

ARTICLE

Attitudes of Dilated Cardiomyopathy Patients and Investigators Toward Genomic Study Enrollment, Consent Process, and Return of Genetic Results

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Precision medicine genetics study design requires large, diverse cohorts and thoughtful use of electronic technologies. Involving patients in research design may increase enrollment and engagement, thereby enabling a means to relevant patient outcomes in clinical practice. Few data, however, illustrate attitudes of patients with dilated cardiomyopathy (DCM) and their family members toward genetic study design. This study assessed attitudes of 16 enrolled patients and their family members (P/FM), and 18 investigators or researchers (I/R) of the ongoing DCM Precision Medicine Study during a conjoint patient and investigator meeting using structured, self-administered surveys examining direct-to-participant enrollment and web-based consent, return of genetic results, and other aspects of genetic study design. Survey respondents were half women and largely identified as white. Web-based consent was supported by 93% of P/FM and 88% of I/R. Most respondents believed that return of genetic results would motivate study enrollment, but also indicated a desire to opt out. Ideal study design preferences included a 1-hour visit per year, along with the ability to complete study aspects by telephone or web and possibility of prophylactic medication. This study supports partnership of patients and clinical researchers to inform research priorities and study design to attain the promise of precision medicine for DCM.

Study Highlights

WHAT IS THE CURRENT KNOWLEDGE ON THE TOPIC?

✓ Involving patients in research study designs may enhance study relevance and improve the value of health research. No studies, however, clarify attitudes of patients with dilated cardiomyopathy, family members, and doctors toward genetic study design.

WHAT QUESTION DID THIS STUDY ADDRESS?

✓ This study aggregated patient and researcher attitudes toward direct-to-participant enrollment, receipt of expanded genetic results, and study design elements to facilitate study design with a patient-centric focus.

WHAT DOES THIS STUDY ADD TO OUR KNOWLEDGE?

✓ The results suggest that patient feedback may help attract and retain enrollees as well as benefit patient

outcomes. Web-based consent was viewed favorably, which coincides with increased implementation of direct-to-consumer technologies in research. Return of complete genetic testing results could be motivating to patient enrollment as long as only actionable results are delivered by an engaged, knowledgeable health-care team.

HOW MIGHT THIS CHANGE CLINICAL PHARMACOLOGY OR TRANSLATIONAL SCIENCE?

✓ Findings from this study may help researchers and clinicians more fully realize the potential of precision medicine and inform the future design of large genetics research studies.

Enrollment and continued engagement of thousands of participants in genetics studies are ideal for precision medicine research. Large, family-based cohorts that include racially diverse and potentially environmentally exposed individuals may be necessary to comprehend complex genetic architecture. To ensure study success and ultimately improve the value of health research, there is a need for recruitment and enrollment motivation strategies.¹ Researchers must then maintain enrollee engagement throughout the study.

We designed a qualitative pilot study to understand study participant preferences and inform design of large genetics research studies.

We surveyed enrolled patients, their family members, and investigators of the ongoing Dilated Cardiomyopathy (DCM) Precision Medicine Study funded by the National Institutes of Health, a multisite consortium-based study that is determining whether idiopathic DCM has a genetic basis.² Capitalizing on the unique opportunity afforded by a conjoint

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patient and investigator meeting, we anonymously queried participants about genetic study enrollment motivations, attitudes toward direct-to-participant consent processes, desirability of return of genetic results, and other aspects of study design. Our goal was to collect this information to facilitate design of future studies with a patient-centric focus.

Due to widespread adoption of direct-to-consumer genome sequencing, electronic results, and data platforms, and mobile health applications (“apps”),³ we aimed to determine desirability of direct-to-participant recruitment and consent, which places patients and research participants at the center of decision making and has been shown to facilitate study enrollment and retention.⁴ Return of electronic, complete genetic testing results were considered another key incentive to maintain study interest.^{5,6} However, no studies about patient attitudes toward DCM genetic studies have been performed, especially in the context of direct-to-participant enrollment and receipt of expanded genetic results.⁷ Furthermore, we wanted to understand patient preferences for length, format, and frequency of study visits, as well as acceptability of proactive treatment. We therefore conducted a pilot study to address these issues and to benefit researchers considering design of genetics studies.

