I, Rebecca Bettin, hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

It is entitled:
Motivations for sharing of genetic testing results and cardiac screening recommendations among a pediatric cardiomyopathy population

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Motivations for Sharing of Genetic Testing Results and Cardiac Screening Recommendations among a Pediatric Cardiomyopathy Population

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Abstract

Cardiomyopathy is a chronic and often progressive disease of the myocardium that is typically inherited. Clinical genetic testing is available for hypertrophic (HCM) and dilated cardiomyopathy (DCM). Identification of the genetic etiology in the proband allows for risk-stratification of relatives. It is recommended that individuals at-risk for HCM or DCM undergo cardiac screening. A written survey was sent to parents of patients evaluated in the cardiomyopathy clinic at Cincinnati Children's Hospital Medical Center to characterize the motivations for genetic testing in their child and mode of information sharing among families with cardiomyopathy. Eighty-eight percent of parents identified two primary motivating factors in the decision to proceed with genetic testing; 1. help to prepare for the future and 2. avoid the worry of uncertainty. The number of affected family members and the presence of sudden cardiac death (SCD) in the family significantly impacted the parent-identified motivators. Families with two or more affected individuals identified two different motivators for genetic testing; 1. confirm the diagnosis in the affected person (92%) and 2. avoid unnecessary screening of unaffected individuals (83%). One hundred percent of families with SCD felt testing to confirm a diagnosis was their primary motivator. Families who had genetic testing were more likely to share results with family members by phone or with a counseling letter while those who did not have genetic testing tended to share results in person. Knowledge regarding motivations for genetic testing and mode of sharing of health information in the context of pediatric cardiomyopathy is important for improving communication between provider and the family.

Keywords: genetic counseling, family sharing, communication, cardiomyopathy, motivations, barriers, modes of sharing
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Introduction

Cardiomyopathy

Cardiomyopathy is a disease of the myocardium, characterized by specific changes in the structure and function of the heart affecting its contractility. There are various types of cardiomyopathy. The two most common being hypertrophic cardiomyopathy (HCM) and dilated cardiomyopathy (DCM), (Schwartz, 1996). HCM is the most prevalent form in the adult general population, affecting 1 in 500 people. While HCM is rare in the pediatric population with a prevalence of 1 in 100,000, it has severe consequences with up to 40% of affected patients progressing to death or transplant within five years of diagnosis (Lipshultz, 2003). This subtype is distinguishable from the other forms of cardiomyopathy by asymmetrical left ventricular hypertrophy. Symptoms of HCM can present at any age with reported onset ranging from prenatal into the late 80s. However, most patients with HCM show no early symptoms (Tome Esteban, 2007 and Maron, 2002). Sudden death may be the initial presenting symptom highlighting a need to identify at risk individuals. DCM is defined as having a dilated left ventricle and systolic dysfunction. It is the “most common form of heart muscle disease today [including adult and pediatric cases], comprising 60% of identified cardiomyopathies” (Fowler, 2009).

Both HCM and DCM have a strong genetic component with anywhere from 20-70% having an identifiable gene mutation and many having a positive family history of disease. Familial HCM and DCM are typically inherited in an autosomal dominant manner (Hershberger, 2009). Incomplete penetrance and variable expressivity are
commonly observed in families (Morales, 2008). This clinical variability ranges from lifelong absence of symptoms to sudden cardiac death (SCD) (Hudecova, 2009; Maron, 2003). “Cardiomyopathy is one of the leading causes of death in children” (Schwartz, 1996), and in adults under the age of 30 (Hudecova, 2009).

**Genetic Testing and Sharing of Cardiac Health Information**

“Genetic testing aims to identify inherited variations in genetic information that may confirm, or exclude a diagnosis or predict future disease in an asymptomatic patient” (Hershberger, 2008). Genetic testing for genes responsible for the familial forms of cardiomyopathy is clinically available (Wheeler, 2009). Identification of a gene mutation in an affected individual allows at-risk family members to be tested for the familial mutation. Individuals found to be mutation positive have the opportunity to undergo early and routine follow up prior to the onset of overt symptoms allowing for the earliest identification of disease and possible prevention of sudden cardiac death. The Heart Failure Society of America published screening guidelines in 2009 recommending routine cardiac evaluation for unaffected individuals with a family history of cardiomyopathy and/or known gene mutation carriers (Hershberger, 2009). An additional benefit to known mutation testing in at-risk family members is the potential to discontinue screening in those found not to carry the familial mutation (Christianns, 2009).

Genotype-phenotype correlations have been reported however yet to impact individual management in the vast majority of cases. Current clinical management continues to be based primarily on the individual’s medical and family history (Maron, 2002). It is recommended that first-degree relatives of an affected person
be informed of the genetic nature of the disease, its treatment, complications, and prognosis (Maron, 2003; van der Roest, 2008). Having a family history of cardiomyopathy, heart failure, or SCD increases a families’ risk for all of these events (Hershberger, 2008). This increased risk is the most important reason families should share their cardiac health information. Previous studies have reported information sharing among families, revealing that family letters are an effective way to inform relatives about inherited cardiac disease (van der Roest, 2008).

**Study Purpose**

Despite the availability of genetic testing and numerous publications recommending cardiac screening for individuals at-risk for cardiomyopathy, very little data exists on parents’ decisions regarding genetic testing for cardiomyopathy and the mode of sharing test results and screening recommendations with other family members. While previous studies have addressed predictive genetic testing, they have primarily reported on cancer predisposition. It is unclear whether these results are generalizable to the pediatric cardiomyopathy population. One objective of this study was to look at factors influencing parents’ decision to proceed with genetic testing for their child among a pediatric cardiomyopathy population. Another objective was to explore the factors that influence information sharing, including influential factors and preferred mode of communication. The final objective was to determine if the number of family members affected or the experience of sudden cardiac death in the family influenced motivations or mode of sharing genetic health information. Results from this study will help guide practitioners of various
disciplines in the pediatric cardiomyopathy field to better understand and care for their patients by providing guidance through the decision making process.
Methods

Participants

The study participants were parents or guardians of pediatric cardiomyopathy patients evaluated in the Cardiomyopathy/Heart Failure Clinic (C/HFC) at CCHMC between October 1, 2006 to December 31, 2010. The parents or guardians of patients who met the following criteria were eligible for the study: 1) A clinical diagnosis of idiopathic or familial dilated cardiomyopathy (DCM) or hypertrophic cardiomyopathy (HCM) 2) Unaffected patients known to be at risk for developing DCM or HCM based on the family history or mutation status. For the purposes of this study, familial cardiomyopathy was defined as an affected patient with at least one affected first or second degree relative or cardiomyopathy in a patient with an underlying sarcomeric or cytoskeletal gene mutation. Parents of patients with DCM or HCM caused by another etiology such as syndromic, metabolic, neuromuscular or required causes and parents who were unable to read English were not asked to participate in this study. Participants were not excluded based on where they live, race, educational background, number of children or frequency of visits in the C/HFC. The parents of 88 patients were eligible and received the study questionnaire by mail (Appendix I). The Institutional Review Boards of both Cincinnati Children’s Hospital Medical Center and the University of Cincinnati approved this study.

