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The Impact of Genetic Testing for Cardiomyopathy on Emotional Well-being and Family Dynamics: A Study of Parents and Adolescents

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Supplemental Methods
Supplemental Tables I–III
Supplemental Figure I
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Abstract

Background ––—Genetic testing is indicated for children with a personal or family history of hereditary cardiomyopathy to determine appropriate management and inform risk stratification for family members. The implications of a positive genetic result for children can potentially impact emotional wellbeing. Given the nuances of cardiomyopathy genetic testing for minors, this study aimed to understand how parents involve their children in the testing process and investigate the impact of genetic results on family dynamics.

Methods ––—A survey was distributed to participants recruited from the Children's Cardiomyopathy Foundation and seven North American sites in the Pediatric Cardiomyopathy Registry. The survey explored adolescent and parent participants' emotions upon receiving their/their child's genetic results, parent-child result communication and its impact on family functionality, using the McMaster Family Assessment Device.

Results ––—One hundred sixty two parents of minors and 48 adolescents who were offered genetic testing for a personal or family history of cardiomyopathy completed the survey. Parents whose child had cardiomyopathy were more likely to disclose positive diagnostic genetic results to their child (p value=0.014). Parents with unaffected children and positive predictive testing results were more likely to experience negative emotions about the result (p value=<0.001) but also had better family functioning scores than those with negative predictive results (p value=0.019). Most adolescents preferred results communicated directly to the child, but parents were divided about whether their child's result should first be released to them or their child.

Conclusions ––—These findings have important considerations for how providers structure genetic services for adolescents and facilitate discussion between parents and their children about results.

Keywords

cardiomyopathy; genetic testing; family study; pediatric; psychology and behavior

Journal Subject Terms:

Genetics; Health Services; Mental Health

Introduction

Cardiomyopathy is a heterogeneous heart disease that can vary in anatomy, age of onset, and associated symptoms. Pediatric cardiomyopathy has an annual incidence of 1–1.5 per 100,000 children[1]. It can be isolated or part of a syndrome that includes extracardiac features. There are currently over 100 genes that cause cardiomyopathies in children, many more than in adults[2], and most of the causative genes are inherited in an autosomal dominant manner[1]. Cardiomyopathy genetic testing (CGT) in children has utility to confirm

a diagnosis, clarify prognosis, anticipate extracardiac manifestations, determine eligibility for disease-specific therapies or clinical trials, inform risk for other family members, and provide information to assist reproductive decisions[3,4,5]. Identifying asymptomatic children at risk for cardiomyopathy facilitates appropriate surveillance and identification of early cardiac disease to guide medical management including restrictions in physical activity and use of medications[4, 6,7].

While there are benefits of CGT, the psychological impact of genetic results on families is understudied. This is especially true for adolescents, who are in a phase of life contending with issues of identity and autonomy. A study evaluating the wellbeing of children between the ages of 8–18 years with positive cardiac genetic testing showed that there was no difference in their overall psychological health compared to the control children group[8]. However, previous studies of cancer genetic testing among minors identified psychosocial concerns involving family tensions that could arise from the genetic testing result[9]. In one study, girls between 6–13 years of age with a family history of breast cancer had higher disease-specific distress than their peers, which was strongly associated with poorer family communication[10].

Genetic testing in minors is complicated as parents are entrusted to provide consent for their testing, even though some children and most adolescents are cognitively capable of understanding their results and its implications[11]. Issues of autonomy become increasingly important as adolescents age, yet existing laws require parental consent but not adolescents' consent[12]. Prior studies of adolescents who underwent diagnostic genetic testing indicated a strong desire to be involved in the process of making decisions about testing and return of results[7,13,14]. A recent study surveying adolescents with congenital heart disease and parents highlighted that the majority in both groups felt it was important for adolescents to know their genetic risk to prevent gaps in cardiac care as they transition to adulthood[15.] Studies in other contexts of genetic testing suggest that parents desire to determine the time and manner of disclosing genetic results to their child[14], notwithstanding evidence that parents lack the necessary medical and genetic knowledge to discuss such information accurately with their child[8,16,17]. Understanding of family dynamics can help medical providers facilitate result communication between the child and family.

Given the development of health practices and behaviors during formative years of childhood, it is important for adolescents' long-term compliance with medical care to understand the impact of CGT on themselves and their families and to develop effective methods to support them through the testing process. Our multi-center study aimed to better understand how families involve minors in the genetic testing and result communication processes by conducting a survey of adolescents and parents of children who were offered CGT. We examined (1) how minors are involved in the genetic testing process, (2) family communication of genetic testing, and (3) the psychosocial impact of result disclosure on adolescents and parents.

Methods

Detailed methodology is available in the Methods section of the Supplemental Materials. The study was approved by the institutional review boards at each of seven study sites that were part of the Pediatric Cardiomyopathy Registry and centrally at Columbia University. Informed consent and assent was obtained from all parent and adolescent participants respectively.

The data that support the findings of this study are available from the corresponding author upon reasonable request.

Results

In total, 162 parents from unique families completed the parental survey. Eighty-one parent participants consented for their adolescent child to participate. Of those, 48 adolescents between the ages of 13–18 years completed the survey for an adolescent response rate of 59%. The number of participants in each sub-cohort based on type of participant and their clinical cardiomyopathy status is shown in Figure 1.

Parents

Demographic data from the 162 parent respondents included in this study are shown in Table 1. One hundred and fifty-four parents had a child who underwent CGT: 100 parents had a child who underwent diagnostic genetic testing (DGT) and were classified as Group1; the remaining 54 parents had a child who underwent predictive genetic testing (PGT) and were classified as Group 2. The clinical and genetic test results for parent participants and their children are summarized in Table 2. Among the parents recruited from the hospitals, there was a 97.3% concordance rate between reported responses on their child's genetic test result and medical records; discordant responses were limited to three parents whose child had a variant of uncertain significance (VUS) but who reported positive results.