METHODS

Parent study

This is a pilot study conducted for The DCM Precision Medicine Study, which aims to test the hypothesis that most DCMs of unknown cause, or idiopathic dilated cardiomyopathy, has mostly a genetic basis.² The study aims to determine if DCM, whether sporadic or familial, has mostly a genetic basis by using cardiovascular clinical and genetic information from 1,300 patients with DCM and their first-degree family members.^{2,8} Additionally, an intervention to help probands discuss genetics with their family is being tested in a randomized trial to determine if it will motivate family member clinical screening. The University of Pennsylvania single institutional review board approved the study, acting as a central institutional review board for all member sites of the consortium. All study participants provided informed consent.

Eligibility and recruitment

Pilot study participants included enrolled patients of the Precision Medicine Study and their first-degree family members (patients (P)/family members (FM)) as well as Precision Medicine Study investigators and other researchers (I/R) consisting of cardiologists, data scientists, clinical genetic counselors, and research administrators. The study recruited all attendees of the Spring Scientific Symposium of the DCM Consortium held at The Ohio State University April 25–26, 2019, a conjoint meeting of the DCM Research Project Consortium members (<https://dcmpproject.com/>), and the DCM Foundation (<https://dcmfoundation.org/>), which aims to provide hope and support to those with DCM. The purpose of the meeting was to discuss the status of the Precision Medicine Study and to plan future studies. Conjoint sessions were intended to integrate insights and feedback of DCM Ps/FMs into the study planning process.

Survey development and administration

Structured surveys were developed to elicit attitudes and preferences toward study design, direct-to-participant recruitment and web-based consent, return of genetic results, communication of genetic risk, and medical interventions. For example, survey respondents were asked to consider if potential study participants would be able to successfully answer study eligibility questions by the web, the optimal time required for a web-based consent process, and the medium for providing support during consent, such as chat, telephone, or email with study personnel. Return of results questions assessed desirability of complete genomic information receipt as well as understanding of results and sharing preferences. Study design questions consisted of medication use and study visit procedures.

Surveys were developed before the Symposium by brainstorming. The brainstorming group, a cardiologist, two genetic counselors, a statistician/geneticist, and a molecular biologist, was asked to record independently as many survey items as possible in 10 minutes. Responses were shared aloud to refine questions. Similar ideas were collated around select themes. Final questions were reviewed by all group members for clarity and consistency with study intent. The 20-question surveys consisted of multiple choice, Likert scale (not likely, somewhat unlikely, neutral, somewhat likely, and likely), and Yes/No questions as well as free text short answer questions. I/R and P/FM questionnaires were identically structured, but the I/R version queried them regarding their patients’ beliefs. For example, researchers were asked how much time they think a participant would be willing to spend completing a web-based consent form.

Surveys were self-administered anonymously immediately after the conclusion of the Symposium. All attendees were invited to participate in the surveys; not all attendees took the survey, or answered all questions, thus the number of responses differed slightly for each question.

Data analyses

Due to the descriptive nature of this pilot study and small sample sizes, responses from P/FM and I/R were examined separately and not compared statistically. Responses from patients and their family members were grouped due to small sample sizes. Percentages of “Likely” or “Very likely” Likert scale questions were tabulated. Short answer written responses were examined for themes using a qualitative descriptive approach, and similar themes were grouped. Noted differences among responses also were reported. Data are available upon request from the corresponding author.

RESULTS

Symposium attendees and respondents

Demographic characteristics of the attendees are presented in **Table 1**. Seven distinct families were represented by the P/FM attendees. Attendees were nearly even split by sex, but the majority (76%) identified as white or Caucasian. Sixteen P/FM (53% of those attending) and 18 I/R (33%) completed the surveys.

Table 1 Symposium attendee demographics

Characteristic	I/R	P/FM	Total
Age range, years	18–66	25–72	—
Male, %	29 (52.7)	14 (46.7)	43 (50.6)
Race, n, %			
White	35 (63.6)	30 (100.0)	65 (76.5)
African American	5 (9.1)	0	5 (5.9)
Hispanic or Latino	4 (7.3)	0	4 (4.7)
Asian	11 (20.0)	0	11 (12.9)
Total	55 (100.0)	30 (100.0)	85 (100.0)

I/R, investigators/researchers; P/FM, patients/family members.

