female, 25, mother died of SUDS, diagnosed with CPVT)

“If it comes back positive, I would be concerned for my other children and grandchildren- it would be a great concern for me. Then I would have to have them tested.” (Female, 57, son and grandson died of SUDS)

This sentiment also extended to individuals of childbearing age, many of whom indicated that they would take their genetic results into account when considering their reproductive options.

“My cardiologists have recommended that we go through IVF and PGD pre implantation genetic [diagnosis]... so that we can guarantee that we have a child without long QT but it’s expensive, and my husband would rather pay the money and just make sure that we don’t have a child with long QT.” (Female, 34, personal and family history of LQTS)

“[My son] said if the genetic test came back positive, he would not have children. He felt that he just couldn’t do that... because he would be afraid that he would be passing the genetic marker on, and he definitely didn’t want to do that.” (Female, 58, son with cardiac arrest and clinical diagnosis of Brugada syndrome)

“And I decided that I am going and try to have another baby. That is when we started... really thinking into it hard, and that is when I discovered it was genetic.” (Female, 45, daughter and brother-in-law died of SUDS, husband diagnosed with LQTS)

External motivators

In addition to the above self-motivated reasons to pursue genetic testing, external pressures also served as a motivator. Some participants indicated that they were only pursuing genetic testing because their relatives stressed the importance of pursuing testing.

“I never even wanted to do the genetic testing. I mean I did it because, for peace of mind for my mother more so, but I really never thought that I had reason to do any of this testing because I never felt that I had heart issues.” (Female, 21, father died of SUDS)

Other participants indicated that genetic testing was their doctor’s recommendation, and that they viewed it as on par with other diagnostic testing that their doctor ordered.

“I didn’t know what it was, so I just went ahead... I just took her word. I just felt like whatever she wanted to do would make me better.” (Female, 17, prolonged QT)

“The cardiologist had come in at that time and said ‘We think you have had a cardiac arrhythmia episode and this can be a family type thing’ and that’s kind of really all it was said... we really didn’t know anything about it.” (Female, 52, son and daughter died of SUDS)

Reasons for choosing not to be tested

While all of the participants in our study for whom genetic testing was appropriate and insurance coverage was obtained pursued testing, many indicated that some of their relatives had not pursued testing and evaluation for LQTS. When outlining the perceived reasons why their relatives have not pursued testing, numerous participants voiced their concern regarding their decision. Having pursued genetic testing to obtain risk information for

themselves and their families, they felt that many relatives were in denial about the severity of the condition and therefore chose to not continue with testing.

“I don’t think [my brother] has taken any of this seriously... I feel there is some denial going on, and whenever I have gotten up there, you don’t talk about [long QT syndrome] at all.” (Female, 48, niece and nephew with cardiac arrest)

“It makes me feel bad because I feel that [my daughter] is gone, and there is a reason she is gone and it is not taken seriously... it’s like they are in denial, I don’t know why they think if they don’t address it then, it doesn’t exist.” (Female, 51, daughter died of SUDS)

“I’m surprised because they haven’t been tested ... So I asked her, ‘What’s going on, are you going to do the test?’ and she said ‘Oh I don’t think they have it... my kids are strong, they look healthy, they don’t need it and that’s it.’ So I guess I’m not sure if it comes from not wanting to face it, or being naïve.” (Male, 31, wife died of SUDS)

Other participants reported that their relatives might be afraid of learning that they or their children may be at risk for sudden death.

“Everybody was really afraid, I brought out all these fears in them about their own health and then their children’s health.” (Female, 46, daughter with cardiac arrest)

“My brother to the best of my knowledge has never been tested and more than likely is not interested in doing so... It’s just easier to not know.” (Female, 67, son died of SUDS)

“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 coulda gave it to them.” (Female, 38, family history of sudden death, personal history of LQTS)

Still other participants report that their relatives have not been tested because of misconceptions or lack of knowledge.

“[My grandmother’s sister] told me of her son fainting 4 or 5 times and how the last time he fainted it was the longest that he was out, and then she followed with ‘But I don’t think that’s our family.’ And I was like: ‘Do you hear yourself?’” (Female, 29, husband died of SUDS)

“My sister refuses to have her children... tested because her husband who is a doctor thinks that they [diagnosing physicians, diagnosis, and affected family members] are all ridiculous.” (Female, 46, daughter with cardiac arrest)

In addition to the above reasons, two participants reported that they discussed the situation with their family members and that their relatives made a conscious decision that they were not interested in knowing their genetic status and the risks associated with testing positive.