Communication with children

While the majority of parents in both Group 1 (n=70; 70%) and Group 2 (n=41; 75.9%) reported their child was informed that they were going to have CGT, the proportion of parents who actively involved their child in the decision making process to have testing was higher in Group 1 (n=24; 24%) than in Group 2 (n=7; 12.9%). See Table 1 in the Data Supplement for total responses on involvement of children in the testing process.

Among the 100 parents in Group 1, 88 were aware of and could recall their child's result at the time of the survey. The majority of these parents (n=62; 70.5%) reported they had communicated the result to their child, and this was strongly associated with the type of result (i.e positive/negative/ VUS) received- 79.7% of parents of children with positive DGT results had disclosed results to their child compared to 57.1% with negative DGT results and 37.5% with VUS DGT results (p value=0.014). Though the proportion of parents in Group 2 who shared PGT results with their child was similar (n=31; 66%), there was no significant association between the parent-child communication of result and the type of

result (p value=0.591). Disclosure of results to the child was significantly associated with the child's age at testing in both Group 1 (mean (IQR) = 11 (8–13) years for disclosure vs 4 (1.5–10.5) years for non-disclosure; p value=0.002) and Group 2 (mean (IQR) = 12 (9–13) years for disclosure vs 4 (0.375, 9) for non-disclosure; p value=<0.001.)

Communication with extended family and impact on family dynamics (Table 3)

Almost equal proportions of parents in Group 1 did (n=37; 42%) and did not share (n=41; 46%) DGT results with the extended family, and the type of result obtained did not impact their sharing. Among Group 2, 72.3% (n=34) of parents shared PGT results with extended family, and a higher percentage of parents (88.3%) shared positive PGT results compared to negative PGT results (66.6%) (p value=0.042).

Of all 67 parents from Groups 1 and 2 who shared results, only six parents (9%) reported that knowledge of results negatively impacted their family dynamics. These six parents all had a child with positive CGT results. The remaining respondents felt that the results did not affect the family (n=13; 19.4%), affected it positively (n=24; 35.8%), or were unsure if anything changed in their family dynamics (n=24; 35.8%).

The average McMaster Family Assessment Device (MFAD) functionality score for Group 1 was 1.9 (out of a maximum of 4), with average scores of 1.9, 1.8 and 1.8 for families with positive, negative and VUS DGT results, respectively. A score of ≥2 indicates unhealthy family functioning. The genetic results of DGT was not significantly associated with how well a family functioned in Group 1 (% healthy functioning families were 49%, 52% and 49% for positive, negative and VUS DGT results, respectively, p value=0.842). The average MFAD functionality score for Group 2 was 1.7, with average scores of 1.6, 1.8 and 1.7 for families with positive, negative and VUS PGT results, respectively. Among Group 2, there was a significant difference in the functionality of families based on genetic results. Ninety-one percent (n=22) of families with positive PGT results met criteria for healthy family functionality while 57% (n=12) of families with negative PGT results and 100% (n=2) of families with VUS results met criteria for healthy functioning (p value= 0.019).

Feelings about genetic test results (Figure 2)

There was no significant association of feelings after receiving the child's result with the type of result in Group 1 (p value=0.189), although a majority of parents in this group expressed negative emotions after their child had a positive (n=38; 64.4%) or uncertain (n=5; 62.5%) DGT result. The responses of the 21 parents whose child had a negative DGT result were more varied, but the most commonly reported emotions were still negative. There was a significant association between the feelings of parents in Group 2 and the type of result their child had (p value=<0.001). The majority of parents whose child had a positive PGT result reported negative emotions (n=13; 61.9%) and inversely, the majority of parents whose child had a negative PGT result reported positive emotions (n=15; 62.5%).

We also investigated if cardiomyopathy status in the 135 parents who knew their child's genetic test result affected the way they felt about the result. Six (6.8%) of the 88 parents in Group 1 had cardiomyopathy and 14 (29.7%) of the 47 parents in Group 2 had cardiomyopathy. There was no significant association between parental cardiomyopathy status and feelings about the child's genetic result for Group 1 (p value=0.425). However, there was a trend for significant association between parents' cardiomyopathy status in Group 2 and their feelings about their child's result (p value=0.052). In both Groups 1 and 2, more parents who had cardiomyopathy had negative feelings about their child's result. Among Group 1, 83.3% (n=5) of parents who had cardiomyopathy had negative feelings while 57.3% (n=47) of parents who did not have cardiomyopathy had negative feelings. Among Group 2, 64.3% (n=9) of parents who had cardiomyopathy had negative feelings compared to 27.3% (n=9) of parents who did not have cardiomyopathy and had negative feelings. See Table 2 in the Data Supplement for comparison of parental feelings towards their child's result with their own cardiomyopathy status.

Adolescents

A total of 48 participants completed the adolescent survey. The mean age was 16.7 years. The demographics are summarized in Table 1. Forty-one (85%) adolescent participants had undergone CGT. Twenty-one adolescents had DGT (Group 3) and 20 adolescents had PGT (Group 4). Table 2 summarizes the clinical and genetic test results for adolescent participants. Among adolescents recruited from the hospital sites, there was a 93.7% concordance between reported responses on their genetic test result and medical records with three participants who had a VUS reporting that they had positive results. Two of these participants' parents also misreported their child's VUS as a positive result in their parental survey responses. This indicates that these adolescents correctly reported the information given to them by their parents.