Survey multiple choice responses

Most P/FM (93%) responded favorably to web-based recruitment and consent (**Table 2**) and were likely to return to the consent form to complete it after exiting (88%). Further, most (87%) believed they could assess their own eligibility for enrollment. The majority (81%) of respondents thought web-based consent form completion should require 30 minutes or less. Live chat, telephone call, or email assistance from study staff were similarly preferable.

The return of genetic results was seen as a motivating factor for research study participation (60%), but participants admittedly would not necessarily understand results without counseling (56% unable to understand). Participants would share results with their physician regardless of whether these results were negative or positive. In terms of study design, 69% of P/FM were willing to take a disease-preventative drug and 93% would be willing to return for ongoing study visits. P/FM preferred these visits to be 1 hour or less in length (59%) and occur yearly or less (88%), and would complete parts of the visit by telephone (40%) or by web (60%).

Like P/FM, most (88%) I/R believed that P/FM would respond favorably to web-based recruitment and consent (**Table 2**), but I/R were not convinced that participants would return to a web consent form after exiting (44% favorable responses) or that participants would answer eligibility questions accurately (56% favorable responses). The majority (89%) of I/R thought participants would be willing to spend 30 minutes or less completing a web-based consent form, and that assistance via phone or live chat would be preferred (76%).

I/R viewed receipt of complete genomic information motivating for study participation (74%), but doubted that participants could understand these results without counseling by medical professionals. I/R also believed that participants would share positive (94%), but not necessarily negative (28%), results with physicians. Researcher responses varied regarding the possibility of participants taking medicine to prevent a disease; however, I/R believed family members would take medication once diagnosed with early stages of disease, or if a family member was diagnosed with DCM or experienced a serious DCM-related outcome. Researchers were generally in agreement with P/FM in terms of study visit format (79% 1 hour or less long, 56% yearly or less, 65% by tele phone, and 78% by web).

Survey free text, short answer responses

P/FM short answer questions and responses are highlighted (**Table 3**). Representative themes in the responses were grouped and presented together, whereas unique viewpoints are also highlighted.

P/FM multiple choice responses indicated favorable views of complete genomic information receipt (60%), but written responses revealed misgivings. One participant did not want to know about other disease risks because she “has plenty to already think about,” especially if she was unable to reduce her risk. Another participant expressed concerns about data privacy. Actionable and accurate results were preferable. In terms of how they receive genetic testing information, patients welcomed more communication, especially via telephone. Further follow-up after the return of results by telephone call was desirable. One researcher remarked, “I have seen some genetic blame being placed—your side, my side, etc.”

Researchers and participants agreed that motivation to enroll in genetics studies was due to concern for family members and altruism. One researcher commented: “Even though we all want to be altruistic, in reality we want to protect our kids. I think that’s really a big driver.” Conversely, one participant expressed frustration with participation in a study they would ultimately not directly benefit from (**Table 3**).

The most important aspect of treatment was interaction with and quality information from healthcare personnel. Likewise, the most beneficial aspect of participating in the DCM Precision Medicine Study was information, and P/FM expressed a need for DCM research information and communication. I/R agreed the DCM Precision Medicine Study had provided patient benefit, primarily through gained actionable knowledge of the familial nature of DCM as well as provision of hope, peace of mind, and engagement with the disease. I/R were admittedly unsure about what participants would want others to know about their disease. A few said participants would want others to know that it is genetic. One stated, “even though they look ok, it is a real disease,” similar to a patient response. No I/R respondents replied that the most important aspect of participant treatment was interaction with physicians. In fact, P/FM seek this interaction and as much disease-specific information as available.

For future studies, P/FM again wanted information and for their doctors to better understand genetic disease. I/R believed P/FM wanted to receive genetic testing results faster as well as information about advances in the field and new research findings, while dealing with less study-related paperwork, time commitment, and procedures.

The greatest impediment to study participation cited by both I/R and P/FM was logistics, including time and money spent, travel to study site, etc. Lack of symptoms also was a common response from P/FM. I/R responses included lack of motivation by unaffected family members, desire to receive a nongenetic cause, and lack of perceived direct benefit.