Informed Consent

Informed consent was incorporated into the cover page of the questionnaire and was obtained from the pediatric patient’s parent or guardian. All participants were
parents or guardians of pediatric patients under the age of 18. The document explained that participation was voluntary, would in no way impact clinical care and that healthcare providers would not have access to individual responses. The questionnaires were sent by mail. Completing and returning the questionnaire served as consent to participate in the study. If the parent or guardian did not want to participate and preferred no further contact regarding the study, they could return the blank questionnaire in the prepaid envelope.

*Questionnaires*

The data was collected using a written questionnaire (Appendix I) designed by the study team. Previously reported barriers and motivators for genetic testing were incorporated into the questionnaire (Levine, 2010; McLaren, 2003; Michie, 1996; van der Roest, 2008). Four motivating factors were borrowed from Levine and others entitled, *Parental attitudes, beliefs and perceptions about genetic testing for FAP and colorectal cancer surveillance in minors*. Three reasons for genetic testing were adapted from interview questions and responses reported by Michie and others, *Parents’ responses to predictive genetic testing in their children*. Two demographic questions, one-barrier to testing and two motivations for testing questions were included from the HEIRS (Hemochromatosis and Iron Overload Screening) Study by McLaren et al, 2003. The questionnaire designed for this study consisted mostly of closed-ended questions aimed at characterizing factors that influenced parent’s or guardian’s decisions about genetic testing and information sharing with other family members in regard to test results and recommended cardiac screening practices. There were two versions of the questionnaire; one
version did not include specific questions regarding sharing of genetic testing results with family members. This modified version of the questionnaire was used for the participants who had not undergone genetic testing for cardiomyopathy at the time of the study. The other version of the questionnaire went to the parents or guardians of patients that were known to have had genetic testing. The two versions of the questionnaires were printed on three different colors of paper representing positive genetic testing results (green), negative genetic testing results (yellow) and no previous genetic testing for cardiomyopathy (red).

Each participant was assigned a study identification code which was linked to their child's genetic testing results, medical record number (MRN), date of birth, family history, echocardiogram and ECG in addition to other information contained in their medical record. Only study staff not involved in clinical care, had access to the individual responses. For the protection of the participants' confidentiality, the study key was destroyed upon completion of the study.

Data Storage

The responses from the questionnaires were entered by hand and managed using Research Electronic Data Capture (REDCap), an electronic data capture tool hosted at the Center for Clinical and Translational Science and Training provided to researchers affiliated with the University of Cincinnati Academic Health Center (Harris, 2009). REDCap is a secure, web-based application designed to support data capture for research studies.
Data Analysis

Basic descriptive statistics were used for this study, calculating frequencies and means as appropriate. Likert scales were used to address motivations for genetic testing and sharing of screening recommendations. A five-point scale was used where 1 was very important and 5 was least important. Values for 1 and 2 were combined to form the important category and 3, 4, and 5 were combined to form the non-important category. In addition, severity of disease was also assessed using a Likert scale in which 1 meant least severe and 5 being most severe. Those who responded with an answer of 1 or 2 were considered to view the disease as least severe, 3 was moderately severe, and a response of 4 or 5 was most severe. Reporting results as dichotomous variables was done for the ease of interpretation. For the secondary analysis, we used chi-square tests to compare the proportions between two groups. All analyses were performed in JMP Genomics 4.
Results

Demographic Characteristics

Eighty-eight parents or guardians of patients seen in the C/HFC within CCHMC were invited to participate in this study. Fifty-five (63%) participants were sent the questionnaire designed for those who’s child previously had genetic testing and thirty-three (37%) were sent the questionnaire designed for those who’s child had not previously had genetic testing.

The overall response rate was 28%. Seventeen (31%) were from the tested group in which 15 (88%) tested positive and two (12%) tested negative and 8 (25%) were from the non-tested group. One questionnaire was returned blank. Table 1 summarizes the demographic variables for the twenty-five parent/guardian participants. The average age of the participant was forty-one years old. All the participants reported being Caucasian. The majority were female, had a two year college degree or higher, and had one child diagnosed with cardiomyopathy. Forty percent of the parent respondents reported having a personal diagnosis of cardiomyopathy. One participant reported having both a diagnosis of cardiomyopathy and arrhythmia. Of the non-participants, eighty-two percent were Caucasian, fifteen percent were African American/Black, and three percent were Hispanic or Puerto Rican. Marital status and education was not available in the patient’s medical record for comparison to the study population. The majority of the children whose parents did not respond had either a personal or family history of HCM, which correlates with our study population. However, the number of non-
respondents with a family or personal history of DCM was greater compared to the number of respondents with a family or personal history of DCM.

**Participants who Previously Completed Genetic Testing (Testers)**

Of the participants who responded to the questionnaire designed for those who had genetic testing, 13 (76%) reported their affected child(ren) as having a diagnosis of HCM, while only 2 (12%) reported a diagnosis of DCM in their affected child(ren); 2 (12%) participants did not respond to the question. When these same participants were asked about the diagnosis of cardiomyopathy in the family, 12 (71%) reported a history of HCM, 2 (12%) reported a history of DCM, and 2 (12%) stated that there was no diagnosis of cardiomyopathy made within their family. The majority (75%) reported that their affected child did not experience symptoms related to cardiomyopathy. Eighty-two percent of affected children had not been admitted to the hospital overnight because of their heart disease. When asked to rank how severe they felt their child’s heart condition was, forty percent said low severity, twenty-seven percent said moderate severity and thirty-three percent said high severity.

**Participants who had Previously NOT Completed Genetic Testing (Non-testers)**

Of the participants who were sent the questionnaire designed for those who had not completed genetic testing, only one (13%) was offered genetic testing for their child and declined. Seven (87%) were not offered genetic testing because there was not an appropriate person in the family available for testing.
Genetic Testing

Fifteen of the twenty-five participants (60%) reported that they had heard of genetic testing for cardiomyopathy prior to their evaluation in the CHFC. Fifty-three percent of the testers reported first learning about genetic testing for cardiomyopathy from their cardiologist and 35% first learned about genetic testing from a genetic specialist (genetic counselor or geneticist) and 12% first learned about genetic testing from a support group, primary care physician or family member. Thirteen percent of the non-testers reported first learning about genetic testing for cardiomyopathy from a cardiologist, 63% first learned about genetic testing from a genetics specialist and 23% first learned about genetic testing from a support group, primary care physician or family member. Ten (40%) of the parent respondents personally had genetic testing for cardiomyopathy, seventeen (68%) reported their child had had genetic testing, nineteen (76%) reported that multiple family members (first and second degree relatives) had had genetic testing. Out of the twenty-five participants, thirteen (52%) reported that their child was the first person in the family to have genetic testing, three (12%) reported that they were the first person in the family to have testing, four (16%) said it was the child’s other biological parent, one (4%) stated it was another family member related to the child who had genetic testing first, one (4%) reported no one had been tested yet and four (16%) did not answer the question.

Motivators for Genetic Testing

Parent participants who were sent the genetic testing questionnaire were asked to consider several factors that may have influenced their decision to proceed with
genetic testing for their child. These factors were ranked according to importance and the results are summarized in Figure 1.