Communication with family members and impact on family dynamics

While the majority (n=30; 80.4%) of the 41 adolescent participants who had CGT were informed they were having genetic testing during the pre-test counseling prior to their sample collection, the number of adolescents who were actively involved in the decision making process to have CGT was higher in Group 3 (n=12; 57.5%) than in Group 4 (n=4; 20%). See Table 1 in the Data Supplement for total responses on involvement of adolescents in the testing process.

A majority of the 34 adolescent participants from both Groups 3 and 4 who knew their results reported that their results were shared with relatives (n=30; 88.2%) and 21 of those respondents (70%) said they were the ones who shared the results with other family members. The type of result was not significantly associated with sharing results (p value=1 for Group 3 and p value= 0.464 for Group 4). Majority of our participants reported that adolescent patients are most likely to discuss their results with their mother (n=31; 64.5%), while fewer felt they would discuss results with their father (n=5; 10.4%), their doctor (n=5; 10.4%), their best friend (n=8; 6%) or someone else (n=1; 2.08%).

In response to whether knowledge of results affected dynamics in the 30 families that shared the adolescent's result with relatives, only two (6.6%) felt that it had made things worse. Both the participants who felt things were worse had positive results and one of them further commented that the knowledge "increased stress levels, however not knowing was far more harmful." The type of genetic test result was not significantly associated with family functioning for adolescents in Groups 3 and 4 (p value=1 for both). The average MFAD score for Group 3 was 1.59, and the average score for Group 4 was 1.8.

Feelings about genetic test results (Figure 2)

Among the 21 adolescent participants in Group 3, 17 could remember the result of their DGT at the time of the survey, and there was no significant association between type of result and emotions felt after learning of their result (p value= 0.457). Seventeen of the 20 adolescents in Group 4 were aware of their results. While not significant, there was a trend of emotions being associated with type of result among this group (p value=0.089); most of the 11 participants with negative PGT results felt positive emotions after learning their result (n=9; 81.8%). When comparing parental cardiomyopathy status with adolescents' feelings about their results, a significant association was noted for Group 3 (p value=0.041) but not for Group 4 (p value=0.862). Among Group 3, the majority of adolescents (n=7; 63.7%) whose parents had cardiomyopathy reported negative feelings.

Overall, the most commonly reported feelings among adolescents in both Groups 3 and 4 prior to testing were negative (n=16; 47%) and neutral emotions (n=11; 32.3%) while fewer reported positive (n=4; 11.7%) and mixed (n=3; 8.8%) emotions. The most frequently reported post-test emotions were positive (n=13; 38.2%). Most of the eight participants who had negative feelings post-testing (n=5; 62.5%) reported that they now felt better about their results, and the remainder did not feel differently. Comparison of emotions in pre- and post-test result stages for adolescents in summarized in Table 3 in the Data Supplement.

Of all 34 adolescent respondents with and without cardiomyopathy who were aware of their results, 31 (91.1%) were glad to know their results, two (5.8%) were 'not sure' and one (2.9%) did not answer the question.

Parent-Adolescent comparisons

We asked all parent and adolescent participants (N=210) who they believed should receive results from CGT of children between the ages of 13–18 years (Figure 3). Fifty percent of parents (n=81) said that the parents should get the results first and the child later, while 40.7% of parents (n=66) said that the child and the parents should receive results at the same time. The responses from adolescent participants were markedly different: 70.8% of adolescents (n=34) thought that the child and the parents should receive results at the same time. Only 16.6% (n=8) believed that the parents should get the child's results first and the child later.

Parent and adolescent participants were then asked the minimum age at which they felt a child should have the autonomy to decide whether to pursue CGT for themselves (Figure 4). About 30.8% of parents (n=50) felt that the decision should be based on maturity rather than

chronological age, and a similar number of parents (n=49; 30.2%) felt that 13–16 years is the age range at which someone should have the autonomy to decide if they want CGT. A higher proportion of adolescents (n=21; 43.7%) felt that the ability to decide to have CGT should be based on level of maturity and not age.

Discussion

Due to the risk for sudden death with cardiomyopathy and the ability to initiate medical therapy with first signs of disease, it is recommended that genetic testing be considered for children with cardiomyopathy or a family history of hereditary cardiomyopathy[18] to determine surveillance approaches and preventive measures to mitigate risks. Compliance with medical recommendations is key to reducing the risk of serious health consequences but can be challenging, especially for adolescents who seek autonomy over aspects of their lives, including health and lifestyle choices and decisions about sports participation. It is important to better understand the impact of genetic testing on adolescents and investigate best practices to engage them in the decision-making process of genetic testing for cardiomyopathy. Our study sought to recognize how minors are included in the pre-test counseling process and identify how genetic testing impacts the emotions of parents and adolescents and the dynamics of their families.

Impact on families

Our results show that families have different experiences depending on the clinical status of their child. Most parents whose child had cardiomyopathy had negative feelings about their child's DGT result, whether it was positive, negative or uncertain. We also found that more parents who have cardiomyopathy are likely to feel negatively about their child's CGT results. Among adolescents with positive DGT results, there were more neutral feelings. This is in line with previous studies demonstrating that significant psychological distress is not significantly associated with positive genetic results in probands with cardiomyopathy given that a positive result currently does not typically alter prognosis or treatment for most individuals who already have a clinical diagnosis[19,20]. Based on our findings however, having a parent with cardiomyopathy may negatively impact the way adolescents feel about their DGT result. The parents' overall negative feelings may also be reflective of their own burden of caregiving as well as concern for the child's quality of life related to their cardiac disease rather than the test results[8]. However, among parents of children with PGT, those whose child had a negative result reported predominantly positive emotions, while parents whose child had a positive PGT result were more likely to report negative emotions. This same pattern was seen in adolescents with PGT, indicating that emotions felt in response to the PGT results are related to whether or not the potential genetic risk for cardiomyopathy is present. Despite the differing emotions after receiving the test results, nearly all adolescents were glad they knew their results, regardless of the outcome. This is consistent with previous research showing low regret among minors over the decision to have CGT[21]. It also supports work affirming that the knowledge from testing results can make adolescents feel empowered[7,9,22].

