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Patient experiences across a genetic screening and testing program pathway

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ABSTRACT

Background: Population-based genetic screening and testing programs have substantial potential to improve cancer-related outcomes through early detection and cancer prevention. Yet, genetic testing for cancer risk remains largely underutilized. This study aimed to describe barriers and facilitators to patient engagement at each stage of a California-based genetic screening program, from completing the electronic screener to receiving the test, and to identify potential improvements that could support precision medicine-based approaches to patient care.

Methods: We conducted 26 semi-structured interviews among program participants who did not complete the screener (n=9), those who did not receive the recommended test (n=7), and those who received a genetic test (n=10). Interviewees were selected from patients who recently received a mammogram through one of the participating Southern California clinics. Interviews were transcribed and coded using Atlas.ti. The study used a qualitative descriptive approach to identify similar and contrasting themes among the participant groups.

Results: This study found that barriers and facilitators to engagement were largely the same regardless of how far participants had moved through the process toward getting a genetic test. We identified four overarching themes: participants wanted clear communication of personal benefits at each stage; participants needed additional information and knowledge to navigate genetic screening and testing; a trusted provider could be instrumental in participants following a recommendation; and repetition and timing strongly impacted participants' likelihood to engage.

Conclusions: Providing education around the benefits of genetic screening and testing to patients and their families, as well as clear communication about what each step entails may help patients engage with similar programs. Strategies aimed at increasing coordination among a patient's healthcare team can also help ensure information reaches patients in multiple ways, from multiple providers, to increase the likelihood that recommendations for testing come from trusted sources, which supports the uptake of genetic testing.

INTRODUCTION

A growing number of genetic screening programs have been implemented in multiple settings (e.g., primary care, specialty ambulatory care like cardiology, etc.) to identify high-risk patients in need of additional preventive treatment and connect them to needed follow up care [1,2]. Programs that offer population genetic screening to assess cancer risk are increasingly viewed as a key means for supporting population health overall because of their potential to identify patients in need of enhanced monitoring or who could benefit from prophylactic treatments to prevent or mitigate symptoms [3,4]. However, genetic screening and testing for cancer risk remains largely underutilized among patients [5,6], raising questions about patient perceptions of these programs, including benefits and barriers, to understand how to encourage participation.

Leveraging genetic testing to assess cancer risk has substantial potential to improve cancer-related outcomes through early detection and cancer prevention [7–9]. In addition to improving health outcomes, studies demonstrate that these programs are cost-effective and can potentially result in cost-savings for healthcare systems compared to using family history alone to assess cancer risk [10]. Yet, for these programs to be effective, patients must be actively engaged through a multi-step process. This includes the completion of a personal and family history screener to qualify for testing, the completion of a genetic test if they meet the recommended risk threshold, and connection to appropriate cancer screening and prevention care if patients are identified to have elevated cancer risk.

Despite population-based cancer risk genetic testing programs requiring multiple steps, previous studies tend to assess general perceptions of genetic testing from individuals who were not being offered a genetic test [11–13] or post-test reactions of participants who completed the genetic testing [1,14–16]. Few studies have focused on learning about the barriers and facilitators to engagement from patients who drop out of the program at each stage – including those who do not complete the risk assessment and those who do not receive a recommended genetic test. For these programs to be ultimately successful it is essential to understand the factors that shape patients' uptake and engagement in genetic testing at each stage of the process.

As genetic screening programs become more common, it is important to understand patient experiences of these programs, and what may motivate patients to complete the full genetic screening and testing process. This qualitative study used interviews with patients who had engaged to various degrees with a cancer screening program in a clinical setting to identify barriers and facilitators to patient engagement at each stage in the process, patient perspectives on the program, and what could be improved to support precision medicine-based approaches to patient care.

MATERIALS & METHODS

Setting

In 2020, the [*PROGRAM*] team in Southern California launched the Comprehensive Assessment, Risk, Education (CARE Program™) developed by Ambry Genetics, a population-based approach designed to increase cancer-related genetic testing for preventive purposes [17]. The program uses a text or email-based electronic screener to gather information about personal and family cancer history from patients who are due to receive a mammogram through one of six participating clinics. Patients who meet the National Comprehensive Cancer Network (NCCN) criteria of "high risk" [18] are recommended to receive a multi-gene "panel" genetic testing for alterations in more than 80 genes associated with hereditary cancer risks across a broad spectrum of tumor sites in addition to breast (uterine, ovarian, colorectal, pancreatic, etc.). Genetic testing is offered the same day following the patient's mammogram visit. Patients who undergo genetic testing and test positive for a pathogenic variant in any of the genes are offered genetic counseling and receive detailed risk reduction, prevention, and management recommendations. Program data on which patients completed the screener, which received the testing recommendation but did not receive genetic testing, and which completed the full process are collected as part of routine care and program oversight.