The responses were evaluated in three groupings; important, moderately important and minimally important. A factor was considered important if 80% of respondents ranked it on the Likert scale as either a 1 or 2, moderately important if 60-80% ranked the factor a 1 or 2 and minimally important if 60% ranked the factor as a 1 or 2. The two most important motivations for genetic testing identified included *help prepare for the future* and *avoid the worry of uncertainty*. The moderately important factors were *avoidance of unnecessary screening of unaffected individuals*, *confirm a diagnosis in an affect individual*, *the ability to share the information with extended family members* and *parents have the right to decide their child’s health care practices*. The factors identified as minimally important included *cost of genetic testing*, *fear of discrimination*, *fear of psychological reaction to the results* and *child has the right to decide if they want to be tested*.

The motivations for testing differed on how many people in the family had a diagnosis of cardiomyopathy and are summarized in Figure 2. All of the families with one or less affected individuals designated *preparing for the future* as an important factor. More (88%) of these participants also reported *ability to share the results with extended family* as an important factor when compared to families with 2 or more affected individuals (67%). More families with one or less affected family member also identified *fear of discrimination* (insurance, employment, society, etc) (50%) as an influential factor compared to those with two or more affected (25%). Families with one or less member affected felt that it was important to let the child
have a right to decide if they want to be tested (43%). Whereas one hundred percent of the families with two or more members affected felt *letting the child have a right to decide if they wanted to be tested* was not important. Families with two or more affected individuals felt strongly about testing to *confirm the diagnosis in an affected person* (92%), to *avoid unnecessary screening of unaffected individuals* (83%). Both groups were in agreement on the importance of *parents have the right to decide their children’s health care practices* (1 or less 63%; 2+ 66%) and *avoid the worry of uncertainty* (1 or less 88%, 2+ 92%).

In addition, there were notable differences in motivations for testing between those who reported sudden cardiac death (SCD) in their family and those who did not. One hundred percent of families with SCD in their family felt it was important to have testing to confirm a diagnosis. Yet they did not feel as strongly about testing to prepare for the future (60%) as those families without SCD in the family (100%). Conversely, those families without SCD considered *fear of discrimination* and *child’s right to decide about testing* as important factors (44%, 20%), whereas one hundred percent of the families with SCD in family considered those two factors not important in their decision about genetic testing for their child.

*Motivators for Sharing Cardiac Screening Recommendations*

The parent participants were also asked to rank their motivations for sharing screening recommendations as important or not important. We compared the answers provided by the tested and non-tested groups. These results are summarized in Figure 3. Both the testers and the non-testers identified, *to encourage family members to begin screening practices* as the most important factor.
influencing sharing of cardiac screening recommendations. All respondents in the non-tested group identified the following factors as important: to educate the family about cardiomyopathy, to make the family aware of other risks associated with cardiomyopathy, and to make family aware of sudden cardiac death. The tested group also identified the above factors as important. Eighty-seven percent of the tested group felt that educating their family about cardiomyopathy and making their family aware of other risks associated with cardiomyopathy were important factors when considering sharing of cardiac screening recommendations. Only 73% of the tested group felt that making their family aware of the risk of SCD was an important motivator for sharing. Eighty-six percent of non-testers felt that having a genetic counselor or geneticist say that the sharing of screening recommendations was important was a significant motivator for them to share; while only 60% of the tested group felt that it was an important motivator.

Eighty-six percent of the non-tested group also felt that they were motivated to share by hoping that their family would understand and be accommodating of the cardiac diagnosis. However this seemed to be a lower motivator for the testing group since only 53% identified it as important. The testing group did feel stronger about sharing the screening recommendations if they knew family members were interested in having genetic testing (33%) than the non-tested group (17%). Both groups felt that just having cardiac screening recommendations come up in a conversation was not a motivator to share information about screening.
Family Members with whom Screening Recommendations were Shared

There was no significant difference between the testing and the non-testing groups regarding with whom information was shared. At least half of each group (50% non-tested, 59% tested) shared screening recommendations with their siblings or the siblings of their child’s other biological parent. Twenty-five percent more participants in the testing group shared the recommendations with their child’s grandparents. Few shared information with their child’s great grandparents or second cousins. In contrast, the methods of sharing screening recommendations differed greatly between the two groups. No one from the non-tested group shared the information over the phone, whereas half (53%) of the tested group did. In addition, only participants in the tested group (50%) used a letter provided by a genetic counselor to share the information. The majority of sharing for both groups occurred in person (non-tested 38%, tested 59%). E-mail was used rarely among both groups (non-tested 13%, tested 6%). Sharing through word of mouth, by telling one family member and asking them to share the information with other family members, was used slightly more often than email by both groups (non-tested 25%, tested 12%).

Motivators for Sharing Genetic Test Results versus Screening Recommendations

The motivations for sharing genetic testing results and screening recommendations were compared within the testing group. The results are summarized in Figure 4. Overall, the participants identified similar motivations for sharing genetic testing results and cardiac screening recommendations.
The greatest motivating factor in either case was a desire to educate their family about cardiomyopathy (screening 78%, testing 75%). This was followed closely by making the family aware of the risk of sudden cardiac death (screening 73%, testing 75%). Participants felt it was slightly more motivating to have a genetic counselor or geneticist state the importance of sharing screening recommendations compared to sharing genetic testing results (screening 60%, testing 50%).
Discussion

Genetic testing for cardiomyopathy and consideration of genetic testing and cardiac screening for potentially at-risk family members are part of routine clinical care in the C/HFC at CCHMC. All the patients in this study were provided genetic counseling by either a genetic counselor or a medical geneticist who, in conjunction with other members of the multidisciplinary team, addressed genetic testing and cardiac screening with patients and families. The current study characterizes factors that may have influenced parents’ decisions to proceed with genetic testing for cardiomyopathy in their child. In addition, factors that influenced the decision to share and the mode of communicating genetic testing and cardiac screening recommendations were also explored.

Overall, there was a high uptake of genetic testing for cardiomyopathy among our cohort. This is not surprising given the previous report by Fitzgerald-Butt et al, 2009. Parents in this study expressed an interest in genetic testing and counseling, however had not been offered these services in a clinical setting. The uptake of testing in our population suggests what when offered testing and counseling in a clinical setting, parents are interested and proceed with testing. Of the twenty-five completed questionnaires, only one family chose not to proceed with genetic testing when offered the option. These results suggest that parents of children with possible familial cardiomyopathy are interested in the information genetic testing may provide and are comfortable with their child being tested. Within the non-tested group, seven of the eight participant families were not offered genetic testing because there was not a suitable candidate for testing within the family. In general,
the most informative person to test is the most severely affected individual. For the families who were not offered genetic testing, the only affected individual had passed away, lived far away or was not interested in genetic testing.