There was a significant and perhaps unexpected difference in level of healthy family functioning among families with children who had PGT that correlated with genetic test results. Families with positive PGT results had better functioning scores than families with negative PGT results. Considering that this group represents children without cardiomyopathy, it is reassuring to see that positive predictive results were not significantly associated with dysfunctional family dynamics. For the majority of families, the dynamics of the family were perceived by participants to be preserved or improved after knowledge of genetic test results. This is an important finding of the study as there is limited data on the impact of genetic testing on family dynamics.

Communication among family members

The majority of families in our study reported good communication between parent and child in the pre- and post-testing stages. In both parent groups, the child's age was significantly associated with the likelihood of the parent disclosing their result to them. There was also a significant trend for more parents to disclose results to their child with cardiomyopathy when the results were positive, perhaps because positive results may impact future reproductive decisions. Overall, our parent cohort stated that the strongest reason they would not disclose results to their child is that the knowledge might be stressful or anxiety-inducing for their child. Prior studies in the context of cardiac and non-cardiac disease have shown that most parents desire to have conversations with their child about their genetic risk but feel they lack the skill to approach the subject[15–17,23]. Other stated reasons have included lack of parental access to appropriate knowledge resources[22], preference for their child to receive genetic education from a medical provider[15], perceived risk of disease for the child[17], and distress between parents regarding making a unified decision about when to have such conversations with children[16]. Medical providers should help facilitate communication about genetic health between parents and children. Standardized educational materials for parents to discuss information with their children would be helpful.

Autonomy

In general, adolescent and parent participants shared similar views about the age when a minor should make the decision to have genetic testing. Both weighed individual maturity level rather than chronological age. However, parents and adolescents expressed different opinions on who should be the first person to receive a child's genetic result. Most adolescent children would like to be informed of their results at the same time as their parents, demonstrating a desire to be treated equally about issues related to their health. At the same time, the fact that they did not feel the need to know results before their parents may indicate they value having the support of a parent when receiving results. The difference in parents' and adolescents' opinions regarding this is important to explore. Previous work has highlighted that the opportunity to make choices during the pre-test counseling helps adolescents maintain empowerment and adapt better to information about health threats[21].

Study limitations

The study is limited by the modest sample size and demographically homogenous population of largely non-Hispanic, Caucasian participants. There may be important cultural differences in family dynamics and feelings about genetic results in other racial/ethnic groups.

Because most participants were enrolled retrospectively, the time period between receiving genetic results and completing the survey varied from a few months to a few years. It is possible that this time lag may have affected the ability to recall how they felt after receiving results. Future studies can prospectively investigate the impact of testing on families to better understand how time affects their experiences.

Most of the parents who participated had children who were affected with cardiomyopathy and had positive genetic test results; those families may have been more inclined to participate in a study of genetic testing.

Medical records could not be reviewed from the 46 parent respondents who were recruited through the Children's Cardiomyopathy Foundation (CCF), so their responses to clinical status and genetic testing questions were not confirmed.

Conclusion and considerations for practice

Genetic counselors and other healthcare providers who provide care to children undergoing CGT should consider taking a family-centered approach to counseling. When feasible, including adolescents during results disclosure is preferred. As parents may express concerns about their adolescent's age, maturity level, clinical status, and social stressors, discussion with genetic professionals can support families to optimize the timing of testing and utility of knowing this information.

A clear plan for how results will be disclosed should also be agreed upon during pre-test counseling, including whether the adolescent will be present for the results disclosure at the same time as their parents. Providing adolescents and their parents information to understand the genetic results in stages over time is helpful, and parents can continue to reinforce key messages as children get older and are able to better understand how their lives are impacted. Exploring existing dynamics to identify a family member who is trusted and close to the adolescent can help facilitate these conversations. For our adolescent cohort, mothers were identified to be the best family member to discuss results with. Follow up after results disclosure with the family is encouraged to address questions and identify misunderstanding of concepts.

Genetic testing can have implications for the patient and the family as a whole. It is important to understand how children and adolescents are involved when dealing with genetic testing in minors and the impact the results can have on the family. While our sample size is modest, it demonstrates the importance of engaging adolescents in conversations about their genetic health. Overall, our results suggest that genetic testing for cardiomyopathies in children and adolescents does not cause emotional harm to families or

adversely impact family dynamics. Further research into the adolescent perspective on the impact of genetic testing is needed and can be valuable in informing healthcare practitioners about how to best provide care to young adult patients.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Non-standard abbreviations and acronyms used:

PCMR	Pediatric Cardiomyopathy Registry
CCF	Children's Cardiomyopathy Foundation
CGT	Cardiomyopathy Genetic Testing
DGT	Diagnostic Genetic Testing
PGT	Predictive Genetic Testing
VUS	Variant of uncertain significance
MFAD	McMaster Family Assessment Device