At the time of the study, around 150,000 electronic screeners had been sent to patients, with an approximately 70% completion rate. A little less than 30% of these patients met NCCN criteria and received a testing recommendation. Of these, about 15% of patients eligible to receive testing go on to

complete genetic testing. The institutional review board of [*ORGANIZATION #1*] approved the study (#2023000128).

Participant Selection

In May through June 2023, we conducted qualitative interviews with patients who had recently received or were due to receive a mammogram through one of the participating clinics. We used program data to identify potential interviewees and to classify them according to where they were along the workflow of receiving the electronic screener, completing the information, receiving the recommendation that they meet guidelines for hereditary risk assessment, and receiving the genetic test. Of note, these classifications were based on the patient's point along the workflow for their current appointment rather than any past appointments in which they may have been exposed to the electronic screener. As described in more detail in the limitations, a greater proportion of participants had previously received genetic testing than anticipated during recruitment. Email invitations were sent to all patients who met the study criteria; outreach materials and patient interviews were conducted in English. Participants reviewed consent forms with the interviewer and gave verbal consent separately to the interview procedure and to electronic health records (EHR) release.

Public and Patient Involvement

Patients or the public were not involved in the design, conduct, analysis, or dissemination of the research study. However, patients were central to the study in determining the relevant barriers and facilitators to genetic testing. In addition, based on the results of patient interviews and what we learned about patient misconceptions and areas of interest, we provided feedback to program staff, who updated a Frequently Asked Questions document that was distributed to all participants in the study.

Data Collection

Three semi-structured interview guides were developed for the groups: (1) those who had received the screener but had declined to fill out the information or had not yet completed it, (2) those who met NCCN criteria for high risk but declined testing or had not yet decided to receive testing, and (3) those who received a positive genetic test result (i.e., pathogenic variant(s) detected). Participants were asked about their views on genetic testing, what could or did motivate them to receive a genetic test (including their history of prior testing), and any barriers or facilitators they encountered during the process. Participant experiences with genetic testing did not always align with data captured by the program data; interview questions were adjusted based on this information.

We aimed to contact patients in the first two groups who had completed their mammograms within a month of study outreach. For patients in the tested-positive group, we conducted outreach with those who had tested positive at least five months prior. Trained members of the research team (SR, a female PhD research scientist; CD, a female Master's-level research associate) conducted interviews which lasted between 30 and 60 minutes and were conducted either virtually or via the phone, based on participant preference. Interviews were recorded and transcribed verbatim by a transcription service and reviewed for accuracy by the research team.

Data Analysis

Interview data were analyzed using a qualitative descriptive approach, which stays closer to the data than other qualitative approaches, provides in-depth content from the participants' perspective, and is well-suited to health care research [19]. Initial codebooks were developed from the interview guide and revised iteratively when emergent codes were identified. To help maintain methodological rigor, three of the interviews were double coded. After each double-coded interview, the coding team met to discuss codes until consensus was reach. The team reviewed the content of the codes to develop a set of central themes. Illustrative quotes are presented in the results section to support the themes and recommendations. Additional verbatim quotes are provided in Table 1 to further validate findings. All analyses were completed in Atlas.ti version 23. Additionally, information from electronic health record data was pulled to demographically describe the sample.

RESULTS

A total of 26 interviews were conducted. Participants were primarily White and English as a preferred language. Table 1.

Table 1. Participant Demographics

Characteristic	Number or Percent
Participant Categorization	26
Did not complete screener	9
Did not receive genetic test	7
Tested positive	10
Age in years (mean)	59.2
Gender	
Women	88.5%
Preferred Language	
English	88.5%
Race/Ethnicity	
White	80.8%
Asian	7.7%
Hispanic/Latina	3.8%
Insurance Type	
Commercial	61.5%
Medicare	23.1%
Unknown/Uninsured	3.8%

Data do not sum to 100% due to missingness. Two participants did not consent to have their interview data linked to their electronic health record data.

Four themes emerged from interviews with all participants, regardless of how far they had moved through the process toward completing a genetic test. These included: (1) the desire for clear communication of benefits, (2) the need for more information and knowledge, (3) the importance of a trusted provider, and (4) the advantages of repetition and timing. Additional exemplar quotes found in Table 2.