“She has never been tested, and she is not married, and she doesn’t have children and she said... ‘If something happens it happens’... she doesn’t want to be tested.” (Female, 46, daughter with cardiac arrest)

“We finally talked about it and he said he didn’t want to know. He knows he has got a genetic disposition.” Female, 58, husband had cardiac arrest

DISCUSSION

An individual’s motivation to either pursue or not pursue genetic testing when there is a personal or family history of a genetic disorder is unique to each individual’s personal experience, health philosophy, and psychosocial factors (Armstrong et al. 2000; Cohen, Stolerma, Walsh, Wasserman & Dolan, 2012), however, recurrent motivations do emerge. In the context of inherited cardiac arrhythmia syndromes or a family history of sudden death, 37 of our participants discussed their motivations for pursuing genetic testing. These motivations include to: seek answers for a previously unexplained death; provide certainty regarding a diagnosis; make health management decisions and attempt to avoid poor outcomes; provide information for children and other family member; and comply with recommendations from family members or health care providers. Although inherited cardiac arrhythmias present unique health concerns, many of the ideas expressed by these participants are echoed in the existing genetics literature, as detailed below.

Providing certainty regarding a diagnosis

One major theme in the literature is the desire to obtain more information regarding the condition and defining genetic risks; many of our participants identified a desire for a definitive diagnosis and to relieve uncertainty as significant motivators for genetic testing. In the context of both LQTS and hypertrophic cardiomyopathy (HCM), the need to relieve uncertainty about one’s mutation status is one of the prime motivators (Charron et al., 2002; Smart, 2010). Among our participants, it was the most commonly cited motivation for individuals with a personal or family history of cardiac diagnosis or syncope and the least commonly cited motivation for individuals with a family history of SUDS, which may suggest that genetic testing is becoming a significant factor in certain cardiogenetic diagnoses. This desire for certainty, however, is limited by current testing technology. In individuals with a clinical diagnosis of LQTS, approximately 30% will not have a detectable mutation (Genereviews, Familion, GeneDX). A negative genetic testing result, therefore, does not rule out the condition, nor does it allow for genetic identification of at-risk relatives.

In the context of congenital deafness, most individuals reported that their motivation was fueled by curiosity about the etiology of their or their family member’s deafness rather than by an interest in using the information to make healthcare or reproductive decisions (Boudreault et al., 2010; Withrow, Burton, Arnos, Kalfoglou & Pandya, 2008). It is important to note, however, that non-syndromic deafness rarely has long-term health implications or specific treatments tailored to genetic testing results. A similar sentiment has been expressed by individuals pursuing pre-symptomatic screening for Huntington’s disease, with a majority of people pursuing genetic testing doing so in order to relieve

uncertainty about their disease status (Dufrasne, Roy, Galvez & Rosenblatt, 2011; Novak & Tabrizi, 2010).

Preventive measures and future health management decisions

In contrast to congenital deafness and Huntington's disease, in the case of LQTS, knowledge of a genetic predisposition in an unaffected individual can shape lifestyle modifications, screening protocols, and surgical interventions in order to reduce the risk of mortality. This idea was mentioned both by individuals with a personal or family history of a cardiac diagnosis and individuals with a family history of SUDS. Individuals with a diagnosis have likely been counseled as to the benefits of preventative treatments, while individuals with a family history of SUDS are concerned about another family member dying suddenly.

As is the case with LQTS, there are well established guidelines for the management of hereditary breast and ovarian cancer syndrome (HBOC) and Lynch syndrome (NCCN clinical practice guidelines in oncology). As a result, individuals pursuing genetic testing for Lynch syndrome and HBOC consistently report the availability of specific risk-reducing guidelines as one of their motivations to pursue genetic testing (Clark et al., 2000; Esplen et al., 2007). Similar recommendations exist for the treatment of LQTS, which include lifestyle modifications, beta blocker medication, and ICD implantation (Goldenberg & Moss, 2008; Wilde & Bezzina, 2005), and this is reflected in our participants' interest in pursuing genetic testing to make future medical management decisions.