**Motivations and Factors Influencing Uptake of Genetic Testing**

In order to better understand the high uptake of genetic testing, parent participants were asked to rank motivations and factors influencing the decision to be tested. We speculate that the high uptake of genetic testing among pediatric cardiomyopathy patients may be related to their parents' motivations for testing. Several studies have previously reported cost of testing and fear of discrimination as barriers to genetic testing for various genetic conditions. (Dugan, 2003; Falcone, 2011; Fitzgerald-Butt, 2009; Levine, 2010; Michie, 1996; van der Roest, 2008). Our population did not identify these previously reported barriers as important when deciding about genetic testing for their child. Levine et al in 2010 previously reported parental concern regarding adverse psychological reactions as a barrier to genetic testing. In contrast, parents in this study identified this factor as minimally important. As discussed above, these three factors have been identified as impacting the decision to have or not have genetic testing in numerous studies regardless of the condition or age of the patient. It is not clear how our population and the identified motivators differ. One possibility is the risk for SCD, which is unique to cardiomyopathy. It is possible that fewer parents’ identified cost as a motivating factor due to new laboratory techniques that have lowered the cost of testing. In addition, many of the laboratories offering clinical genetic testing for cardiomyopathy have instituted patient-friendly billing policies limiting the
potential out-of-pocket cost. It is also possible that patients’ and families’ view of
genetic testing and risk of genetic discrimination has changed over time and may be
impacted by the passing of the Genetic Information Nondiscrimination Act (GINA)
and a better understanding of previously existing federal laws such as the Health
Insurance Privacy and Accountability Act (HIPAA).
The motivators that greatly contributed to the parents’ decision to have genetic
testing for their child in our study differs from those previously reported by adult
patients for late onset conditions or conditions with less intra-familial clinical
variability. As a whole, parents identified wanting to gain information to help
prepare for the future and avoid the worry of uncertainty as the top two motivators
for genetic testing. These two motivators were also reported in a case study by
Michie et al 1996, by parents who wanted their children tested for familial
adenomatous polyposis (FAP) at ages 2 and 4 years old. These parents reported
seven reasons for wanting genetic testing, two avoiding the worry of uncertainty and
helping prepare for the future matched our top motivators. In another study,
Parental attitudes, beliefs and perceptions about genetic testing for FAP and colorectal
cancer surveillance in minors, Levine et al reports that knowledge, and reduction of
anxiety and uncertainty were among the reasons parents gave for having their
child(ren) genetically tested for mutations in the APC gene. This may imply that
parents whose child could possibly be affected by a genetic condition with variable
expressivity, incomplete penetrance or later onset of symptoms desire this
information to help adjust the upbringing of their child(ren). The worry and guilt,
which may be associated with the possibility of passing on a genetic condition to a
child could possibly be relieved if genetic testing reveals the child did not inherit the mutation. Knowing that their child does not have the genetic mutation in question may allow the parents to stop worrying about the uncertainty. These data add to the existing body of literature regarding motivations for pursuing genetic testing in a minor child.

*Presence of Sudden Cardiac Death and the Number of Affected Family Members*

The number of affected family members and the presence of SCD in the family did influence the parent identified motivations for genetic testing. *Preparing for the future* was the most important motivator for the cohort as a whole as well as for families with one or less person affected and/or no history of SCD. However, for families with two or more affected relatives and/or the presence of SCD, the primary motivator was *confirm the diagnosis in the affected individual*. This difference may suggest the loss of a family member due to SCD influences, which motivators impact the decision to be tested. These families may be more focused on the present situation and are more motivated by the future benefit of testing compared to families who have not experienced SCD. Interestingly, *fear of discrimination* was an important factor identified by 44% of families without SCD. This may imply that families who have not encountered the unexpected death of a loved one are more focused on the potential societal implications, such as stigma or discrimination, associated with testing, when compared to families who experienced a SCD. In addition, families with one or less person affected felt it was extremely important to have genetic testing in order to *share results with extended family members* compared to those with 2 or more family members affected. One possible
explanation for this observed difference may be a need for families with multiple affected individuals to focus on those with a current diagnosis of cardiomyopathy as opposed to potentially at-risk relatives. Families with one affected individual may be motivated to share this information in order to prepare relatives for a possible diagnosis as the first diagnosis in the family may come without warning. Families with fewer affected individuals may feel a greater individual responsibility to share information with relatives. Whereas families with multiple affected individuals view the responsibility to inform other family as shared or may believe that at risk relatives are already informed.

Motivations for Sharing

To the best of our knowledge this is the first report regarding motivations for sharing genetic testing results and cardiac screening recommendations for cardiomyopathy. It is recommended that first-degree relatives of an affected person be informed of the genetic nature of the disease, its treatment, complications, and prognosis (Maron, 2003; van der Roest, 2008). Having a family history of cardiomyopathy, heart failure, or SCD increases a families' risk for all of these events (Hershberger, 2008). This increased risk is the most important reason families should share their cardiac health information. It is accepted that not all affected or mutation positive individuals share this information with their family members. Dugan et al in 2003, and McGivern et al in 2004, reported the most commonly cited reasons for refusal to share genetic testing results by patients were estranged family relationships, concern for altering family dynamics and not being close to or in contact with relatives. In addition, Dugan et al reported that 46% of genetic
counselors have had a patient openly refuse to notify their at risk relatives of genetic information. In our study, every participant (except one of the non-testers) reported that they shared either genetic testing results, cardiac screening recommendations or both with their family. This is similar previously published data in a Hereditary Breast and Ovarian Cancer (HBOC) population. McGivern et al, 2004, reported that 97% of a BRCA1/BRCA2 positive patient cohort reported sharing genetic information with at least one at risk relative. The most important reasons for discussing BRCA test results with family members in the HBOC population were to inform them of their risk, and fulfill a duty to inform. Despite a different disease, the motivations to share genetic testing results appear to be similar among our cohort. Top motivators for sharing genetic testing results with family included, educate the family about cardiomyopathy, and make the family aware of the risk of sudden cardiac death. The motivations to share cardiac screening recommendations varied slightly from the motivations to share genetic testing results. The primary reason participants gave for sharing screening recommendations was encourage family members to begin screening practices. While this may seem obvious it is not something that is intuitive for most families and is a focus of the genetic counseling session. All but one of our parent participants reported sharing information with family members. However, the mode of sharing this information and whom it was shared with depended upon whether or not their child had genetic testing. Those parent participants who decided to proceed with genetic testing for their child shared cardiac screening recommendations by phone or with a letter provided by the
genetic counselor. Those parent participants whose child did not have genetic
testing primarily shared screening recommendations in person. Patients in C/HFC
who do not undergo genetic testing are not routinely provided with written cardiac
screening recommendations. Given that a letter was identified as a primary method
of sharing among those families that did receive letters, it may be beneficial to
provide a letter about screening recommendations to all families regardless of
whether or not genetic testing was completed.