Communication of Benefits

At each stage of the CARE program pathway, from filling out the electronic screener to receiving the genetic test, participants were motivated to complete the tasks based on their understanding of the importance of the steps. Many of the participants did not understand the purpose of the screener when they received it; nearly all of participants who did not complete the screener mentioned that they would have been more likely to complete it or find the task important had they known that it would be used to determine if they should receive a genetic test or otherwise inform the type of healthcare they would receive. Participants did not differentiate the electronic screener from other pre-appointment paperwork and forms; to most, it appeared to be the same as many of the other family history questionnaires that they were asked to fill out for other medical visits. Participants often expressed frustration at completing a seemingly redundant form.

> "*Sometimes I think 'I just did this last week,' or 'Do you have to know this again?' That's the thing I can say about it that's a negative…It's not difficult [but] it takes time."*
> - Participant who did not complete the screener

In terms of receiving the genetic test, participants did not always realize that the recommendation was tied to their family history and an increased personal risk for certain cancers; participants suggested that understanding this link may have increased their motivation to receive the test. Many of the interviewees who engaged in the testing were motivated by being able to share their results with their family members. This benefit was not always clearly understood by interviewees who did not complete the screener or take the test; a better understanding of the connection may have increased engagement.

> *"I think that [knowing the connection between the screener and a genetic test recommendation] would change [my willingness to complete the screener] tremendously because it's like you're using your family history to figure out if it's a possibility that that relates to you."*
> - Participant who did not complete the screener

Information and Knowledge

A large proportion of interviewees had prior exposure to some kind of genetic testing through genetic testing associated with pregnancy, direct-to-consumer genetic testing, or genetic testing after a personal cancer diagnosis. Despite this previous exposure, some participants were still unsure what the recommended genetic test entailed, including whether it involved a blood test or biopsy or required a hospital stay. One person who chose not to get the genetic test expressed confusion between the genetic tumor testing the participant had already received when diagnosed with breast cancer, and the recommendation to undergo genetic testing to assess cancer risk. Participants who had not completed the screener gave similar responses. Some indicated that they would want more information about the specifics around the test, if it were recommended, including both general information about the test (e.g., how the test is done and how long it takes) and whether there were any medical risks (e.g., one participant who was concerned because they were "allergic to everything").

> "*I'm not even sure what entails genetic testing. Is it a blood test? Is it, you know, a biopsy kind of thing?"*

"I thought it was saying that if you have cancer, you know, if I'm already sick, would I be interested in it. And that was probably in my mind, too. Like 'No, I'm fine, I don't need this" …So I just didn't understand it."

- Participants who did not receive the recommended test

None of the participants (either those who had not received the screener or who did not receive the test) were fully opposed to the idea of genetic testing for cancer risk. Participants who were more hesitant about genetic testing wanted to know the personal benefit and expressed concerns about cost, but none of the interviewees had overriding objections to knowing about their genetic cancer predisposition. A few interviewees mentioned that they had encountered people (including family members they reached out to after receiving a test) who "*put their head in the sand…like an ostrich, they just don't want to know*" but most of the participants who received the testing recommendation already had a heightened awareness of their risk given their family and sometimes personal history with cancer.

Importance of a Trusted Provider

The provider making the recommendation also mattered to some participants. A number of participants expressed confusion based on their providers' level of emphasis on the testing recommendation, as well. Interviewees described the recommendation as a "suggestion" from their mammogram technicians that was not accompanied by a sense of urgency, which made it difficult for them to judge the importance of the recommendation for testing. One participant who eventually received genetic testing did so because when their primary care provider recommended the test, they emphasized how important it was for the participant to be tested given their family history. Interviewees felt that the recommendation from the CARE program could be communicated more strongly, and one interviewee suggested that receiving the recommendation from their doctor (as opposed to a technician or other staff) would make it appear more important.

"It wasn't like, "Oh, we think this would be good for you." […] versus, "If you wanna do it, you can, it doesn't matter." You know, it was just a matter of fact, "You can have it done if you want."

- Participant who received the genetic test

"I'm not sure that coming from a tech, it would carry a lot of weight…if [my oncologist] suggested it to me, I would probably do it."