Concern about family members and future generations

Because of the availability of treatment, participants in cancer predisposition studies, like our participants, reported that they were seeking information in order to clarify both their and their family members' risks for having or developing a life-threatening condition. Of all motivations for pursuing genetic testing, this was the most common motivation cited overall. This motivation is also cited commonly in both the cancer and cardiogenetics literature. Parents with a family history of cancer are especially likely to pursue genetic testing in order to obtain a more accurate risk assessment for their children (Clark et al., 2000; Claes, Denayer, Evers-Kiebooms, Boogaerts & Legius, 2004; Lammens et al., 2010). In individuals with a cardiogenetic diagnosis, they often desire information for their own medical management (Andersen et al., 2008), however, more commonly, individuals pursuing testing are doing so for the sake of their family members. After discovering the family's disease-causing mutation, this information can be used to determine their family members' risks and allow them to pursue informative genetic testing (Andersen et al., 2008; Charron et al., 2002) as well as allow mutation positive individuals to take measures to protect their own health (Smart 2010).

Seeking answers for a previously unexplained death or cardiac event

Both testing for hereditary cancer syndromes and cardiac channelopathies can bring up significant psychosocial issues (Erskine et al. 2013). In most, if not all cases, individuals presenting for counseling have personally experienced a life-threatening event or have experienced the death or near death event in a loved one. This study demonstrates a distinction between cancer and a sudden cardiac death must be made, as additional

psychosocial factors must be taken into account given the often unexpected nature of death attributed to the chanelopathies.

The idea of sudden death and grief as a motivator for genetic testing, however, is one that has not been extensively explored in the existing literature. Especially in cases where the death of a child is sudden, parents often attempt to find meaning in the death (Keese, Currier & Neimeyer, 2008; Wheeler, 2001) and if they are unable to do so, their grieving process can become more complicated (Currier, Holland & Neimeyer, 2006; Keese et al., 2008). In the case of SIDS specifically, Davis et al. (2000) found that over half (68%) of parents searched for meaning in their child's death; most parents were unable to find meaning, and as a result, the loss was viewed as more painful. This search for meaning may include trying to obtain an official cause of death, to make sense of the death in the context of a family's life and belief systems, or to find any positive effects from the death (Keese et al., 2008). When families present for genetic testing following a sudden death, therefore, it is important to take their level of grief and search for meaning into account. While genetic testing may serve as an important step in an individual's search for meaning, both in an effort to understand what happened to their relative as well as to use this information to save other family members, motivations for testing should be discussed.

External motivators

Many participants indicated that the main reason they initiated genetic testing was because of pressure from either a physician or family member. While genetic information can prove important to both unaffected family members as well as a treating physician, it is essential to explore the individual's beliefs and desires for genetic testing. According to the American Medical Association, the informed consent process must include a discussion of the individual's diagnosis, the testing that is being performed and the reason for testing, and the benefits and risks of both pursuing and not pursuing testing (American Medical Association). Therefore, in order to provide true informed consent, an individual's prior knowledge and motivations to pursue testing must be addressed to ensure an understanding of the condition and the implications for themselves and family members by electing to accept or decline genetic testing. As such, it is essential to be mindful of both the internal and external motivators for individuals during the genetic counseling session and during the informed consent process.

Reasons for choosing not to be tested

The vast majority of the participants in this study elected to pursue genetic testing, therefore, motivations to not pursue testing were not extensively explored. Some participants, however, discussed their perceptions of why family members did not pursue testing, identifying denial, fear, and misinformation as primary reasons. These perceptions, while strictly speculative, are largely consistent with the existing body of literature. In studies assessing the motivations of individuals to pursue cancer genetic testing, individuals who opted out cited concerns about effects on the family, not being able to cope with the results, fear of the results, not perceiving the advantages of testing, and insurance or employment discrimination (Balmana, Stoffel, Emmons, Garber & Syngal, 2004; Lammens et al., 2010). Of note, since the passage of the Genetic Information Nondiscrimination Act (GINA) (<http://>

www.eeoc.gov/laws/statutes/gina.cfm) in 2008, most individuals are protected against employment and health insurance discrimination based on genetic information, but not life or long-term disability insurance discrimination.

A limitation of the existing literature examining the reasons against pursuing genetic testing is that most of the individuals declining testing had presented for genetic counseling. These studies, however, were not able to examine the motivations of individuals who never presented for pre-test counseling. Hadley et al. (2003) speculated that individuals did not pursue genetic counseling because of denial of high risk, inability to afford testing, skepticism of the benefit of testing, or unawareness of the availability of testing. The manner in which relatives are notified of family member results as well as the information they receive could also play a part in their desire to pursue or not pursue testing.