All patients evaluated in the C/HFC were seen by a genetics professional. However,
44% reported that they first learned about genetic testing from a genetic counselor
or geneticist, and 40% first learn about genetic testing from a cardiologist. This may
imply that the 44% that learned about genetic testing from the genetic counselor or
geneticist and are not severely affected or being routinely screened because of a
positive family history by a cardiologist may lose out on the opportunity to learn
about the importance of sharing cardiac screening information if it wasn't for the
participation of genetics in a multidisciplinary cardiology clinic. In a
multidisciplinary clinic, genetics professionals are likely to spend more time with
children and families being followed because of increased risk whereas cardiologists
are likely to spend more time with those children and families with a diagnosis. This
may result in a higher proportion of at-risk or mildly affected individuals learning
about genetic testing as opposed to those children and families with an existing
diagnosis. The incorporation of genetic counselors and geneticists into a
multidisciplinary cardiology clinic may lead to improved communication regarding
availability of genetic testing and cardiac screening recommendations.
**Study Limitations**

There are multiple limitations of this study. While the sample size is small, the cohort and response rate are comparable to other studies that have used questionnaires to investigate barriers to testing and communication within a family, (Dugan, 2003; Falcone, 2011; Fitzgerald-Butt, 2009; Levine, 2010). Another limitation was that the questionnaire was never pre-tested or validated. Even though some of the questions were generated from other studies, it was never evaluated for clarity or readability outside of the study group. One more limitation might be participation bias. Individuals that chose to participate might be more willing to share genetic testing results or screening recommendations than those who did not participate in this study.
Conclusion

Our questionnaire-based study explored parental motivations for genetic testing in a child diagnosed with cardiomyopathy. The majority of families who were offered genetic testing elected to proceed and report *help prepare for the future* and *avoid the worry of uncertainty* as the main motivators. The number of affected individuals within a family and the presence of SCD in a family impacted motivations for genetic testing. A significant proportion of families chose to share genetic test results and cardiac screening recommendations. Parents who chose to proceed with genetic testing for their child chose to share genetic test results and cardiac screening recommendations with family members by phone, in person or with a letter written by a genetic counselor. These results support prior recommendations that genetic testing and discussion of cardiac screening recommendations should be incorporated into clinical practice and that involvement of genetics professionals into routine care may optimize implementation of these recommendations. These findings add to current knowledge and understanding about parents’ motivations to pursue genetic testing for their children with cardiomyopathy and motivations and preferred methods of sharing health information with relatives.
## Tables and Figures

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency (n=25)</th>
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<tbody>
<tr>
<td><strong>Gender</strong></td>
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<td>Female</td>
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<td>Male</td>
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<td>2 or 4 year college degree</td>
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<tr>
<td>Widow</td>
<td>4%</td>
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<tr>
<td><strong>Participants Affected</strong></td>
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<tr>
<td>(one person had 2 diagnoses)</td>
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<tr>
<td>CM</td>
<td>40%</td>
</tr>
<tr>
<td>Arrhythmia</td>
<td>4%</td>
</tr>
<tr>
<td>Other</td>
<td>16%</td>
</tr>
<tr>
<td>Don’t know/no diagnosis/no response</td>
<td>44%</td>
</tr>
<tr>
<td><strong>Total number of biological children</strong></td>
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<tr>
<td>At least 1</td>
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</tr>
<tr>
<td>2 or more</td>
<td>76%</td>
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<tr>
<td><strong>Number children undergoing cardiac screening at CHFC</strong></td>
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<tr>
<td>At least 1</td>
<td>72%</td>
</tr>
<tr>
<td>2 or more</td>
<td>28%</td>
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<tr>
<td><strong>Number of children diagnosed with cardiomyopathy</strong></td>
<td></td>
</tr>
<tr>
<td>Zero</td>
<td>33%</td>
</tr>
<tr>
<td>One</td>
<td>63%</td>
</tr>
<tr>
<td>2 or more</td>
<td>4%</td>
</tr>
</tbody>
</table>
Figure 1  Patient Motivations for Genetic Testing

- Help to prepare for the future: 88%
- Avoid the worry of uncertainty: 88%
- Avoidance of unnecessary screening of unaffected individuals: 77%
- To confirm diagnosis in an affected individual: 76%
- Ability to share this information with extended family members: 76%
- Parents have the right to decide their child’s health care practices: 63%
- Cost of genetic testing: 47%
- Fear of discrimination: 41%
- Fear of psychological reaction to the results: 29%
- Child has the right to decide if they want to be tested: 19%
Figure 2  Comparison of Motivators by Affected Family Members

- Help to prepare for the future: 83% (100%), 17% (2+)
- Avoid the worry of uncertainty: 88% (100%), 92% (2+)
- Avoidance of unnecessary screening of unaffected individuals: 75% (83%), 83% (2+)
- To confirm diagnosis in an affected individual: 92% (100%), 63% (2+)
- Ability to share this information with extended family members: 88% (100%), 67% (2+)
- Parents have the right to decide their child’s health care practices: 66% (2+), 63% (2+)
- Cost of genetic testing: 50% (100%), 34% (2+)
- Fear of discrimination: 50% (100%), 25% (2+)
- Fear of psychological reaction to the results: 33% (100%), 25% (2+)
- Child has the right to decide if they want to be tested: 43% (100%), 1% (2+)
It just came up in conversation

Family members are interested in having genetic testing

Figure 3

Important Motivators for Sharing Screening Recommendations

- Encourage family members to begin screening practices
- To educate family about cardiomyopathy
- Make family aware of other risks associated with cardiomyopathy
- Make family aware of the risk of sudden cardiac death
- Genetic counselor or geneticist said it was important
- In hopes that they would be understanding and accommodating
- Family members are interested in having genetic testing
- It just came up in conversation
Figure 4
Factors that Influence Sharing: Genetic Testing vs Screening Recommendations

- To educate family about cardiomyopathy: 75% Genetic Testing (N=16) vs 87% Screening Recommendation (N=15)
- Make family aware of the risk of sudden cardiac death: 75% vs 73%
- Genetic counselor or geneticist said it was important: 50% vs 60%
- Family members are interested in having genetic testing: 31% vs 33%
- It just came up in conversation: 25% vs 7%

Genetic Test Results
Screening Recommendations

Percentage of Respondents
References


Appendix I - Questionnaires

Tested

Section 1: Information about the individual completing the survey.

1. What is your gender?
   [ ] Male
   [ ] Female

2. What is your age?
   _______ Years

3. Which of the broad categories best describes your family’s race?
   [ ] American Indian/Alaskan Native
   [ ] Asian
   [ ] Black or African American
   [ ] Native Hawaiian or other Pacific Islander
   [ ] White or Caucasian

4. How far did you get in school?
   [ ] Less than High School
   [ ] High School diploma or equivalent
   [ ] Some College
   [ ] College Graduate (Associate's Degree, Bachelor's Degree)
   [ ] Master's Degree
   [ ] PhD, MD or the like

5. What is your marital status?
   [ ] Single
   [ ] Married
   [ ] Separated/Divorced
   [ ] Widowed

6. Have you been diagnosed with one of the following?
   [ ] Heart muscle disease (cardiomyopathy)
   [ ] Heart rhythm problem (cardiac arrhythmia)
   [ ] Other heart problem _________________
   [ ] I don’t know

7. Has your child’s other biological parent been diagnosed with one of the following?
   [ ] Heart muscle disease (cardiomyopathy)
   [ ] Heart rhythm problem (cardiac arrhythmia)
   [ ] Other heart problem _________________
   [ ] I don’t know
8. How many biological children do you have?
   [ ] None      [ ] Three
   [ ] One       [ ] Four or more
   [ ] Two

9. How many of your biological children are followed by a cardiologist in the Children’s Hospital Heart Institute Cardiomyopathy Clinic?
   [ ] None
   [ ] One
   [ ] Two
   [ ] Three
   [ ] Four or more