- Participant who did not receive the genetic test

Many participants across the CARE program pathway described strong, trusting relationship with at least one provider – usually a primary care provider or oncologist – whose recommendations they were most likely to follow; none identified mammography staff as that trusted source of information. Participants that received a genetic test and tested positive for at least one genetic marker typically communicated a positive relationship with the genetic counselor. Participants were given the option to see the counselor before receiving the test, however, most opted to take the test first. Some participants who later tested positive wished that they had been directed more strongly toward the option of meeting the genetic counselor prior to testing, so that they could receive more information about possible life insurance effects or additional explanation around possible test results.

"I wasn't aware that I should have life insurance before I do genetic testing in case something shows up…which is a huge factor…I think part of it is I didn't ask questions and I wasn't really proactive in this care. Because I honestly never believed I had a genetic mutation."

- Patient who received the genetic test after a second recommendation from their primary care provider

Repetition and Timing

Interviews with participants who did and did not receive the genetic test suggested that, while there may not be immediate uptake by those who receive the recommendation, hearing about genetic testing multiple times and in multiple ways has a long-term effect of encouraging appropriate patients to receive testing. Many of the participants who were eventually successful at moving through the CARE program pathway (i.e., completed the screener and took the genetic test) had received genetic testing recommendations before. Their decision to receive the test at this point, rather than when it was suggested first, was based on a confluence of contextual factors, including changing family and personal cancer experiences and changes in the cost of the testing, etc. As mentioned previously, some participants received the test after getting the recommendation from multiple types of providers.

"I was just worn out...and I'd been through so much. I think it was at one of those points where I just felt like I just needed to like curl up and lick my wounds and recover and kind of hide for a while."

- Participant who did not receive genetic testing after initial recommendation

Similarly, participants who did not complete the screener described being busy with family and work obligations, but none expressed a complete refusal to share the information or fill out the forms. They described generally completing documentation of information like family history prior to a medical appointment but were unable to do so for their most recent mammography appointment.

"And then this year...I can't remember what was going on…I was like, I don't have time to do this, so I just didn't do it. I figured I'll do it next year."

- Participant who did not complete the screener

Table 2. Themes and exemplar quotations

Communication of benefits	*"You know, when I did the genetic testing, it was basically like, 'Do you wanna do it?' …There wasn't, you know, 'This is gonna be very helpful.' Of course, it's helpful, but the bottom line was, 'You can do genetic testing if you're interested.' So I was, because of all that history that I had on my dad's side, thinking, you know, that has to be it."* *"That website form it said something about genetic testing and have I had it? No. Would you be interested? Yes. And the reason why I wanted to do it was because my father, mother, brother, and sister have all had cancer. I have not, and three of those have passed away due to cancer. And my sister had breast cancer…So I was just interested in how their cancer…pertained to me."* *"So I went in for my first mammogram and I think you do a risk factor analysis that like gives you a number of your like lifetime risk of breast cancer, right?*

	And my number was really high. And so I remember I did the mammogram and then they said, 'Do you wanna do genetic testing?' And I said, 'No, I don't need to know.' And it was just the ultrasound tech, he was offering it and I really didn't have any understanding or-or like, I had no idea like the extent that it could provide information. *What ended up happening is I got a new younger lady as a PCP and I went to see her…and I was sharing my history and she said, 'You absolutely need genetic testing.' She's like, 'You can't get to the bottom of breast cancer. Like there's too many factors involved. Like you need to do genetic testing. I'm sending you to see a genetic counselor. Like you need to get this done.' And she was the first person who ever like flat out told me to my face that like, I need to get it done."*
Information and knowledge	*"I didn't know what it entailed. I think if I would have known more of what it entailed…there would be no fear or anything like that. [But the recommendation] is just in with all the other stuff…it was just real quick. I don't remember them talking about it in particular."* *"I probably wouldn't have asked had my doctor not recommended it. 'cause I just assumed…insurance wouldn't cover it, but, if they just made it easier to get as far as saying, 'Yes…your insurance will cover this, do you want it?' I think a lot more people would get it."* *"Well, I'd wanna know, what do they do? How do they do it? How long it takes."* *"I asked if it was gonna be, um, like a blood test or saliva 'cause I wasn't sure."*
Importance of a trusted provider	*"The only interaction I have is with the techs and ... if they threw it out there, it was very casual, but no one has ever sat down with me and said, 'Let's talk about doing some genetic counseling.' It's never been that formal. And it's never been with a doctor. It's only-- I've only met with techs... You know, I think it would mean more coming from a doctor."* *"Don't send me a letter telling me that you're recommending genetic testing. I mean, you know, that's just gonna freak me out. It's like, 'Okay, well what are you seeing? You know, why is this a concern all of a sudden? You know, why do you care?" Because my first go-to is to panic that, 'Oh my gosh, something's, you know, something's back and you saw something on the mammogram.' So I'd prefer not to be freaked out in the process."* *"I'm not sure that coming from a tech, it would carry a lot of weight. [I would want to hear from primary care nurse practitioner] if it's not gonna be my oncologist [which would be] a whole different ballgame."*
Repetition and timing	*"And then this year...I can't remember what was going on. I think my husband had a…surgery and my son, his [school] team was competing. I was like, I don't have time to do this, so I just didn't do it. I figured I'll do it next year. But yeah...it takes a really, really long time to do those forms."*

