Disclosure of diagnosis to at-risk relatives by individuals diagnosed with hypertrophic cardiomyopathy (HCM)

Janella Hudson^{1,2} · Amy C. Sturm³ · Lisa Salberg⁴ · Simone Brennan⁵ · Gwendolyn P. Quinn^{1,6,7} · Susan T. Vadaparampil^{1,6}

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Abstract

Hypertrophic cardiomyopathy (HCM) affects 1 in 200 people and is the most common cause of sudden cardiac death in the young. Given that HCM usually is inherited in an autosomal dominant pattern, an HCM diagnosis has implications for biologically related family members. The purpose of this study was to explore probands' disclosure of an HCM diagnosis with these biologically related, at-risk family members. An online survey was posted on the website of the Hypertrophic Cardiomyopathy Association (HCMA), an advocacy and support group for HCM patients and their families. Descriptive statistics were used to summarize responses to closed-ended questions and demographics. Using an iterative content analysis with the constant comparison approach, we analyzed the responses to open-ended questions inquiring about the nature and role of disclosure communication with at-risk relatives. A total of 315 individuals with a self-reported diagnosis of HCM completed the survey. Most participants (98%) disclosed their diagnosis to at-risk family members. Sixty-four percent disclosed to family members less than 1 year after diagnosis. Participants also disclosed potential treatment options (74.6%) and the emotional impact of the diagnosis (39%). HCM specialists were ranked by participants as being the most helping in explaining the benefit of genetic counseling, while genetic counselor were ranked as least helpful. Emerging themes address the need to encourage screening and genetic testing among family members and to identify external educational resources for use during the disclosure process. Importantly, our study found that the process of disclosure varies based on individuals' experiences and family communication dynamics. However, almost all participants expressed the importance of disclosing the diagnosis of HCM as well as the importance of being screened and expressed needs for additional support during the disclosure process.

Keywords Hypertrophic cardiomyopathy · HCM · Disclosure · Diagnosis · Family members · At-risk relatives · Genetic

✉ Janella Hudson
janella.hudson@gmail.com

¹ Health Outcomes and Behavior Program, H. Lee Moffitt Cancer Center and Research Institute, Tampa, FL 33612, USA

² Tampa, USA

³ Wexner Medical Center and Dorothy M. Davis Heart and Lung Research Institute, The Ohio State University, Columbus, OH 43210, USA

⁴ Hypertrophic Cardiomyopathy Association, Denville, NJ 07834, USA

⁵ School of Medicine, Wayne State University, Detroit, MI 48201, USA

⁶ Department of Oncologic Science, Morsani College of Medicine, University of South Florida, Tampa, FL 33612, USA

⁷ Department of OB-GYN and Population Health, School of Medicine, New York University, New York, NY 10016, USA

Introduction

Hypertrophic cardiomyopathy (HCM) is a common inherited heart disease characterized by left ventricular hypertrophy that can lead to left ventricular outflow tract obstruction, causing a variety of symptoms and morbidities including chest pain and heart failure (Qintar et al. 2012). The electrical activity of the heart may also be affected, which can lead to palpitations, syncope, and malignant arrhythmias, causing sudden cardiac death (Wexler et al. 2009). A large number of genetic studies have established HCM as a genetically heterogeneous disease, mainly caused by mutations in genes encoding components of the sarcomere. Approximately 50% of affected individuals will have identifiable mutations, with the majority attributed to inherited germline mutations in *MYH7* and *MYBPC3* (Maron et al. 2014). HCM typically follows an autosomal dominant Mendelian inheritance pattern with variable

expression and age-related and incomplete penetrance (Shah 2016). As one of the most common inherited cardiovascular diseases, HCM occurs in about 1 in 200 people (Semsarian et al. 2015), with the most severe manifestations of HCM in younger people (Mayo Clinic 2016). However, HCM has a comparatively low diagnostic prevalence in clinical practice (Maron et al. 1994), suggesting significant populations of individuals with HCM are undiagnosed with the condition, including at-risk relatives who may be unaware of their HCM risk. HCM carries a low yearly risk of overall mortality at 1.3 and .7% per year for sudden cardiac death (Leonardi et al. 2010). Frequently, sudden cardiac death is the only symptom of HCM.