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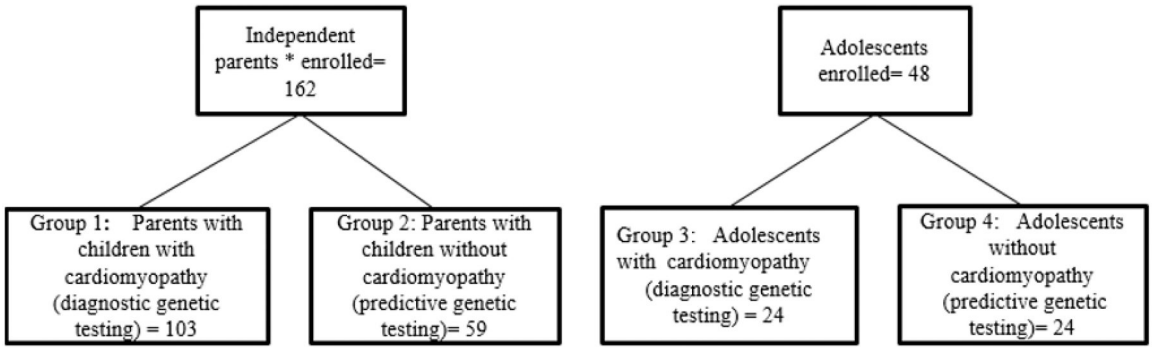
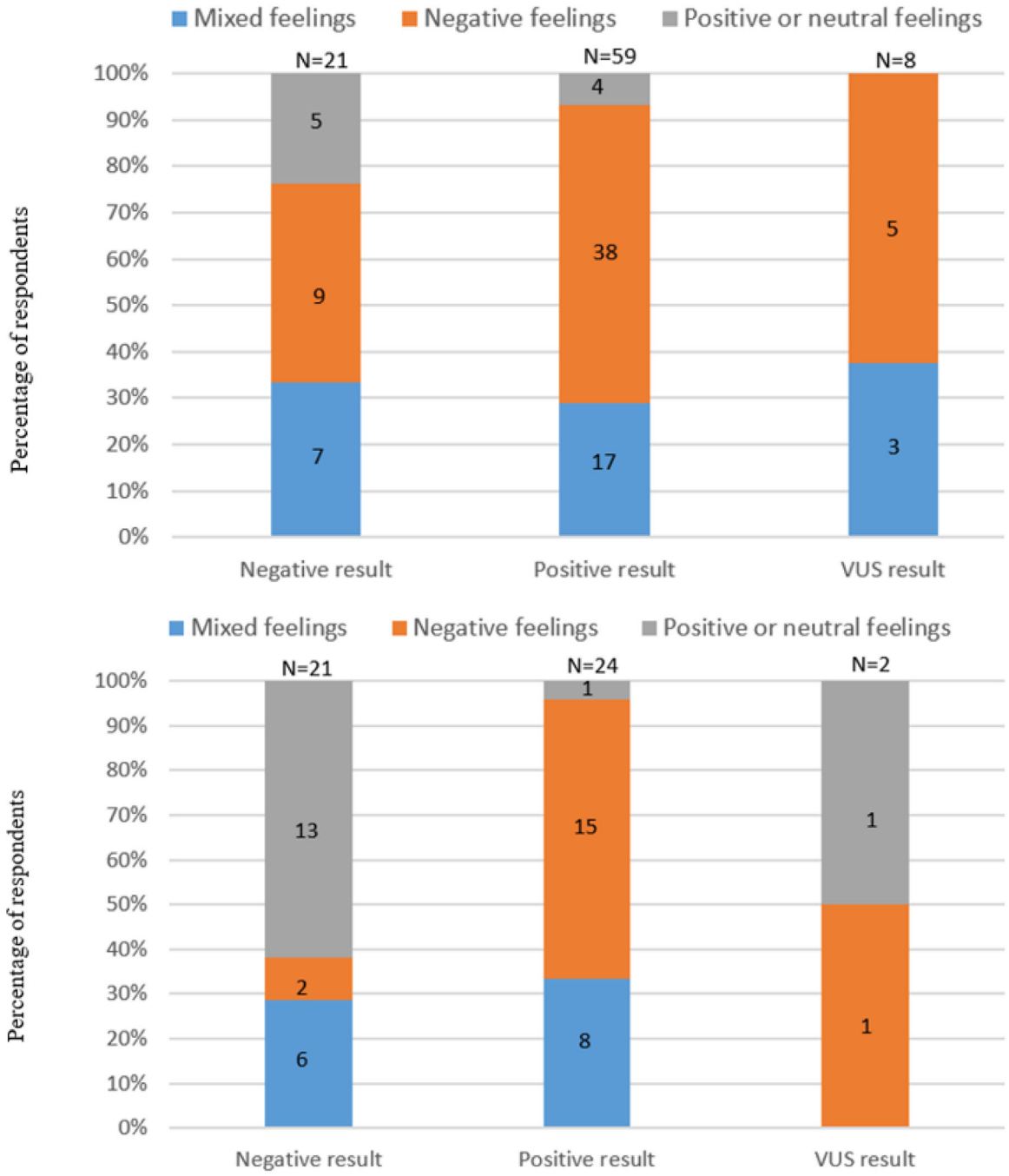
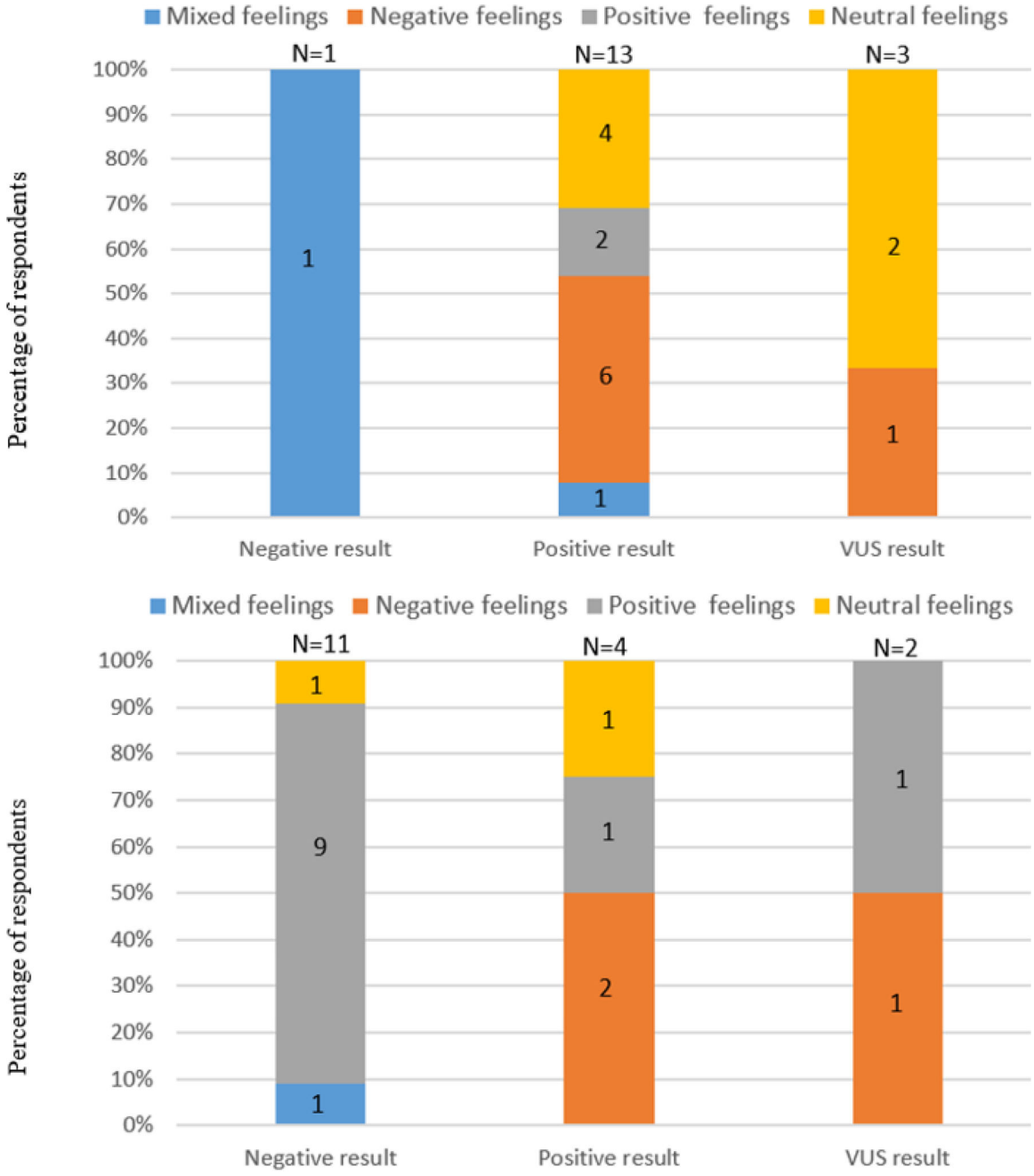