> *"I found that…a lot of the stuff I just didn't wanna know about, you know. I was just pretty focused on getting rid of the cancer and jumping through the hoops that the doctor wanted me to do. So all the other stuff, it's like, 'Okay, you just do your thing and-and I'm just gonna try not to die during chemo.'"*

DISCUSSION

Population-level genetic screening and testing programs can help identify patients at an increased risk of cancer and direct them toward enhanced cancer screening services (e.g., mammograms, colonoscopies, ultrasounds, magnetic resonance imaging etc.) or other recommendations. However, for these programs to be successful, patients must be effectively engaged at each step of the program pathway. Although study participants had varying degrees of engagement in the CARE program pathway, four themes emerged highlighting barriers and facilitators to participation that cut across the different groups: the benefits around clarity of communication, the need for increased information and knowledge, the importance of a trusted provider, and the effects of repetition and timing.

The themes we identified largely indicate that the reasons participants have for not engaging in testing are potentially mutable, and that patients are willing to engage in genetic screening and testing when they have a clear understanding of the specific purpose and procedures at each point in the pathway. Similar to prior research, our study found that patients are open to hearing about their genetic risks [11] but are most likely to support testing when there are clear medical benefits, and when testing is for specific diagnostic and predictive purposes [12,20,21]. These findings reiterate that a genetic risk assessment program, such as CARE, needs to clearly articulate to patients the diagnostic and predictive purposes of genetic screening and testing and how the testing would impact their future healthcare. Programs could consider opportunities to integrate patient communication and education during appointments, such as when mammogram patients are waiting to be seen.

Interestingly, while a limited number of individuals suggested that "not wanting to know" factored into their thoughts around receiving a genetic test, the fear of emotional and psychosocial effects from genetic testing results that have been cited in other literature [20,22,23] was not frequently identified by participants in this study. In fact, most participants felt that "knowledge was power" and having more information could help in their care. These findings may suggest a growing acceptance of genetic testing, with knowledge and provider relationships as the main factors affecting engagement.

Other barriers to genetic testing identified by patients in the study included effects on insurance and potential costs, which aligns with findings from previous studies [1,13,24]. Interviewees also expressed being frustrated by redundant requests for information that did not feel relevant to a given appointment or were not addressed by their healthcare provider during appointments. This highlights a need for improved portability and visibility of patient-provided history, particularly for patients who return year over year to the same center or see multiple providers within the same network. Moreover, patients at each stage in the pathway described their own limited capacity, in terms of time and attention, to prioritize, navigate, and complete each request and recommendation offered by the healthcare system. Communication from a trusted provider helped counteract these barriers and was identified as a key facilitator to moving forward in the process for many participants. Previous research

has also underscored the importance of hearing the recommendation for genetic testing from a trusted provider, especially among racially and ethnically diverse patients [25–27]. In addition, patient navigators have been shown to improve patient follow-through on genetic testing [28].

These findings highlight the importance of supporting the coordination of health care team around genetic testing and risk assessment. Improved coordination of the health care team may help ensure that multiple providers address the recommendation with patients at multiple time points and increase the feeling of importance around the test. Additionally, expanding the languages the screener is available in is an important way to support equitable access to testing, especially in linguistically diverse regions like Southern California.

Limitations

As mentioned before, self-selection bias was a limitation in this study; participants who were completely opposed to genetic testing or had the largest concerns around privacy issues or distrust of the healthcare system may not have chosen to participate in the qualitative interviews. This limitation was mitigated by performing outreach in three separate categories – including those who did not fill out the family history screening and those who did not receive a test after a recommendation – which ensured that the study included patients that stopped engaging early in the pathway. Additionally, these participants likely reflect to some extent a patient population that would be willing to consider screening and testing, if aspects of the program were improved; this study gives insight into what those considerations and changes might be.