Either healthcare providers (cardiologist or genetic counselors) or family members communicated disease information to P/FM. Most of these individuals communicated the information in person, and the information was

Table 2 Multiple choice survey questions and responses

Patient/family member facing question	Patient/family member responses (n = 16)	Researcher responses (n = 18)
<i>Web-based consent themed questions</i>		
1. You are considering enrolling in a research study. If given the option of a web-based study consent form to be performed at your convenience as an alternative to scheduling an in-person, face-to-face consent appointment, how likely are you to opt to consent through a web-based form? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	93% Likely or somewhat likely	88% Likely or somewhat likely
2. Suppose that you are unable to complete the web-based consent form at once. How likely are you to return to the online form to complete after saving your progress and exiting? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	88% Likely or somewhat likely	44% Likely or somewhat likely
3. How confident are you in your ability to accurately answer questions about your eligibility for the DCM Research Project by answering a series of questions about your cardiovascular health history? Likert Scale Responses: 1—Not Confident, 2—Somewhat Non Confident, 3—Neutral, 4—Somewhat Confident, 5—Confident	87% Confident or somewhat confident	56% Confident or somewhat confident
4. How much time would you be willing to spend completing a web-based consent form? Less than 10 minutes, 10–20 minutes, 20–30 minutes, 30–60 minutes, over an hour	81% Preferred 30 minutes or less	89% Responded 30 minutes or less
5. If you would like to ask a member of the study staff a question about content of the web-based consent form, which type of communication would you prefer in order to reach a member of the study staff to address your question: Live Chat, Telephone Call, Email, Other [Specify] (U.S. mail, Online, Text)	29% Live chat, 35% telephone call, 29% email	36% Live chat, 40% telephone call, 22% email
<i>Return of genetic results themed questions</i>		
6. Would you be more willing to participate in a research study if you could receive your complete genomic information? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	60% Likely or somewhat likely	74% Likely or somewhat likely
7. Do you think you would be able to understand your results by yourself without further counseling? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	56% Unlikely or somewhat unlikely	89% Unlikely or somewhat unlikely
8. Do you think you would be able to understand your results without further counseling by a Clinical Genetic Counselor? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	56% Unlikely or somewhat unlikely	83% Unlikely or somewhat unlikely
9. Do you think you would be able to understand your results without further counseling by an MD? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	31% Unlikely or somewhat unlikely	33% Unlikely or somewhat unlikely
10. Would you discuss your positive results with your physician/s? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	88% Likely or somewhat likely	94% Likely or somewhat likely
11. Would you discuss your negative results with your physician/s? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	94% Likely or somewhat likely	28% Likely or somewhat likely
<i>Study design themed questions</i>		
12. How likely are you to take a medication to prevent a disease you don't yet have? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	69% Likely or somewhat likely	53% Likely or somewhat likely
13. Do you think that family members would rather have early signs of a disease (e.g., DCM) rather than some very early findings that will lead to DCM (but that do not meet a "disease" classification) to start a medication? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	56% Likely or somewhat likely	100% Likely or somewhat likely
14. Do you think that it is only after a family member has DCM that the family will be proactive in this regard? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	87% Likely or somewhat likely	94% Likely or somewhat likely
15. Do you think that it is only after a family member has DCM and some bad outcome (e.g., sudden death, advanced heart failure, heart transplant, death), that they would seriously consider taking a medication with very early findings? Likert Scale Responses: 1—Unlikely, 2—Somewhat Unlikely, 3—Neutral, 4—Somewhat Likely, 5—Likely	71% Likely or somewhat likely	84% Likely or somewhat likely
16. Would you be willing to return for ongoing visits for study-related testing (e.g., possibly to include heart checks, medical testing, surveys, and other data collection)? Yes/No	93% Yes	94% Yes

(Continues)

Table 2 (Continued)

Patient/family member facing question	Patient/family member responses (n = 16)	Researcher responses (n = 18)
17. What would you consider to be a reasonable length for such study-related visits? ^a One half hour, one hour, two hours, three hours, four or more hours	59% Responded 1 hour or less	79% Responded 1 hour or less
18. What would you consider to be the optimal frequency of such study-related visits (yearly, biennial, etc.)? ^a Every 6 months, every year, every 2 years, between 2 and 5 years, every 5 years	88% Responded every year or less	56% Responded every year or less
19. Would you be willing to complete parts of the study visit by telephone or web? ^a No; Yes, by Phone; Yes, by Web	40% by Phone, 60% by Web	65% by Phone, 78% by Web
20. Would you be comfortable with longer study visits if the visits were less frequent? Yes/No	87% Yes	53% Yes

DCM, dilated cardiomyopathy; MD, medical doctor.