In the USA, commercial genetic testing for HCM via next-generation sequencing panels is offered by a number of molecular genetic testing laboratories. In other countries, including Argentina, Brazil, Cuba, and Mexico, genetic testing is done in non-commercial labs and can even be covered by health insurance. Genetic testing, when informative, allows cascade genetic testing to commence through a family, identifying those family members who are at risk to develop HCM and require cardiac screening from those who do not.

While no cure exists, screening for and diagnosis of HCM allows family members to engage in risk-reducing measures to manage the condition and plan for any subsequent manifestations of the disorder, including medical therapy, withdrawal from intense competitive sports or strenuous exercise, and identification of risk for malignant arrhythmias with subsequent placement of an implantable cardioverter defibrillator (ICD) when necessary.

An at least 3-generation pedigree, or family history, is recommended for all patients with HCM. In the USA, genetic counseling is recommended for all patients and families with HCM, and in addition, genetic testing should be considered for the one most clearly affected patient in the family to facilitate cascade genetic testing and family screening (Writing Committee Members 2011). Conversely, European guidelines direct all index patients diagnosed with HCM be referred for genetic testing, with no specifications for severity of the disease (Charron et al. 2010). During genetic counseling for HCM, it is recommended that individuals diagnosed with HCM disclose their diagnosis to their biologically-related at-risk family members, so that they may learn about their increased risk and consult their own health care provider for clinical screening, and if a known familial mutation has been identified, genetic testing. However, receiving a new diagnosis of any potentially life-threatening disease, including HCM, triggers a myriad of medical and psychosocial challenges. Disclosing a potential genetic risk to relatives may trigger concerns such as privacy, and may also introduce complex family communication issues (Forrest et al. 2003; Gaff et al. 2010; Gallo et al. 2009; Galvin and Grill 2009).

Unlike the robust literature exploring disclosure and family communication surrounding genetic risk in the context of breast cancer (Forrest et al. 2003; Hughes et al. 2002; Olson and Cheung 2010; McGivern et al. 2004; Himes et al. 2015), relatively little research has explored this issue in HCM (Geelen et al. 2011a; Batte et al. 2014). Studies examining the disclosure patterns of patients diagnosed with hereditary breast and ovarian cancer have described a complex process occurring over time (Gaff et al. 2007), influenced by pre-existing family dynamics, and the proband's reaction to perceived risk (Forrest et al. 2003; Claes et al. 2003). Disclosure of genetic risk to at-risk family members does not necessarily occur immediately after diagnosis, as families and probands are often preoccupied with potentially traumatic experiences related to diagnosis. Initial discussions of diagnoses often prioritize the health of the proband and are intended to elicit support from family members, with lesser priority given to discussions concerning genetic risk occurring over time (Forrest et al. 2008). HCM includes the possibility of sudden death as the first sign of disease, which makes its disclosure unique when compared to genetic risk for other diseases such as cancer. Such a risk may alter perceptions and deliberations of timing, approach, and context of disclosure. We explored patterns of disclosure of the diagnosis of HCM probands to their at-risk family members, including preparation related to the disclosure of genetic risk and experiences related to genetic counseling uptake.

Materials and methods

The lead author worked with the Hypertrophic Cardiomyopathy Association (HCMA) to collaboratively develop an online questionnaire that assessed members' experiences concerning disclosure of diagnosis to biological families. The HCMA is an international advocacy group devoted to "improving the lives of those with HCM, preventing untimely deaths and advancing global understanding." (HCMA 2016). In addition to its advocacy work for HCM patients, the HCMA confers a "Center of Excellence" (COE) status to HCM programs that demonstrate expertise in patient care and research. The application process for the COE designation includes a site visit, interviews with key clinicians and institutional leaders, and a review of the department as the HCMA seeks to determine if sufficient individual and institutional support are present. Additional details concerning the COE application process can be found at www.4hcm.org. While the HCMA's membership is international, most participants reside in the USA. Website users who self-reported an HCM diagnosis (clinical or genetic) were eligible for the study. As many participants may not have undergone genetic testing, inclusion criteria included registered HCMA website users who self-reported a diagnosis of HCM.