Another limitation was in identifying participants from each of the three groups. The CARE™ patient portal was used for outreach; however, this portal was designed to support patient care rather than research purposes, and patient experiences and history with genetic testing was not always included. For example, of the seven interviews conducted with participants categorized in the program data as Group 2 (i.e., did not receive genetic screening after the recommendation), only four had never actually received a genetic test for cancer risk in their lifetime. The other three had received genetic testing for cancer risk years prior to the CARE recommendation, received the genetic test in another country, or received the test after the CARE recommendation when they were referred again by their primary care provider. We addressed this limitation by adjusting interview guides based on a participant's actual experiences, rather than our initial categorization.

Additionally, the study only included participants who were scheduled to receive or recently received a mammogram. Individuals who do not receive this kind of screening, including those that are generally not engaged with the health care system, are not represented in our findings. This population would likely face different barriers to receiving genetic screening or testing recommendations. Future research could focus on the equity considerations of engaging missing populations in these genetic risk programs.

CONCLUSION

Interviews with participants at multiple stages of a genetic screening and testing program pathway suggest that patients face similar barriers at each step in the program, as they try to understand and prioritize the relative importance of screening and testing for their personal health and healthcare. Patient education around the benefits of the program to patients and their families, as well as clear communication about what testing entails may help patients engage with the program. Strategies aimed

at increasing coordination among a patient's healthcare team can help present information to patients in multiple ways, from multiple providers, to help ensure that recommendations come from trusted sources and support the uptake of genetic testing.

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This study was funded by [*ORGANIZATION #1 and 2*].

Competing Interests

The authors have no competing interests to declare that are relevant to the content of this article.

Ethics Statement

The protocol for this study was approved by the [*ORGANIZATION #1*] Institutional Review Board (IRB # 2023000128). Participants provided verbal informed consent prior to enrollment in the study.

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Authors' Contributions

The author contributions were as follows: All authors contributed to the study conception and design. Material preparation, data collection and analysis were performed by [*AUTHOR #1 and #6*]. The first draft of the manuscript was written by [*AUTHOR #1 and 6*] and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

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Experiences across a genetic screening and testing program pathway: a qualitative study of mammogram patient perspectives

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dverseTITLE: Experiences across a genetic screening and testing program pathway: a qualitative study of mammogram patient perspectives

ABSTRACT

Background: Population-based genetic screening and testing programs have substantial potential to improve cancer-related outcomes through early detection and cancer prevention. Yet, genetic testing for cancer risk remains largely underutilized. This study aimed to describe barriers and facilitators to patient engagement at each stage of a California-based genetic screening program, from completing the electronic screener to receiving the test, and to identify potential improvements that could support precision medicine-based approaches to patient care.

Methods: We conducted 26 semi-structured interviews among program participants who did not complete the screener (n=9), those who did not receive the recommended test (n=7), and those who received a genetic test (n=10). Interviewees were selected from patients who recently received a mammogram through one of the participating Southern California clinics. Interviews were transcribed and coded using Atlas.ti. The study used a qualitative descriptive approach to identify similar and contrasting themes among the participant groups.

Results: This study found that barriers and facilitators to engagement were largely the same regardless of how far participants had moved through the process toward getting a genetic test. We identified four overarching themes: participants wanted clear communication of personal benefits at each stage; participants needed additional information and knowledge to navigate genetic screening and testing; a trusted provider could be instrumental in participants following a recommendation; and repetition and timing strongly impacted participants' likelihood to engage.

Conclusions: Providing education around the benefits of genetic screening and testing to patients and their families, as well as clear communication about what each step entails may help patients engage with similar programs. Strategies aimed at increasing coordination among a patient's healthcare team can also help ensure information reaches patients in multiple ways, from multiple providers, to increase the likelihood that recommendations for testing come from trusted sources, which supports the uptake of genetic testing.

Strengths and Limitations of this Study:

- In-depth interviews with patients expand our understanding of patients' experiences and preferences for a population-based genetic screening program.
- This study offers insights on how to promote engagement at multiple stages of a genetic screening and testing pathway.
- The majority of interviewees identified as white, women, with English as a preferred language, which limits generalizability.
- The study was limited to participants already engaged in healthcare and does not address overall barriers and access to care.