10. How many of your biological children have a diagnosis of: (Please circle)
    Heart muscle disease (cardiomyopathy)  0  1  2  3  4+
    Heart rhythm problem (cardiac arrhythmia)  0  1  2  3  4+

Section 2: Information about your first child to be evaluated by Cardiology at Cincinnati Children’s Hospital. (Not necessarily your first-born child)

1. Does your child have a diagnosis of cardiomyopathy? (If No skip to Section 3)
   [ ] Yes
   [ ] No

2. At what age did your child first experience symptoms?
   ________________ months old
   ________________ years old
   [ ] Never had symptoms

3. Please choose the symptoms experienced by your child:
   [ ] Shortness of breath
   [ ] Chest pain
   [ ] Abnormal heart rate
   [ ] Difficulty exercising
   [ ] Excessive sweating
   [ ] Fainting
   [ ] Heart Failure
   [ ] Other______________________________________________
   [ ] No symptoms

4. Has your child ever been admitted to the hospital to stay overnight because of their heart disease?
   [ ] Yes
      If yes, about how long was the stay_________________________
   [ ] No
5. On a scale of 1 to 5 how severe do you feel your child's heart disease is; 1 being the least severe and 5 being the most severe?

1 2 3 4 5

6. At what age was your child diagnosed with cardiomyopathy?

___________ prenatally (before birth)
___________ months old
___________ years old

7. What type of cardiomyopathy has been diagnosed in your child?

[ ] Hypertrophic Cardiomyopathy (HCM; thick left heart muscle)
[ ] Dilated Cardiomyopathy (DCM; large left pumping chamber)
[ ] Other _______________
[ ] I don’t know
[ ] None

Section 3: Information about immediate and extended biological family members of the first child evaluated at Cincinnati Children’s.

1. How many people in the family, other than your child, have cardiomyopathy diagnosed by a doctor?

[ ] One
[ ] Two
[ ] Three
[ ] Four
[ ] Five or more
[ ] No one

2. What type of cardiomyopathy has been diagnosed in the family?

[ ] Hypertrophic Cardiomyopathy (HCM; thick left heart muscle)
[ ] Dilated Cardiomyopathy (DCM; large left pumping chamber)
[ ] Other _______________
[ ] I don’t know
[ ] None

3. Which of the listed factors led to the initial diagnosis of cardiomyopathy in the first person in the family?

[ ] Heart murmur detected
[ ] Electrocardiogram (EKG)
[ ] Echocardiogram (Echo)
[ ] Symptoms (fainting, difficulty exercising, chest pain, excessive sweating, etc)
[ ] Sudden (cardiac) death of a family member
[ ] Other ___________________________
4. How old was the **youngest person** in the family at the time of diagnosis?

- _____________ months old
- _____________ years old

Section 4: Information about **genetic testing** for cardiomyopathy.

1. Had you heard about genetic testing for cardiomyopathy before your appointment in Cardiology at Cincinnati Children’s?
   - [ ] Yes
   - [ ] No

2. How did you first learn about genetic testing? (Please select one)
   - [ ] Genetic counselor
   - [ ] Geneticist
   - [ ] Cardiologist
   - [ ] Primary Care Physician
   - [ ] Support Group
   - [ ] News
   - [ ] Internet
   - [ ] Family member
   - [ ] Friend
   - [ ] Other

3. Has anyone in your family had genetic testing for cardiomyopathy?
   - [ ] Yes (circle all that apply)
     - [ ] You
     - [ ] Child
     - [ ] Sibling
     - [ ] Your child’s other biological Parent
     - [ ] Uncle
     - [ ] Aunt
     - [ ] Grandparent
     - [ ] Cousin
   - [ ] No (If No, skip to #8)

4. Who was the **first person** in the family to have genetic testing for cardiomyopathy?
   - [ ] Yourself
   - [ ] Your child’s other biological parent
   - [ ] Your child
   - [ ] Another family member related to your child (please circle)
     - Sibling, Parent, Aunt, Uncle, Grandparent, Cousin
   - [ ] No one has been tested
5. What were the genetic testing results for the first person tested?
   [ ] Disease causing mutation (a gene change was found and is known to be associated with cardiomyopathy)
   [ ] Variant of uncertain significance (a gene change was found, but unclear if it is associated with cardiomyopathy)
   [ ] No mutation was found (no gene change was identified by testing)
   [ ] Results are pending
   [ ] No testing
   [ ] Unsure of test results

6. What were the results of your genetic test?
   [ ] Disease causing mutation (a gene change was found and is known to be associated with cardiomyopathy)
   [ ] Variant of uncertain significance (a gene change was found, but unclear if it is associated with cardiomyopathy)
   [ ] No mutation was found (no gene change was identified by testing)
   [ ] Not had genetic testing
   [ ] No testing
   [ ] Results are pending

7. What were the results of your child’s genetic test?
   [ ] Disease causing mutation (a gene change was found and is known to be associated with cardiomyopathy)
   [ ] Variant of uncertain significance (a gene change was found, but unclear if it is associated with cardiomyopathy)
   [ ] No mutation was found (no gene change was identified by testing)
   [ ] Not had genetic testing
   [ ] No testing
   [ ] Results are pending

8. What were the results of your child’s other biological parent’s genetic test?
   [ ] Disease causing mutation (a gene change was found and is known to be associated with cardiomyopathy)
   [ ] Variant of uncertain significance (a gene change was found, but unclear if it is associated with cardiomyopathy)
   [ ] No mutation was found (no gene change was identified by testing)
   [ ] Not had genetic testing
   [ ] No testing
   Results are pending
9. Please circle the number that best represents the degree of importance the listed factor had on the decision for genetic testing for your child:

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<thead>
<tr>
<th></th>
<th>Very Important</th>
<th>Not Important</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cost of genetic testing</td>
<td>1 2 3 4 5</td>
<td></td>
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<tr>
<td>Help to prepare for the future</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Avoiding the worrying of uncertainty</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Fear of psychological reaction to the results</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>To confirm diagnosis in an affected individual</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Ability to share this information with extended family members</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Parents have the right to decided their child’s health care practices</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Avoidance of unnecessary screening of unaffected individuals</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Fear of discrimination (insurance, employment, society, etc)</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Child has the right to decide if they want to be tested</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
</tbody>
</table>

10. How did your perception of the severity of the condition change after genetic testing in the family?
   [ ] Condition seems more severe after testing
   [ ] Condition seem less severe after testing
   [ ] Severity of the condition is unchanged after testing
   [ ] No one in the family has had genetic testing

11. Did you or your child’s other biological parent share genetic testing results with family members?
   [ ] Yes
   [ ] No

12. Who did you or your child’s other biological parent share/discuss the test results with in your family?
   [ ] Your/ your child’s other biological parent’s siblings
   [ ] Your/ your child’s other biological parent’s parents
   [ ] Your/ your child’s other biological parent’s grandparents
   [ ] Your/ your child’s other biological parent’s first cousins
   [ ] Did not share results with family
13. How did you or your child’s other biological parent share the genetic testing results? (mark all that apply)
   [ ] Shared a letter from the genetic counselor with family members
   [ ] Shared a copy of the genetic testing results/report
   [ ] Spoke with family members in person about the results
   [ ] Discussed results with family by phone
   [ ] Emailed family members regarding the test results
   [ ] Shared results with one family member and asked them to share the results with other family members

14. Please circle the number that best represents the degree of importance the listed factor had on whether test results were shared with family members.