^aRespondents could provide more than one answer to this question.

most often shared immediately after receiving a diagnosis. At least one proband acknowledged difficulty with explaining the ambiguity of DCM to family members and getting them to participate. Another factor affecting communication was fear and anxiety surrounding the uncertain potential for disease.

Providers could best support P/FM by understanding the genetic basis of diseases. In terms of their own needs, providers want more information in the form of point of care resources to help with genetic information, support for clinical care post diagnosis, lay summaries of evolving research, and best therapies and preventative measures. They also want greater access to genetic counselors.

Similar to the multiple-choice responses for this question, P/FM responded positively to a web-based consent process, but wanted the capability to contact a representative with questions. I/R questioned patient self-assessment accuracy. All expressed concern about generational adoption of the technology. One researcher asked, “What about the absence of a conversation? It’s very cold, right?”

I/R correctly predicted that P/FM want to understand complete risks, benefits, and side effects before agreeing to start a drug to reduce DCM risk. P/FM nearly unanimously would be willing to try non-drug interventions, such as exercise, diet, and enhanced screening, but I/R predicted low adherence to these lifestyle modifications. One asymptomatic FM had already been prescribed a low dose ACE inhibitor. Another FM of childbearing age was not sure she would take a preventive drug. One researcher commented, “There is a cost to everything we do. Taking a pill every day is a psychological reminder to someone that they could have a major heart problem; may cause psychological harm to someone. Evidence of preclinical disease is still disease.” Another said, “The thing is, we haven’t proven this concept. It’s not evidence-based is the problem. We are trying to create that evidence base. There is nothing benign about any medication we give.”

DISCUSSION

A scientific symposium comprised of participants from the DCM Precision Medicine Study provided an ideal opportunity for pilot data collection regarding attitudes toward aspects of large DCM genomic research studies. This project is unique in that both patients and researchers were

queried to construct a thematic analysis intended to inform future, patient-centered genetics studies. Our study suggested that patient feedback may inform a study design that can attract and retain enrollees as well as benefit patient outcomes. Consideration of enrollment and retention strategies for large studies is crucial. Our findings are consistent with recent studies that engaged research participants in the design or conduct of research to enhance relevance to patients.^{9–14}

We used a conjoint meeting to elicit study design details, where topics relevant to all participants of the DCM Precision Medicine Study were explored in a shared setting. Prior studies also have demonstrated impact of and rationale for patient and researcher involvement in research meetings.^{15,16} In our particular study, collected survey responses from our meeting were examined for three key themes regarding genetic research design that constituted the major outcome measures of our analysis.

Web-based consent

Given the popularity of web-based, direct-to-consumer genetic testing,¹⁷ we believed that providing potential study participants with an option to consent at their convenience via the web would motivate study participation. Web-based consent is increasingly accepted in all types of studies.¹⁸ Our data supported this finding, although we clarified that provision of technical support and awareness of generational preferences was desirable. Web-based consent for genetics studies could be successful if: (i) the survey ideally requires 30 minutes or less to complete; (ii) technical support is readily available; and (iii) the web interface can accommodate varying skillsets.

Return of results

Receipt of complete genetic results was not universally desired or a motivating factor for enrollment. Instead of being viewed as a benefit of study enrollment and consistent with previous studies, P/FM expressed anxiety over receipt of genetic results and the potential for revealing new genetic risks unrelated to their primary diagnosis.¹⁹ Considerations such as limitations of direct-to-consumer genetic testing, accuracy of results, potential for loss of patient confidentiality, etc. increased the anxiety.