Survey instrument

The questionnaire consisted of 9 open- and 17 closed-ended questions. In the first part of the questionnaire, participants provided answers to closed-ended questions regarding demographic information, information about their diagnosis, and their decisions about whether to disclose their diagnosis to biological family. Participants were further asked to specify what information they shared, which family members they disclosed to, whether they sought support or guidance when considering disclosure, whether they utilized any external resources for assistance during disclosure, and if they received genetic counseling. Open-ended questions further probed the disclosure process, motivation, or considerations when deciding whether to share the diagnosis, as well as any recommendations they would give about the disclosure process to other HCM individuals. Participants were also asked to provide feedback about how they managed HCM in their daily lives (Tables 1, 2, 3).

Participants

After receiving IRB approval and a waiver of written consent from Wayne State University, the HCMA's founder posted an

invitation to participate on the Web message board. In addition, an email invitation was sent to the approximately 1600 registered users who previously reported a diagnosis of HCM, indicated a willingness to participate in future studies, and provided an email address. A follow-up email invitation was sent 4 weeks later. Participants who clicked on the survey were directed to a cover letter detailing the study's purposes and parameters of informed consent. Participants were advised that members under the age of 18 required parental consent to participate and that clicking the survey launch button indicated their consent to participate.

Analysis

Descriptive statistics were used to summarize closed-ended questions, including data related to diagnosis preparation methods, genetic counseling uptake, and demographics. Open-ended responses were coded using content analysis and the constant comparison approach. First, the researchers identified emerging themes and interpretively constructed an understanding of the dynamics involved when disclosing HCM to biologically related family members. Simultaneously, JH and SB analyzed responses in an iterative process in which a code list was constantly refined as new

Table 1 Participant demographic and genetic counseling uptake

Demographic or characteristic	No. of participants (<i>N</i> = 315)	No genetic counseling	Received genetic counseling
Sex			
Female	146	91 (61.5%)	55 (37.2%)
Male	155	72 (45%)	83 (51.9%)
Race			
White	278	150 (52.6%)	128 (44.9%)
Minorities (non-white)	26	16 (61.5%)	10 (38.5%)
Ethnicity			
Hispanic or Latino	9	6	3
Education			
Some HS/high school diploma equivalent/	25	18 (69.2%)	7 (26.9%)
Some college/bachelor's degree/graduate or	176	95 (52.8%)	81 (45%)
prof. degree			
Other	16	9 (56.3%)	7 (43.8%)
Household income			
US\$ < 20,000	13	6 (46.2%)	7 (53.8%)
US\$ > 20,000	229	126 (53.6%)	103 (43.8%)
Age groups ($\bar{m}$ = 51.25, sd = 15.0)			
≤ 45 years old	83	42 (50.6%)	41 (49.4%)
Age 46 and older	189	111 (56.6%)	78 (39.8%)
Received care at HCM Center of Excellence			
No	125	87 (68.5%)	38 (29.9%)
Yes	131	53 (39.8%)	78 (58.6%)
Unsure	50	28 (56%)	22 (44%)