INTRODUCTION

A growing number of genetic screening programs have been implemented in multiple settings (e.g., primary care, specialty ambulatory care like cardiology, etc.) to identify high-risk patients in need of additional preventive treatment and connect them to needed follow up care [1,2]. Programs that offer population genetic screening to assess cancer risk are increasingly viewed as a key means for supporting population health overall because of their potential to identify patients in need of enhanced monitoring or who could benefit from risk-reducing treatments to prevent or mitigate symptoms [3,4]. Indeed, conducting additional cancer-risk assessment during routine screening appointments such as mammograms has become a recommended standard of care [5] which creates a critical need for these centers to implement efficient, scalable tools and workflows to support the risk assessment and genetic screening of their entire served population. Population-based genetic screening programs can also help mitigate some of the barriers that have been identified in more standard genetic service delivery models, such as long wait-times, limited genetic counselors, and overall accessibility. However, genetic screening and testing for cancer risk remains largely underutilized among patients [6,7], raising questions about patient perceptions of these programs, including benefits and barriers, to understand how to encourage participation.

Leveraging genetic testing to assess cancer risk has substantial potential to improve cancer-related outcomes through early detection and cancer prevention [8–10]. Yet, for these programs to be effective, patients must be actively engaged through a multi-step process. This includes the completion of a personal and family history screener to qualify for testing, the completion of a genetic test if they meet the recommended risk threshold, and connection to appropriate cancer screening and prevention care if patients are identified to have elevated cancer risk.

Despite population-based cancer risk genetic testing programs requiring multiple steps, previous studies tend to assess general perceptions of genetic testing from individuals who were not being offered a genetic test [11–13] or post-test reactions of participants who completed the genetic testing [1,14–16]. Few studies have focused on learning about the barriers and facilitators to engagement from patients who drop out of the program at each stage – including those who do not complete the risk assessment and those who do not receive a recommended genetic test. For these programs to be ultimately successful it is essential to understand the factors that shape patients' uptake and engagement in genetic testing at each stage of the process.

As genetic screening programs become more common, it is important to understand patient experiences of these programs, and what may motivate patients to complete the full genetic screening and testing process. This qualitative study used interviews with patients who had engaged to various degrees with a cancer screening program in a clinical setting to identify barriers and facilitators to patient engagement at each stage in the process, patient perspectives on the program, and what could be improved to support precision medicine-based approaches to patient care.

MATERIALS & METHODS

Setting

In 2020, the Providence Clinical Genetics and Genomics team in Southern California launched the Comprehensive Assessment, Risk, Education developed by Ambry Genetics (The Ambry CARE Program®), a population-based approach designed to increase cancer-related genetic testing for preventive purposes [17]. The program uses a text or email-based link that directs patients to an

electronic screener to gather information about personal and family cancer history from patients who are due to receive a mammogram through one of six participating clinics (See Figure 1). Patients are sent the screener one week prior to their scheduled mammogram and reminders one to three days prior. Patients who meet the National Comprehensive Cancer Network (NCCN) criteria of "high risk" [18] are recommended to receive a multi-gene "panel" genetic testing for alterations in more than 80 genes associated with hereditary cancer risks across a broad spectrum of tumor sites in addition to breast (uterine, ovarian, colorectal, pancreatic, etc.). After being identified as meeting NCCN criteria, the electronic platform provides patients with their recommendation for genetic testing and a pre-test educational video. Patients receive the screening and/or genetic testing recommendation at their mammogram appointment, and have the opportunity to ask questions, provide consent for testing, and collect their sample on the same day of their visit; they may also elect to schedule a traditional pre-test genetic counseling appointment with a genetic counselor if they wish. For eligible patients who do not present for genetic testing on the day of their mammogram, the genetics clinic follows up to offer sample collection through alternate means, such as returning to a testing center on a different day, sending a kit to the patient's home, or scheduling a genetic counseling appointment to discuss further. Patients who undergo genetic testing and test positive for a pathogenic variant in any of the genes are offered genetic counseling and receive detailed risk reduction, prevention, and management recommendations. Program data on which patients completed the screener, which received the testing recommendation but did not receive genetic testing, and which completed the full process are collected as part of routine care and program oversight.

At the time of the study, around 150,000 electronic screeners had been sent to patients, with an approximately 70% completion rate. A little less than 30% of these patients met NCCN criteria and received a testing recommendation. Of these, about 15% of patients eligible to receive testing go on to complete genetic testing. The institutional review board of Providence Health & Services (Oregon) approved the study (#2023000128).