Conversely, when P/FM receive results pertinent to their diagnosis, they prefer as much information as possible to

Table 3 Free text short answer survey questions and responses

Question	Selected responses from patients and their family members
Q1: How do you think you would feel about receiving more information (than just DCM related genetic information) when genetic testing results are returned to you?	<p>"I want a doctor to share all information with me, even if it is messy." "The way I work is the more information the better, but I noticed in my session that not everyone's like that. There's a lot of fear out there. Not everyone wants to know everything I want to know." "I want to be able to pick what information I want to receive." "I would like if there were an option to only find out about defined, treatable diseases." "I have done ancestry.com and that was fine, but I have enough on my plate with the diagnosis. It wouldn't be motivating to me." "It seems like it's fine to provide it, but I don't know that it would necessarily motivate me more to join the study. I'm feeling motivated for my husband who has DCM and my children already." "I love 23andMe and so do my friends. What's cool about the Corel [sic] study is that it's been seven years and I'm still getting genetic results. As the research continues to develop, they will continue providing information. The opportunity is available on the website to see or not see my result."</p>
Q2: What motivated you/would motivate you to enroll in a study returning genetic information?	<p>"My family is affected. We don't know what's going on genetically, but we're happy to contribute to research and get answers." "I have a hard time agreeing to do the study—I already have the disease and don't have children. What are the other benefits? Why should I participate? I so appreciate what you guys do, but it didn't change anything. My treatment is still the same, my condition is still the same; it didn't change my daily life."</p>
Q3: What aspects of your treatment have been most important to you? Has the study been beneficial to you? Why or why not? What do you want other people to know about your disease or situation?	<p>The most important aspect of treatment was interaction with and quality information from healthcare personnel. The most beneficial aspect of participating in the DCM Precision Medicine Study was information.</p>
Q4: What would you like to see more of/less of in future genetic studies?	<p>"Dissemination of information. Doctors have all this knowledge and we aren't getting that." "General cardiologists need to be on board with this. Honestly, my husband was diagnosed with a heart condition at age 50. I never heard about a genetic connection until a few years ago, when we had dinner with (a cardiologist involved with the study). She asked if he had been tested and the kids could be tested. And so, no cardiologist has ever mentioned it. They are the ones who could say, it would be really interesting for you to go and get involved in this study."</p>
Q5: What do you think is the greatest impediment to family member participation in a DCM genetic study?	<p>"Not everyone wants to know their genetic information, especially when they have no symptoms." "Sometimes the physicians are reluctant to screen a family member if they have no symptoms." "My brother has six kids and a defibrillator. It's been a lot of years since my son was transplanted, so a lot of years since anyone was acute. The younger kids, my brother's kids, don't see the need to get further screening. I think they think we're healthy, what's the big deal. They don't see the imminent risk." "A confusing process discourages people." "It didn't do a thing for me. I don't have any change in my care, daily routine."</p>
Q6: What was your/your family's experience with communicating risk? Who communicated it/relationship to you? How was it shared (telephone, email, etc.)? What was stated? When was it shared (immediately after finding out; after letter received; or much later...after personal processing of info or after another event occurred in family)?	<p>"I was the driver. I was able to get nine people in my family tested. I convinced them via calling, face to face. Logistics of it is difficult, and that may be why 80% of the resistance is encountered—the echo, ECG, blood draw, expense." "Some family members may see affected relatives as anxious or worried. Why would other family members want to go through that voluntarily?" "There is a fear of insurance being adversely affected if a pre-existing condition is identified." "We just said, we don't want to scare you because we don't know because my husband hadn't been tested yet. They said, we wouldn't have wanted to know because what would we do?"</p>
Q7: What do providers need to conduct and support participants and families with genetic DCM?	<p>"My PCP had told me there was no need to be tested for DCM, she was completely unaware of the genetic link." "General cardiologists need more information to direct their patients." "Providers need to understand the genetic basis of disease."</p>
Q8: You are considering enrolling in a research study. If given the option of a web-based study consent form to be performed at your convenience as an alternative to scheduling an in-person, face-to-face consent appointment, how likely are you to opt to consent through a web-based form?	<p>"It could be helpful for reluctant or out-of-town family members." "I would prefer it, even if it was long. I would rather do it that way than face-to-face." "I would prefer it. The medical community is still stuck in the 1970s." "One of the difficulties (of in person consent) is that it is going to take me an hour of my time. Whereas if I was on a web-based thing I could do it for five minutes and then come back to it. It is going to be a hassle either way but if you can reduce the time and hassle—you're still going to have to look at some blood tests—but still for me one of the drawbacks is to drive somewhere and spend an hour doing all this stuff." "I think one of the advantages of this approach is to enroll patients from sites distant from any coordinating site. ... and distant from any genetic counselor so where are they going to go have their blood drawn? There may not be any genetic counselor or someone associated with the study to complete the process." "I would like that a lot. I just retired and I did every form possible online because I didn't have time to talk to individual people or to wait on the phone. I know my kids that are in their late 30s and early 40s have even less time than I have to spend on the phone. They don't even like talking on the phone. They are very literate with the web." "Level of comfort with technology goes down after about age 55 in the current population. Depends on ease of use and platform." "I'm okay with it, but if I get stuck, I want to call somebody on the phone. I don't want to have this chat. For me, as an older person, I'm okay starting on the web, but I want a phone number so I can talk to somebody."</p>