Table 2 Participants' disclosure to at-risk family members

1.1 Content of participant's disclosure		
"What information did you share? Choose all that apply"		
Diagnosis	309	98.1%
Treatment options	235	74.6%
Emotional impact	123	39%
Other	41	13%
1.2 Family members to whom probands disclosed diagnosis		
"Who have you told? Please check all that apply"		
Mother	198	62.7%
Father	139	44%
Sister	202	63.9%
Brother	214	67.7%
Cousins	208	66%
Aunt/uncle	177	56.2%
Grandparent	70	22.2%
Other	113	35.9%
1.3 Probands' preparation prior to disclosure of diagnosis		
1.4 "Did you seek external guidance or support before sharing this information?"		
Yes	67	21.2%
No	239	75.6%
1.5 Resources consulted in preparation of probands' disclosure of diagnosis		
"What source did you consult for guidance or support? Choose all that apply."		
Physician	55	17.5%
Nurse	16	5.1%
Genetic counselor	16	5.1%
Advocacy/support group	19	6.0%
HCMA phone call	27	8.6%
Other	12	3.8%

phenomena emerged. Codes were subsequently collapsed into categories with shared properties and results were presented thematically. Inter-rater reliability was reached through consensus gaining.

Results

Demographics

Three hundred fifteen individuals completed the questionnaire (Table 1). Participants overwhelmingly reported that they shared their diagnosis with biological family members, most often to first-degree relatives (Fig. 1).

Quantitative findings

The majority of participants reported disclosing their diagnosis to at-risk family members (98%). (Table 2). Disclosure

most often occurred immediately after diagnosis, with 64% reporting disclosure less than a year after diagnosis and 8.5% reporting disclosure within a year after disclosure (Fig. 2). In addition to disclosing the diagnosis of HCM, participants also reported disclosing potential treatment options (74.6%) as well as the emotional impact of the diagnosis (39%). Probands disclosed their diagnosis most often to a first-degree relative (Fig. 1), and most often to their mother (62.7%), sister (63.9%), brother (67.7%), and cousins (66%) (Table 2). Twenty-one percent (21.2%) of participants sought external guidance or support prior to their disclosure, with 17.5% of those participants seeking support from a physician and 5% consulting a genetic counselor. Genetic counseling uptake rates were highest among males (58.6%) and those who received care at an HCM Center of Excellence (Table 1).

Qualitative findings

Responses ranged from brief comments such as, "It is my understanding it is hereditary," to more elaborate comments. Participants articulated a sense of urgency in making others aware of HCM and conveying the possibility of sudden cardiac death for those with HCM yet unaware and undiagnosed. Throughout the related responses, the dynamics of family communication were revealed with several participants indicating that informing their family members was a foregone conclusion, as their families readily shared medical information. Conversely, others' decisions to inform their families revealed potentially underlying family tensions.

I shared it to inform them it's genetic. Our grandmother and aunt are believed to have this. Only they were never diagnosed correctly back then. We had a 16-year-old cousin drop dead at the bus stop ... again ... diagnosed as enlarged heart. It's now through me this disease has been brought out to light correctly.

I wanted to be sure that they could prevent the worst case scenario and be educated by my symptoms; Survival of cardiac arrest as a result of (this) condition.

To find out where it came from and who else is at risk. Basically they all ignored me.

Encourage testing and screening

Beyond discussing HCM's signs, symptoms, and genetic cause, many participants encouraged testing to determine the presence of the disease and/or its genetic predisposition. Family dynamics played a role in forming the decision to share and participants' perspectives regarding screening modalities varied. Some participants offered a generic

Table 3 Survey questions

1. When did you receive the diagnosis of HCM?
yyyy
2. Please indicate your gender
Female
Male
3. What is your date of birth?
mm/yyyy
4. Please indicate your race
White
Black
Hispanic
Asian
Middle Eastern
Native
Mixed
Other
5. Please indicate the highest level of education that you completed
Some HS
HS
Some college
Bachelor's
Grad Degree
Other
6. Please indicate your household income level
< 20 K
20–40 K
40–60 K
60–80 K
80–100 K
100 K
No response
7. Have you shared your diagnosis with biologically related family members?
Yes
No
8. Who have you told? Please check all that apply
Mother
Father
Sister
Brother
Cousins
Aunt/uncle
Grandparents
Other
9. What information have you shared? Choose all that apply
Diagnosis
Treatment
Emotional impact
Other
10. Why did you share this information?
[Open text]
11. Did you seek external guidance or support before sharing this information?
Yes
No
12. What source did you support for guidance and support? Choose all that apply
Physician
Nurse
Genetic counselor
Advocacy/support group