Figure 1:

Participant sub-cohorts. Classification of sub-cohorts based on cardiomyopathy genetic testing status used for data analysis. *Independent parent indicates only a single parent was enrolled from each family.





Figures 2(A-D):

Feelings of participants correlated with type of genetic test result received, i.e., positive results (pathogenic variant identified), negative result (no pathogenic variant identified) or variant of uncertain significance. Feelings were classified into categories of positive (*happy, glad etc.*), negative (*sad, angry etc.*) and neutral (*indifferent, curious etc.*) **A:** Reported feelings of parents in Group 1 after receiving their child's diagnostic genetic result. N=88; No significant association was seen between the feelings of parents and the type of result their child with cardiomyopathy had (p value=0.189), though the majority of parents whose child had a positive (64.4%) or uncertain result (62.5%) reported negative feelings. B:

Reported feelings of parents in Group 2 after receiving their child's predictive genetic result. N=47; There was a significant association between the feelings of parents and the type of result their child without cardiomyopathy had (p value=<0.001). Most parents whose child had a negative result (61.9%) reported positive feelings and most parents whose child had a positive result (62.5%) reported negative feelings. C: Reported feelings of adolescents in Group 3 after receiving their diagnostic genetic result. N=17; No significant correlations between feelings of adolescents with cardiomyopathy and their genetic test result (p value=0.457).D: Reported feelings of adolescents in Group 4 after receiving their predictive genetic result. N=17; No significant correlations between feelings of adolescents with cardiomyopathy and their genetic test result (p value=0.089). Most with negative results (81.8%) reported positive feelings.

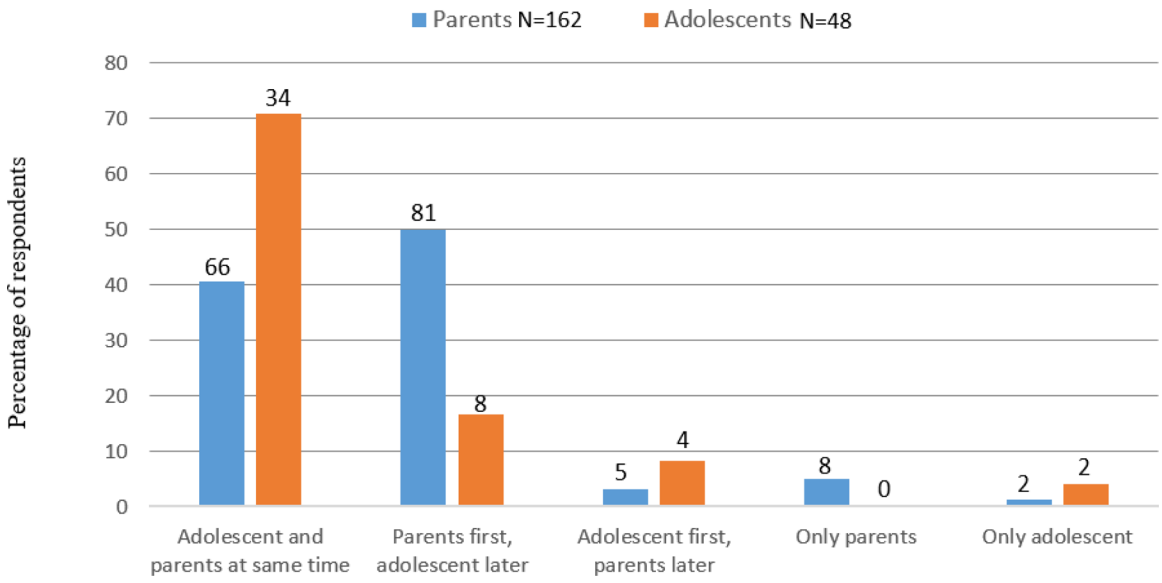


Figure 3:

Parents vs. adolescent responses about order and timing of genetic results disclosure. All 210 parent and adolescent participants (including those who did not have genetic testing) were surveyed about preference for child's result disclosure. The 162 parents were split between parents getting the child's result first and the child later (50%) and the child and the parents receiving results at the same time (40.7%). Whereas, the majority of adolescents (70.8%) felt the child and the parents should receive results at the same time and 16.6% believed that the parents should get the child's results first.

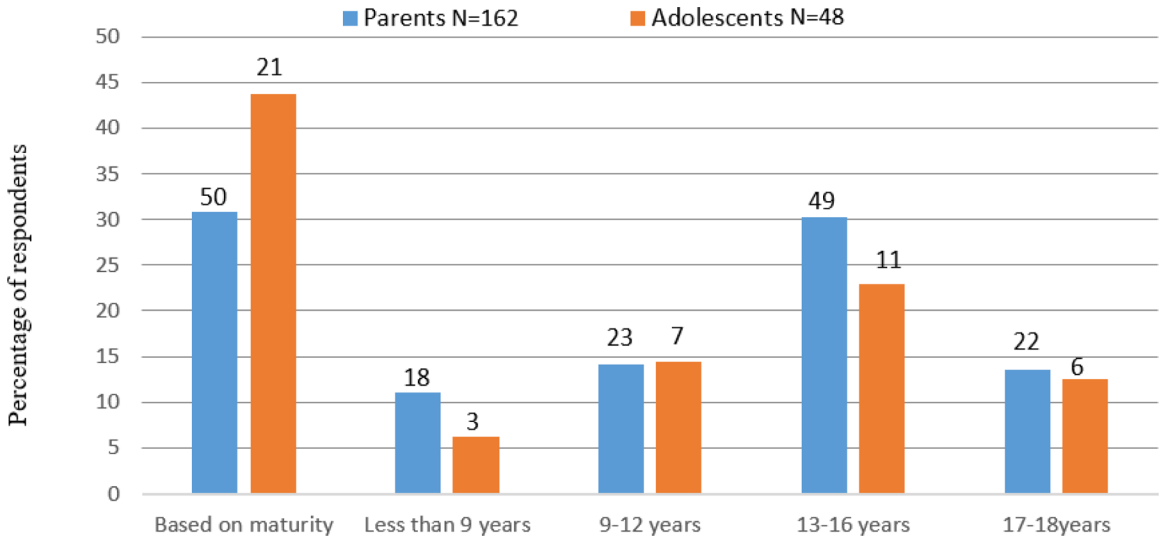


Figure 4:

Parents vs. adolescent responses about the minimum age for autonomous decision making about genetic testing. All 210 parent and adolescent participants (including those who did not have genetic testing) were surveyed about the age at which they felt someone can could autonomously decide to have genetic testing. Most parents were split between opinions of basing the decision on maturity rather than age (30.8%) and 13–16 years being the minimum age to make an independent decision (30.2%). A similar trend was observed in adolescents where many felt that the decision should be based on maturity rather than age (42.7%) or that the decision could be made at 13–16 years of age (22.9%).

Table 1.

Demographics of participants

Demographics of parent participants	**N= 162**	**Demographics of adolescent participants**	**N=48**
Average age (years) at time of survey	41.5 (SD=7.3)	Average age (years) at time of survey	16.7 (SD=2.6)
Sex			
Male	65 (40.2%)	Male	24 (50%)
Female	97 (59.8%)	Female	24 (50%)
Race/ethnicity			
Caucasian	122 (75.3%)	Caucasian	41 (85.4%
Hispanic	15 (9.2%)	Hispanic	3 (6.2%)
Asian or Pacific Islander	8 (5%)	Asian or Pacific Islander	2 (4.1%)
Black or African American	5 (3%)	More than one race	2 (4.1%)
More than one race	4 (2.4%)		
Other	4 (2.4%)		
Native Hawaiian or other Pacific Islander	2 (1.2%)		
American Indian or Alaskan Native	1 (0.6%)		
Highest education completed			
Some high school	10 (6.2%)	Less than grade 8	10 (20.8%)
High school or GED	21(13%)	Grade 9	10 (20.8%)
Some college/vocational school	31(19.2%)	Grade 10	11(22.9%)
College graduate	56 (34.5%)	Grade 11	9 (18.8%)
Master’s degree	33 (20.3%)	Grade 12	3 (6.2%)
Doctoral degree	7 (4.3%)	Some college/vocational school	5 (10.4%)

Table 2.