<table>
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<tr>
<th>Factor</th>
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<tr>
<td>Genetic counselor or geneticist said it was important to share</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Family members are interested in having testing</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>To educate family about cardiomyopathy</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Make family aware of the risk of sudden cardiac death</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Make family aware of other risks associated with cardiomyopathy</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Encourage family members to begin screening practices</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>It just came up in conversation</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
</tbody>
</table>

15. After sharing the information, do you know if anyone in the family saw a cardiologist?
   [ ] Yes
   [ ] No
   [ ] Did not share results

16. After sharing the information with the family, was anyone diagnosed with cardiomyopathy?
   [ ] Yes
   [ ] No
   [ ] Did not share results

17. After sharing the information, do you know if anyone in the family chose to have genetic testing?
   [ ] Yes
   [ ] No
   [ ] Did not share results
Section 5: Information about the screening recommendations for cardiomyopathy.
1. Was it recommended that family members undergo cardiac screening?
   [ ] Yes
   [ ] No (If no, skip to Section 6)

2. What screening procedures were recommended?
   [ ] Physical Exam
   [ ] Electrocardiogram (EKG)
   [ ] Echocardiogram (Echo)
   [ ] Holter Monitor
   [ ] Exercise treadmill test
   [ ] Not sure/can’t remember
   [ ] Other_____________________________________________________________________

3. For which of the following family members was screening recommended? (Please mark all that apply)
   [ ] First-degree relatives (Children, Siblings, Parents)
   [ ] Second-degree relatives (Aunt, Uncle, Niece, Nephew, Grandparent)
   [ ] Third-degree relatives (First Cousins)
   [ ] No one

4. Do you feel like you had a resource in cardiology to call with questions about screening?
   [ ] Yes
   [ ] No

5. Did you share the screening recommendations with family members?
   [ ] Yes
   [ ] No

6. Who did you share/discuss the screening recommendations with in your family? (Please mark all that apply)
   [ ] Your/ your child’s other biological parent’s siblings
   [ ] Your/ your child’s other biological parent’s parents
   [ ] Your/ your child’s other biological parent’s grandparents
   [ ] Your/ your child’s other biological parent’s first cousins
   [ ] Did not share recommendations with family

7. How did you or your child’s other biological parent share the screening recommendations? (Mark all that apply)
   [ ] Shared a letter from the genetic counselor with family members
   [ ] Spoke with family members in person about screening
   [ ] Discussed screening option with family by phone
   [ ] Emailed family members regarding screening recommendations
   [ ] Shared screening recommendations with one family member and asked them to share the results with other family members
8. Please circle the number that best represents the degree of importance the listed factor had on the why screening practices were shared with family members.

<table>
<thead>
<tr>
<th>Factor</th>
<th>Very Important</th>
<th>Not Important</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic counselor or geneticist said it was important to share</td>
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<td>2 3 4 5</td>
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<tr>
<td>Family members are interested in having genetic testing</td>
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<td>2 3 4 5</td>
</tr>
<tr>
<td>To educate family about cardiomyopathy</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Make family aware of the risk of sudden cardiac death</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Make family aware of other risks associated with cardiomyopathy</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Encourage family members to begin screening practices</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>In hopes that they would be understanding and accommodating</td>
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<td>2 3 4 5</td>
</tr>
<tr>
<td>It just came up in conversation</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
</tbody>
</table>

10. After sharing the screening recommendations, do you know if anyone of your blood relatives saw a cardiologist?
   [ ] Yes
   [ ] No

11. After sharing the information with the family, was anyone diagnosed with cardiomyopathy?
   [ ] Yes
   [ ] No

12. After sharing the information, do you know if anyone of your blood relatives chose to have genetic testing?
   [ ] Yes
   [ ] No

Section 6: The following is a set of statements about heart muscle disease (cardiomyopathy).
Select ONE answer for each question.

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<tr>
<th>Statement</th>
<th>True</th>
<th>False</th>
<th>Unsure</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Heart muscle disease is always caused by genetic factors.</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
</tr>
<tr>
<td>2. All family members who carry a gene mutation will develop heart muscle disease in their lifetime.</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
</tr>
<tr>
<td>3. A parent who carries a gene mutation can pass this mutation to their children.</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
</tr>
</tbody>
</table>
Non Tested

Section 1: Information about the parent completing the survey.

1. What is your gender?
   [ ] Male
   [ ] Female

2. What is your age?
   _______Years

3. Which of the broad categories best describes your family’s race?
   [ ] American Indian/Alaskan Native
   [ ] Asian
   [ ] Black or African American
   [ ] Native Hawaiian or other Pacific Islander
   [ ] White or Caucasian

4. How far did you get in school?
   [ ] Less then High School
   [ ] High School diploma or equivalent
   [ ] Some College
   [ ] College Graduate (Associate’s Degree, Bachelor's Degree)
   [ ] Master’s Degree
   [ ] PhD, MD or the like

5. What is your marital status?
   [ ] Single
   [ ] Married
   [ ] Separated/Divorced
   [ ] Widowed

6. Have you been diagnosed with one of the following?
   [ ] Heart muscle disease (cardiomyopathy)
   [ ] Heart rhythm problem (cardiac arrhythmia)
   [ ] Other heart problem _________________
   [ ] I don’t know

7. Has your child’s other biological parent been diagnosed with one of the following?
   [ ] Heart muscle disease (cardiomyopathy)
   [ ] Heart rhythm problem (cardiac arrhythmia)
   [ ] Other heart problem _________________
   [ ] I don’t know
8. How many biological children do you have?
   [ ] None     [ ] Three
   [ ] One      [ ] Four or more
   [ ] Two

9. How many of your biological children are followed by a cardiologist in the
   Children's Hospital Heart Institute Cardiomyopathy Clinic?
   [ ] None
   [ ] One
   [ ] Two
   [ ] Three
   [ ] Four or more

10. How many of your biological children have a diagnosis of: (Please circle)
   Heart muscle disease (cardiomyopathy)   0 1 2 3 4+
   Heart rhythm problem (cardiac arrhythmia) 0 1 2 3 4+

Section 2: Information about your first child to be evaluated by Cardiology at
Cincinnati Children's Hospital. (Not necessarily your first-born child)

1. Does your child have a diagnosis of cardiomyopathy? (If No skip to Section 3)
   [ ] Yes
   [ ] No

2. At what age did your child first experience symptoms?
   ___________ months old
   ___________ years old
   [ ] Never had symptoms

3. Please choose the symptoms experienced by your child:
   [ ] Shortness of breath
   [ ] Chest pain
   [ ] Abnormal heart rate
   [ ] Difficulty exercising
   [ ] Excessive sweating
   [ ] Fainting
   [ ] Heart Failure
   [ ] Other____________________________________________________________
   [ ] No symptoms

4. Has your child ever been admitted to the hospital to stay overnight because of
   their heart disease?
   [ ] Yes
      If yes, about how long was the stay________________________
   [ ] No
5. On a scale of 1 to 5 how severe do you feel your child's heart disease is; 1 being the least severe and 5 being the most severe?