Table 3 (continued)

- HCMA phone call
Other
13. Did you prepare for this conversation with any of the following steps?
• I did not prepare
• Wrote out what I planned on saying
• Sought information online and printed or copied information there
• Talked it out with a friend
• Talked it out with a healthcare professional
• Talked it out with an advocacy organization
14. Provide the year that you shared this information
yyyy
15. In 3–4 sentences, tell us what advice would you give to someone else (who has also been diagnosed with HCM) about sharing this information with biologically related family members? Would you recommend they follow what you did?
[Open text]
16. In 3–4 sentences, tell us what advice would you give to someone else (who has also been diagnosed with HCM) about sharing this information with biologically related family members? Would you recommend they follow what you did?
[Open text]
17. Is there anything else you think is important for us to know?
[Open text]
18. In 3–4 sentences, how has receiving the diagnosis of HCM most affected your life?
[Open text]
19. In 3–4 sentences, how do you manage the unpredictable nature of HCM in your daily life?
[Open text]
20. Do you currently receive care at an HCM Center of Excellence/HCM program as recognized by HCMA?
Yes
No
21. Have you received genetic counseling?
Yes
No
22. What resources did you find helpful in explaining your diagnosis (choose all that apply)?
Physician
Nurse
Genetic counselor
Advocacy/support group
HCMA phone call
Other
23. What resources did you find to be most helpful in explaining the importance and/or benefit of genetic counseling? [Rank in order of most helpful. 1 = Most helpful, 5 = least helpful]
Physician
Nurse
Genetic counselor
Advocacy/support group
HCMA phone call
Other

recommendation to be checked by a cardiologist and others advocated genetic testing, and many described a screening process consisting of both an electrocardiogram and genetic testing. Other participants endorsed screening using echocardiography but expressed concerns regarding genetic testing

Fig. 1 To whom did you disclose?

and the potential value for their family members, and larger quality of life dilemmas raised by test results.

Because I had a son that had HCM who died when he was 9 years old, I thought it would be important for my family and their kids to go to a cardiologist and get checked out.

Give them information about genetic testing as well as physical exam in order to get as early a warning as possible of having the DX.

Encourage genetic testing but get your doctor to write letter stating the benefits of the genetic testing vs annual EKGs.

Share a sense of concern

Several respondents did not write about HCM as a potential genetic condition or the need to educate/advocate for testing, but simply stated they shared their diagnosis out of a sense of concern for others and/or to engender compassion for themselves. Concern for others was evidenced in the comments that referred to others' welfare, general interest, and benefit. Again, children were often the primary focus of concern, particularly as children are often engaged in sports or other physical activity. In terms of concern for themselves, many respondents indicated they felt more in control of their emotions and health when they could discuss their fears. Others felt it imperative to alert their families to symptoms that may indicate

Fig. 2 Time elapsed between diagnosis and disclosure

they needed immediate assistance, or offer explanations of their modified behavior:

Talking about it helps me emotionally because the more I learn about it—the better I feel about having it.

Also (to) explain my slowness—lack of energy—missed parties ...”

Disclosure is an obligation

Overwhelmingly, respondents perceived diagnosis disclosure as an ethical obligation. Many participants viewed themselves as being the individual who could “give them [family] a heads up that they may have this disease.”

Moreover, the risk of sudden cardiac arrest, as the first sign of a medical problem, coupled with the genetic nature of the illness increased many participants sense of power over others’ lives. Whether talking about their own disclosure or offering advice, respondents repeatedly and emphatically referred to disclosure as a “responsibility” and the “right thing to do.” They stressed the importance of allowing family members the opportunity to make informed decisions about screening options, lifestyle choices, and reproduction decisions in light of an increased risk of HCM. Withholding disclosure was perceived as leaving family members unacceptably vulnerable to the risk of sudden death and other possible complications from HCM. Although most participants saw disclosure as a benefit for the other, whereby the family member could benefit from awareness and preemptive action, a few participants also saw disclosure from a gain frame for all involved.