Study Limitations and Research Recommendations

The results of the study are limited by the method of participant recruitment and possibly by interview method. All participants were self-selected and pursued genetic testing, if appropriate. Participants who volunteered from SUDC and SADS were aware that the study involved experiences with genetic testing in the context of sudden death. Participants from the MECC were receiving treatment, kept their appointments, and followed-up with their results; individuals who were not interested in genetic testing may not have kept appointments with the clinic or declined participation in the study. By nature, our participants were not only motivated to learn more about their condition and possible genetic contributors but were also more likely to have experienced the sudden death of a family member as a consequence of LQTS. Additionally, the manner of interview was not consistent for every participant: some were interviewed individually, some with a focus group of people they did not know, and some with a focus group with family members. As a result of multiple individuals being interviewed at once, responses could have been influenced by other individuals present.

While this study interviewed 50 individuals, it does not have sufficient data to discern thematic differences related to age, gender, ethnicity, or education level. Further study with more structured interviews in diverse populations could provide further insight. Additionally, because all individuals interviewed pursued genetic testing, their reports of motivations against pursuing testing were purely speculative. While challenges exist in recruiting individuals who decline genetic testing, further discussion would be essential to examine their motivations against testing and as well as their knowledge and attitudes towards genetic testing in order to eliminate barriers to accessing genetic services and improve medical decision making and treatment.

CONCLUSIONS

Knowledge of patients' motivations for pursuing genetic testing is an integral part of the genetic counseling process. Over the course of a session and while obtaining informed consent, an individual's personal experiences and attitudes about genetic testing come to the surface and must be addressed accordingly. Having a body of literature outlining some common motivations for pursuing testing can allow for the anticipation of certain issues that

may arise as well as serve to help to normalize an individual's feelings about genetic testing. This knowledge can also provide a basis for addressing a family's fear, denial, and need for information may help them make informed choices about genetic testing. Additionally, the ideals of autonomy and non-directiveness are central to the genetic counseling profession. During the genetic counseling session, therefore, it is essential to separate an individual's own motivations from possible external pressures in order to obtain informed consent and providing effective counseling.

As we learn more about the preventive genomic medicine, genetic testing is becoming more available and has many implications for personalized medicine. In conjunction with medical experts, genetic counselors, with their knowledge of genetic concepts and patient-centered counseling, will be essential in continuing to translate this new information into routine clinical practice.

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Table 1

Participant Demographic Information

Characteristics	Number of Participants (N=50)
Sex	
Male	11
Female	39
Geographic Region	
Northeast	39
Midwest	6
South	3
West	2
Race	
White	42
Black/African-American	7
Biracial	1 (White Asian)
Ethnicity	
Non-Latino/Hispanic	40
Latino/Hispanic	10
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
Single	15
Widowed	3
Cohabiting	2
Separated	2
Divorced	1
Annual Household Income	
<25,000	6
\$26,000-\$50,000	6
\$51,000-\$80,000	10
>\$80,000	21
Refused	2

Characteristics	Number of Participants (N=50)
Unknown	5
Age	
<20	1
20-30	9
31-40	9
41-50	14
51-60	11
>60	6

Table 2

Participant Diagnosis, Family History, and Genetic Testing Status

Cardiac Status Characteristics	Number of Participants
Diagnosis	
LQTS	26
Unknown	13
Brugada Syndrome	4
SQTS	2
CPVT	2
ARVD	1
Presenting Feature	
SIDS/SUDS	29
Cardiac arrest- non fatal	10
Other	7
Syncope/arrhythmia	4
Relationship	
Affected relative	16
Unaffected at-risk relative	16
Unaffected/ not at-risk (unaffected parent/spouse)	12
Proband	6
Genetic Testing	
Yes	34
positive result	15
negative result	13
VUS	11
unknown result	5
No	14
Unknown	2

Table 3Themes Identified Regarding Motivations for Pursuing Genetic Testing and Participant Demographics ($n=37$)

Theme	# Participants	Participant Characteristics		
		Personal history of cardiac diagnosis or syncope	Family history of cardiac diagnosis or syncope	Family history of SUDS
Concern about family and future generations	15	4	2	9
Preventive measures	12	3	2	7
External factors	9	1	3	5
Relieve uncertainty	9	4	3	2
Seeking answers	6	0	0	6

Note: Some participants indicated multiple motivations for testing