1 2 3 4 5

6. At what age was your child diagnosed with cardiomyopathy?

______________prenatally (before birth)
______________months old
______________years old

7. What type of cardiomyopathy has been diagnosed in your child?

[ ] Hypertrophic Cardiomyopathy (HCM; thick left heart muscle)
[ ] Dilated Cardiomyopathy (DCM; large left pumping chamber)
[ ] Other _____________________
[ ] I don’t know
[ ] None

Section 3: Information about immediate and extended biological family members of the first child evaluated at Cincinnati Children’s.

1. How many people in the family, other than your child, have cardiomyopathy diagnosed by a doctor?

[ ] One
[ ] Two
[ ] Three
[ ] Four
[ ] Five or more
[ ] No one

2. What type of cardiomyopathy has been diagnosed in the family?

[ ] Hypertrophic Cardiomyopathy (HCM; thick left heart muscle)
[ ] Dilated Cardiomyopathy (DCM; large left pumping chamber)
[ ] Other _____________________
[ ] I don’t know
[ ] None

3. Which of the listed factors led to the initial diagnosis of cardiomyopathy in the first person in the family?

[ ] Heart murmur detected
[ ] Electrocardiogram (EKG)
[ ] Echocardiogram (Echo)
[ ] Symptoms (fainting, difficulty exercising, chest pain, excessive sweating, etc)
[ ] Sudden cardiac death of a family member
[ ] Other _____________________
4. How old was the **youngest person** in the family at the time of diagnosis?

   _______________ prenatally (before birth)
   _______________ months old
   _______________ years old

**Section 4:** Information about genetic testing for cardiomyopathy.

1. Had you heard about genetic testing for cardiomyopathy before your appointment in Cardiology at Cincinnati Children’s?
   - [ ] Yes
   - [ ] No

2. How did you first learn about genetic testing? (Please select one)
   - [ ] Genetic counselor
   - [ ] Geneticist
   - [ ] Cardiologist
   - [ ] Primary Care Physician
   - [ ] Support Group
   - [ ] News
   - [ ] Internet
   - [ ] Family member
   - [ ] Friend
   - [ ] Other

3. Has anyone in your family had genetic testing for cardiomyopathy?
   - [ ] Yes (circle all that apply)
     - [ ] You
     - [ ] Child
     - [ ] Sibling
     - [ ] Your Child’s Other Biological Parent
     - [ ] Uncle
     - [ ] Aunt
     - [ ] Grandparent
     - [ ] Cousin
   - [ ] No (If No, skip to #8)

4. Who was the **first person** in the family to have genetic testing for cardiomyopathy?
   - [ ] Yourself
   - [ ] Your child’s other biological parent
   - [ ] Your child
   - [ ] Another family member related to your child (please circle)
     - Sibling, Parent, Aunt, Uncle, Grandparent, Cousin
   - [ ] No one has been tested
5. Please circle the number that best represents the degree of importance the listed factor had on your decision regarding genetic testing for your child:

<table>
<thead>
<tr>
<th>Factor</th>
<th>Very Important</th>
<th>Not Important</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cost of genetic testing</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Help to prepare for the future</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Avoiding the worrying of uncertainty</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Fear of psychological reaction to the results</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>To confirm diagnosis in an affected individual</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Ability to share this information with extended family members</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Parents have the right to decided their child’s health care practices</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Avoidance of unnecessary screening of unaffected individuals</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Fear of discrimination (insurance, employment, society, etc)</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
<tr>
<td>Child has the right to decide if they want to be tested</td>
<td>1 2 3 4 5</td>
<td></td>
</tr>
</tbody>
</table>

6. What were the genetic testing results for the first person tested?
   - Disease causing mutation (a gene change was found and is known to be associated with cardiomyopathy)
   - Variant of uncertain significance (a gene change was found, but unclear if it is associated with cardiomyopathy)
   - No mutation was found (no gene change was identified by testing)
   - Results are pending
   - No testing
   - Unsure of test results

7. What were the results of your child’s genetic test?
   - Disease causing mutation (a gene change was found and is known to be associated with cardiomyopathy)
   - Variant of uncertain significance (a gene change was found, but unclear if it is associated with cardiomyopathy)
   - No mutation was found (no gene change was identified by testing)
   - Not had genetic testing
   - No testing
   - Results are pending

Section 5: Information about the screening recommendations for cardiomyopathy.

1. Was it recommended that family members undergo cardiac screening?
   - Yes
   - No (If no, skip to Section 6)

2. What screening procedures were recommended?
3. For which of the following family members was screening recommended? (Please mark all that apply)
   - First-degree relatives (Children, Siblings, Parents)
   - Second-degree relatives (Aunt, Uncle, Niece, Nephew, Grandparent)
   - Third-degree relatives (First Cousins)
   - No one

4. Do you feel like you had a resource in cardiology to call with questions about screening?
   - Yes
   - No

5. Did you share the screening recommendations with family members?
   - Yes
   - No

6. Who did you share/discuss the screening recommendations with in your family? (Please mark all that apply)
   - Your/ your child's other biological parent’s siblings
   - Your/ your child’s other biological parent’s parents
   - Your/ your child’s other biological parent’s grandparents
   - Your/ your child’s other biological parent’s first cousins
   - Did not share recommendations with family

7. How did you or your child’s other biological parent share the screening recommendations? (Mark all that apply)
   - Shared a letter from the genetic counselor with family members
   - Spoke with family members in person about screening
   - Discussed screening option with family by phone
   - Emailed family members regarding screening recommendations
   - Shared screening recommendations with one family member and asked them to share the results with other family members
8. Please circle the number that best represents the degree of importance the listed factor had on whether screening practices were shared with family members.

<table>
<thead>
<tr>
<th>Factor</th>
<th>Most Important</th>
<th>Least Important</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic counselor or geneticist said it was important to share</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Family members are interested in having genetic testing</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>To educate family about cardiomyopathy</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Make family aware of the risk of sudden cardiac death</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Make family aware of other risks associated with cardiomyopathy</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>Encourage family members to begin screening practices</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>In hopes that they would be understanding and accommodating</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
<tr>
<td>It just came up in conversation</td>
<td>1</td>
<td>2 3 4 5</td>
</tr>
</tbody>
</table>

10. After sharing the screening recommendations, do you know if anyone of your blood relatives saw a cardiologist?
   
   [ ] Yes
   
   [ ] No

11. After sharing the information with the family, was anyone diagnosed with cardiomyopathy?
   
   [ ] Yes
   
   [ ] No

12. After sharing the information, do you know if anyone of your blood relatives chose to have genetic testing?
   
   [ ] Yes
   
   [ ] No

**Section 6:** The following is a set of statements about heart muscle disease. Select ONE answer for each question.

1. Heart muscle disease is always caused by genetic factors.
   
   [ ] True
   
   [ ] False
   
   [ ] Unsure

2. All family members who carry a gene mutation will develop heart muscle disease in their lifetime.
   
   [ ] True
   
   [ ] False
   
   [ ] Unsure

3. A parent who carries a gene mutation can pass this mutation to their children.
   
   [ ] True
   
   [ ] False
   
   [ ] Unsure