Without a doubt share it [HCM diagnosis] could save someone’s life! You love your little daughter or son? Want to get those kisses and share those moments? Then you make sure you tell them. You don’t want to share the moment or remember them as you stand next to their coffin. It’s too late then.

My brother died from the disease and never told us he had it. It was not fair to us, not be informed, since there was a possibility of it being genetic. My DX confirmed it is genetics.

Disclosure imperative for adolescents and young adults

Participants also described disclosure as a responsibility when biologically related children and youth and age

groups commonly considered especially vulnerable for most health issues, are present in the family. Many participants mentioned that they shared their diagnosis to benefit related adolescents and young adults in their lives who were very physically active, and in some cases, participated in sports. As such, participants stressed the need to share the diagnosis of HCM when they had young family members.

You owe it to your children.

Your descendants certainly have the right to know in advance that they may have inherited the gene for HCM so they know how to react in the event of symptoms or an emergency—this is especially important in the case of teenage athletes. I have two grandsons who participate in sports and a granddaughter who is a gymnast. I also have a daughter who suffered a cardiac arrest which was not recognized in time to save her from oxygen deprivation and vision loss. You owe it to your family to safeguard their health.

Disclosure process unique for each proband and family

Several participants indicated that they could not offer advice about the disclosure process without having a comprehensive understanding of their family’s communication dynamics. While the need to disclose seemed to be universally recognized as imperative, several respondents were sensitive to the fact that one’s disclosure of the diagnosis might require a personalized approach. Participants acknowledged that pre-existing family communication patterns might influence the disclosure approach. Notwithstanding openness to many processes of disclosure, the majority described a similar process executed and a similar process as recommendation.

I think this ends up being a highly personal thing. Each person needs to know their family and do what they think is best. In the end, giving people the info and letting them do what they will with it is the best you can do.

Just be honest and prepared for drama and potential misunderstanding.

Use Open and Direct Communication

Participants advocated an upfront, honest, and factual delivery of the diagnosis. Participants recommended sharing

the facts, as represented by credible resources. While the diagnosis of HCM often elicited varied reactions, respondents overwhelmingly recommended sharing facts and not emotion. Sudden cardiac death is a concern with HCM, but participants stressed that this should not be presented to biologically related family members as the only prognosis; instead, family members should be advised of the fact that HCM often affects individuals differently and can allow for an unaffected lifestyle.

I believe that this is important information to be shared in a factual way.

I spoke bluntly—matter of fact and with follow-up documentation.

Refer to external experts and resources

Participants recommended that diagnosed individuals consult HCM resources to become more “educated” before disclosing. Overwhelmingly, they reported the need to be equipped with the information that would assist family members in understanding the need to be screened. Additionally, they also suggested providing family members with resources that they could independently consult in order to acquire additional information, especially resources or physicians that specialize in HCM or at an HCMA Center of Excellence (COE). These recommendations emphasized the resources rather than the discloser as the “expert” for family members when they embarked on independent information gathering and decision-making.

Participants noted that genetic counselors and providers at their respective HCM Centers of Excellence relieved the burden of disclosure by relaying information to biologically related family members with helpful handouts and family letters. Participants also utilized materials provided by the HCMA during the disclosure process.

It helps to have the corny pamphlets with the diagrams to show how it works, symptoms possible to expect.

I met with the genetic counselors at (redacted). They provided me with the fact sheet and materials in order to share information with my family.

Some family members may NOT want to hear anything! I found it best to write a brief summary based on HCMA information. On my second try, I attached a letter from a genetic counselor. This letter was better received!

Disclosure amidst turbulent family communication patterns

While participants stress the importance of disclosing the diagnosis to family members, they also acknowledge the likelihood of facing negative reactions. Participants described responses from family members that ranged from blaming and accusation to denial.