Reported cardiomyopathy (CM) status and genetic testing results of participants

Clinical and genetic testing status of parent participants (N=162)*				
Clinical status	**Had CM genetic testing**	**Positive result**	**Negative result**	**Uncertain† result**
Diagnosed with CM (n=23)	22	19	1	2
Not diagnosed with CM (n=129)	60	21	21	18
Clinical and genetic testing status of child of parent participants (N=162)*				
Clinical status	**Had CM genetic testing**	**Positive result**	**Negative result**	**Uncertain† result**
Child diagnosed with CM (n=103)	100	59	21	8
Child Not diagnosed with CM (n=59)	54	24	21	2
Clinical and genetic testing status of adolescent participants (N=48)*				
Clinical status	**Had CM genetic testing**	**Positive result**	**Negative result**	**Uncertain† result**
Diagnosed with CM (n=24)	21	13	1	3
Not diagnosed with CM (n=24)	20	4	11	2

*N is discordant with response numbers because 10 parent participants did not answer if they were affected with cardiomyopathy, 19 parent participants did not know/could not recall their child's result, 7 adolescents did not know/could not recall their result

†Uncertain result indicates variant of uncertain significance was identified

Table 3:

Parent responses on family communication and impact on family dynamics correlated to genetic results

Group 1: Parents who knew child's DGT result (N=88)					
Result communication and impact on family	**Negative results (N=21)**	**Positive results (N=59)**	**Uncertain* results (N=8)**	**Total (N=88)**	**P value**
Communication with child					
Medium age of child at disclosure with whom results were shared (IQR)	11 (8–13)			10 (5–13)	0.002
Medium age of child at testing with whom results were not shared (IQR)	4 (1.5, 10.5)				
Communicated result to child	12 (57.1%)	47 (79.7%)	3 (37.5%)	62 (70.5%)	0.014
Did not communicate result to child	9 (42.9%)	12 (20.3%)	5 (62.5%)	26 (29.5%)	
Communication with family					
Shared with other family	8 (38.1%)	26 (44%)	3 (37.5%)	37 (42%)	0.827
Did not shared with other family	11 (52.1%)	25 (42.4%)	5 (62.5%)	41 (46.5%)	
Unsure if it was shared	2 (9.5%)	8 (13.6%)	0 (0%)	10 (11.3%)	
MFAD scores †					
Healthy functioning (<2)	11 (52.4%)	29 (49.2%)	3 (37.5%)	43 (48.9%)	0.842
Unhealthy functioning (≥2)	10 (47.6%)	30 (50.8%)	5 (62.5%)	45 (%1.1)	
Average (SD)	1.832 (0.640)	1.923 (0.620)	1.839 (0.642)	1.894 (0.621)	
Group 2: Parents who knew child's PGT result (N=47)					
Communication with and impact on family	**Negative results (N=21)**	**Positive results (N=24)**	**Uncertain* results (N=2)**	**Total (N=47)**	**P value**
Communication with child					
Median age of child at disclosure with whom results were shared (IQR)	12 (9–13)			10(5.5, 13)	< 0.001
Medium age of child at testing with whom results were not shared (IQR)	4 (0.375, 9)				
Communicated result to child	13 (61.9%)	17 (70.8%)	1 (50%)	31 (66%)	0.591
Did not communicate result to child	8 (38.1%)	7 (29.2%)	1 (50%)	16 (34%)	
Communication with family					
Shared with other family	14 (66.7%)	20 (83.3%)	0 (0%)	34 (72.3%)	0.042
Did not shared with other family	5 (23.8%)	4 (16.7%)	2 (100%)	11 (23.4%)	
Unsure if it was shared	2 (9.5%)	0 (0%)	0 (0%)	2 (4.3%)	
MFAD scores †					
Healthy functioning (<2)	12 (57.1%)	22 (91.7%)	2 (100%)	36 (76.6%)	0.019
Unhealthy functioning (≥2)	9 (42.9%)	2 (8.3%)	0 (0%)	11 (23.4%)	
Average (SD)	1.886 (0.660)	1.630 (0.420)	1.708 (0.177)	1.748 (0.543)	
Group 3: Adolescents who knew their DGT result (N=17)					
Communication with and impact on family	**Negative results (N=1)**	**Positive results (N=13)**	**Uncertain* results (N=3)**	**Total (N=17)**	**P value**
Communication with family					

Group 1: Parents who knew child's DGT result (N=88)					
Result communication and impact on family	**Negative results (N=21)**	**Positive results (N=59)**	**Uncertain* results (N=8)**	**Total (N=88)**	**P value**
Shared with other family	1 (100%)	8 (61.5%)	2 (66.7%)	11 (64.7%)	1.000
Did not shared with other family	0 (0%)	5 (38.4%)	1 (33.3%)	6 (35.3%)	
MFAD scores †					
Healthy functioning (<2)	1 (100%)	11 (84.6%)	3 (100%)	15 (88.2%)	1.000
Unhealthy functioning (≥2)	0 (0%)	2 (15.4%)	0 (0%)	2 (%11.8)	
Average (SD)	2.000 (NA)	1.697 (0.445)	1.083 (0.083)	1.593 (0.463)	
Group 4: Adolescents who knew their PGT result (N=17)					
Communication with and impact on family	**Negative results (N=11)**	**Positive results (N=4)**	**Uncertain* results (N=2)**	**Total (N=17)**	**P value**
Communication with family					
Shared with other family	5 (45.5%)	3 (75%)	2 (100%)	10 (58.8%)	0.464
Did not shared with other family	6 (54.5%)	1 (25%)	0 (0%)	7 (41.2%)	
MFAD scores †					
Healthy functioning (<2)	7 (63.6%)	4 (100%)	0 (0%)	11 (64.7%)	0.063
Unhealthy functioning (≥2)	4 (36.4%)	0 (0%)	2 (100%)	6 (35.3%)	
Average (SD)	1.780 (0.681)	1.729 (0.322)	2.129 (0.054)	1.809 (0.570)	

*Uncertain result indicates variant of uncertain significance was identified

†MFAD score refers to score from the G12 subscale of the McMaster Family Assessment Device