(Continues)

Table 3 (Continued)

Question	Selected responses from patients and their family members
Q9: What information would you want to have before starting on a drug to reduce DCM risk? What other types of non-drug interventions would you be willing to do (exercise, diet, enhanced screening, etc.)?	"A positive genetic test doesn't necessarily mean disease." "I would say no. My older son does take preventative medication, and last year (his doctor) didn't know that. And when (his doctor) saw him, he just said to keep doing what he's doing. The reason his cardiologist even gave it to him is because he told him about his family history. He also did have a slightly decreased ejection fraction." "I don't have the bad genes, but my brother and his son have ventricular arrhythmias and they both have defibrillators. So if you found those findings on me, even though I don't have the bad genes, I would probably still take the preventative medication." "If the risks of taking the medicine were clearly communicated, I would be receptive."

DCM, dilated cardiomyopathy; ECG, echocardiogram; PCP, primary care provider.

include the latest disease research and detailed follow-up with genetic counselors and physicians. Patients preferred to receive life-threatening results through a personal meeting or telephone call,²⁰ perhaps because the professional could explain the findings and assist the patient with coping. Future studies might consider the use of web-based result return only when returning routine results without health implications.²¹ Options for receipt of complete or only selected genetic testing results may be preferable, as is consistent with previous research.^{19,22-25} Indeed, the American College of Medical Genetics' current guidelines recommend the option to opt out of receipt of complete genome analysis.²⁶ Consistent with our study, the most important patient-reported aspects of results return included receipt of no cost, actionable information, especially for serious diseases for which treatments exist,^{22,25,27-29} and availability of detailed written information to explain the results.¹⁹

Study design preferences

P/FM and I/R shared similar views regarding study participation motivation, prophylactic drug use, and other aspects of study design. Motivation for enrollment in our family-based genetic studies was largely due to concern for children and other family members, as is consistent with participation in medical research studies in general.^{22-24,29,30} Some participants also mentioned altruism as a motivator.^{23,28,30,31} Researchers should take note of the emphasis on concern for family and use that information to increase study enrollment, motivation, and participation. Other motivators for study participation include some form of personal gain and improving knowledge and medical advancement.^{28,32}

P/FM noted that increased physician awareness of the genetic basis of diseases would improve P/FM support and conduct of future studies. Importantly, I/R often correctly predicted and agreed with the responses of P/FM, showing in particular that physicians recognize patient need for information as well as their desire to provide that information. I/R recognized their limitations with regard to understanding the genetic basis of diseases and expressed a need for more genetic counselor support. These results are consistent with patient perception that clinician genetic knowledge is poor and their desire for providers to be knowledgeable and work closely with genetics experts to provide the most informed care.²⁹ This study showed that I/R did not fully understand the importance of their own interactions with patients. Providers should place importance on their ability to provide as much disease-specific genetic knowledge to their patients as possible.

There are several limitations of this study that could be addressed in future research. First, sample sizes were small, as this project was intended to be a qualitative pilot study. Second, the race/ethnicity of our P/FM sample was homogeneous. A more diverse population is recommended for true generalizability of these results. In addition, because survey data were collected at the conclusion of the meeting, it is possible that presentations and personal interactions may have influenced P/FM and I/R opinions on these thematic areas. Finally, the attitudes of enrolled participants may differ from those not enrolled. As a result, the opinions may not be directly generalizable to general DCM study participant populations.

CONCLUSIONS

Three major themes were investigated in this study. Web-based consent was viewed favorably, which coincides with increased implementation of direct-to-consumer technologies in research. Return of complete genetic testing results could be motivating to patient enrollment as long as only actionable results are delivered by an engaged, knowledgeable healthcare team. Patient and provider partnership when designing studies may help researchers and clinicians more fully realize the potential of precision medicine.

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