Despite these possible responses, participants emphasized the importance of giving family members an *opportunity* to pursue screening that could potentially save their lives. This responsibility they noted, is satisfied after the act of disclosure.

Don’t stress too much if your family won’t get tested and refuses to grasp the serious nature of the disease. You’ve given them the information and that’s all you can do. All but one of my family members is in denial and won’t even let me discuss (remains silent or changes the subject) my having HCM never mind the fact that they could have it too (therefore they won’t get tested). Yes—I recommend that people tell their family in case they want to get tested but don’t rely on them for support because if they’re like my family ... if you don’t talk about it, you don’t have it.

Be prepared to receive a cold reception regarding your diagnosis. You still need to provide as much information as you can to people. What they do with it is their business.

Don’t expect a high rate of return from your effort.

Discussion

Overwhelmingly, participants indicated that disclosure was imperative and shared their diagnosis with family members accordingly. Disclosures most often occurred with first-degree relatives, often less than a year after diagnosis. Participants identified the importance of informing family members about HCM and its hereditary nature; the benefits of conducting screening, genetic testing, and the benefits of acting immediately to avoid the worst outcomes. They also expressed a self-imposed and deeply felt sense of obligation and responsibility as the primary motive for disclosure. Finally, they acknowledged the communication process as unique for every individual and family, yet consistently urged direct and factual delivery of the information, referral to experts and resources other than the discloser, and readiness for adverse reactions to the discloser.

Participants’ recommendations for screening of biologically related family members varied, as some recommended

clinical cardiac screening alone, others recommended screening in conjunction with genetic testing, or mentioned genetic testing only. Participants who described positive experiences with genetic testing often simultaneously recommended the same for their family members, while others expressed doubt regarding usefulness and accessibility. Some participants also expressed concern over being discriminated against (i.e., identifying a pre-existing condition) as a result of the test results, demonstrating the need for greater patient education about anti-discrimination legislation, such as the Genetic Information Nondiscrimination Act of 2008 (GINA), the Genetic Non-Discrimination Act of Canada, the UK's Concordat and Moratorium on Genetics and Insurance, and the European Union's treaty concerning Human Rights and Biomedicine concerning Genetic Testing for Health Purposes. Over half of the participants did not receive genetic counseling and it is unclear as to whether or not these patients were referred by their cardiologists. The absence of genetic counseling coupled with the lack of knowledge or unwillingness that participants described concerning genetic testing may represent significant barriers and lack of accessibility to genetic testing. Probands' lack of consultation with a genetic counselor reduces the availability of informative genetic testing for biologically related family members who could benefit from cascade genetic testing. Given the risk of sudden cardiac death for high-risk individuals and the availability of effective treatment modalities (such as an ICD), uptake of genetic counseling and testing in HCM patients should be as high as possible (Charron et al. 2010). Yet, participants described the same ambivalence as found elsewhere among HCM patients (Smart 2010), including uncertainty regarding accessibility of genetic testing, concerns about accuracy of genetic testing results, and anticipated family communication challenges. Our findings underscore the importance of enlisting support from a genetic counselor during the genetic risk assessment, genetic testing, as well as the genetic testing results disclosure process. Once informed, many relatives of HCM index cases wish to know their mutation status and generally regard genetic counseling positively (Christiaans et al. 2009). Interventions during the disclosure and family communication process, such as a letter from the genetic counselor, have been associated with increased uptake of genetic testing among relatives (Suthers et al. 2006) and can help probands communicate risk information to at-risk relatives.

As in other disease contexts (Bradbury et al. 2007; Duncan et al. 2008), participants perceived disclosure as particularly vital for biological children and youth. Participants unanimously agreed that biological children should immediately be screened, given the risk of sudden death and potential severe manifestation of HCM among adolescents and young adults. Consistent with the oncology literature on inherited cancer risk (Patenaude 1996), several participants considered genetic testing to be problematic for adolescents and young

adults and voiced concerns over larger ethical and developmental implications, including introducing possible anxiety and adversely influencing their outlook on the future. Previous research has explored using varying degrees of openness when communicating about potential genetic risk with children, while seeking balance between children's right for information and parents' desire to protect their children from potential anxiety (Hallowell et al. 2005). These concerns are perhaps heightened in the context of a disease characterized by sudden death that can occur during youth. Despite potential harms, youth with earlier onset genetic conditions such as familial adenomatous polyposis (FAP) (Duncan et al. 2008) and those with genetic conditions not actionable in childhood (Hufnagel et al. 2016) have identified clear benefits associated with receipt of gene-positive test results. After diagnosis and identification of the appropriate treatment plan, disclosure of HCM risk to children may best be conceptualized as put forth by Geelen et al. (2011b), a long-term learning process in which parents and providers collaboratively construct a plan of action that is in the best interest of the child.

The hereditary nature and potential risk for sudden death distinguish HCM from previously studied conditions. Unlike other disease contexts characterized by a potentially delayed onset, probands perceived disclosure as imperative shortly after diagnosis. While participants adamantly support the need for disclosure of their HCM diagnosis, they report many of the challenges found elsewhere in the literature concerning adverse reactions from family members, difficulty communicating with family members and obtaining the resources needed to facilitate disclosure. Over half of the participants in our study disclosed less than a year after diagnosis, but further inquiry is needed to explore the experiences and motivations of probands who delayed disclosure several years later.

Our findings demonstrate that the process of disclosing the diagnosis of HCM to at-risk relatives, while perceived by many as a responsibility, can be a complex and potentially turbulent experience. A genetic counselor can supply helpful tools such as informative materials and customized family letters tailored to meet the needs of the proband. As our participants related, these resources are especially beneficial for probands attempting to fulfill their responsibility to disclose in the midst of turbulent family communication dynamics. Further, probands should consult a genetic counselor to address concerns or questions regarding genetic testing in order to optimize informative mutation carrier status information available for at-risk relatives. While probands almost unanimously agreed that disclosure of the diagnosis of HCM is a responsibility and ethical imperative, greater education is needed to demonstrate the availability and importance of genetic testing in order to facilitate cascade testing for at-risk relatives.

Our findings are notable and expand the literature concerning family communication of genetic risk in the

context of cardiovascular disease. However, it should be noted that our sample consisted of a largely homogenous group of well-educated, high income, White participants. Participants' recruitment from an online advocacy group may have contributed to an overrepresentation of individuals who actively support risk dissemination. This bias and the lack of diversity within this sample may limit the generalizability of results to the larger population of individuals diagnosed with HCM. Future research should actively elicit feedback from individuals from diverse racial/ethnic and socioeconomic standing, especially in light of research showing a significant disparity in preemptive detection and treatment of HCM (Maron et al. 2003). It should also be noted that a selection bias may be present in which individuals who strongly endorsed disclosure may have been more likely to opt-in for participation. The HCMA reports that only 14% of its membership was diagnosed with HCM as the result of family screening. It is likely that a significant proportion of invited members who did not participate in this study would share more nuanced histories of disclosure. It should also be noted that many participants described strained communication following the initial disclosure of diagnosis, suggesting that disclosure is an ongoing process that may require facilitation from a genetic counselor. Additional research is needed to determine the degree to which individuals with HCM may choose not to disclose their diagnosis, and the factors that may influence that decision.

Conclusion

This study represents an important step towards understanding the disclosure dynamics for individuals with HCM. Future research with a more diverse sample is needed to examine a more nuanced understanding of individuals' motivations and methods for disclosing their diagnosis, whether disclosure consists of a single event or an evolving process, and how probands manage their own emotional needs while supporting those of biologically related at-risk family members.

Compliance with ethical standards

Conflict of Interests The authors declare that they have no competing interests.

Use of Animals Not applicable.

Human Involvement in Research All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed consent Informed consent was obtained from all individual participants included in the study.

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