Participant Selection

In May through June 2023, we conducted qualitative interviews with patients who had recently received or were due to receive a mammogram through one of the participating clinics. We used program data to identify potential interviewees and to classify them according to where they were along the workflow of receiving the electronic screener, completing the information, receiving the recommendation that they meet guidelines for hereditary risk assessment, and receiving the genetic test. Of note, these classifications were based on the patient's point along the workflow for their current appointment rather than any past appointments in which they may have been exposed to the electronic screener. As described in more detail in the limitations, a greater proportion of participants had previously received genetic testing than anticipated during recruitment. Email invitations were sent to all patients who met the study criteria; outreach materials and patient interviews were conducted in English. Participants reviewed consent forms with the interviewer and gave verbal consent separately to the interview procedure and to electronic health records (EHR) release.

Public and Patient Involvement

Patients or the public were not involved in the design, conduct, analysis, or dissemination of the research study. However, patients were central to the study in determining the relevant barriers and facilitators to genetic testing. In addition, based on the results of patient interviews and what we

learned about patient misconceptions and areas of interest, we provided feedback to program staff, who updated a Frequently Asked Questions document that was distributed to all participants in the study.

Data Collection

Three semi-structured interview guides were developed for the groups: (1) those who had received the screener but had declined to fill out the information or had not yet completed it, (2) those who met NCCN criteria for high risk but declined testing or had not yet decided to receive testing, and (3) those who received a positive genetic test result (i.e., pathogenic variant(s) detected). While patients who tested negative would have insight into what encouraged them to take the genetic test, our study was limited to patients who tested positive for a variant so that interviews could explore patients' experiences receiving genetic counseling and risk reduction recommendations. In addition, the experiences of patients who had a variant were of the most interest to the program team to help ensure quality improvement across all stages as individuals who test negative for a variant are standardly offered genetic counseling as part of the program for tailored empiric risk assessment, whereas it is a requirement for individuals who test positive.

Participants were asked open-ended questions about their views on genetic testing, what could or did motivate them to receive a genetic test (including their history of prior testing), and any barriers or facilitators they encountered during the process. Interview domains were developed after a literature search on barriers and facilitators to genetic testing and review with the Providence Clinical Genetics and Genomics team (See Supplemental Table 1). Participant experiences with genetic testing did not always align with data captured by the program data; interview questions were adjusted based on this information.

We aimed to contact patients in the first two groups who had completed their mammograms within a month of study outreach. For patients in the tested-positive group, we conducted outreach with those who had tested positive at least five months prior. Interviews were between 30 and 60 minutes and were conducted either virtually or via the phone, based on participant preference. Interviews were recorded and transcribed verbatim by a transcription service and reviewed for accuracy by the research team.

Data Analysis

Interview data were analyzed using a qualitative descriptive approach, which stays closer to the data than other qualitative approaches, provides in-depth content from the participants' perspective, and is well-suited to health care research [19]. Initial codebooks were developed from the interview guide and revised iteratively when emergent codes were identified. To maintain methodological rigor, three of the interviews were double coded. After each double-coded interview, the coding team met to discuss codes until consensus was reach. The team reviewed the content of the codes to develop a set of central themes. Illustrative quotes are presented in the results section to support the themes and recommendations. All analyses were completed in Atlas.ti version 23. Additionally, information from electronic health record data was pulled to demographically describe the sample.

RESULTS

A total of 26 interviews were conducted. Participants were primarily White and English as a preferred language. Table 1.

Table 1. Participant Demographics

Characteristic	Number or Mean
Participant Categorization	26
Did not complete screener	9
Did not receive genetic test	7
Tested positive	10
Age in years (mean)	59.2
Gender	
Women	23
Preferred Language	
English	23
Race/Ethnicity	
White	21
Asian	2
Hispanic/Latina	1
Insurance Type	
Commercial	16
Medicare	6
Unknown/Uninsured	1

Data do not sum to 26 (i.e., 100%) due to absence of data. Three participants did not consent to have their interview data linked to their electronic health record data.

Four themes emerged from interviews with all participants, regardless of how far they had moved through the process toward completing a genetic test. These included: (1) the desire for clear communication of benefits, (2) the need for more information and knowledge, (3) the importance of a trusted provider, and (4) the advantages of repetition and timing. Additional exemplar quotes found in Table 2.

Communication of Benefits

At each stage of the CARE program pathway, from filling out the electronic screener to receiving the genetic test, participants were motivated to complete the tasks based on their understanding of the importance of the steps. Many of the participants did not understand the purpose of the screener when they received it; nearly all of participants who did not complete the screener mentioned that they would have been more likely to complete it or find the task important had they known that it would be used to determine if they should receive a genetic test or otherwise inform the type of healthcare they would receive. Participants did not differentiate the electronic screener from other pre-appointment paperwork and forms; to most, it appeared to be the same as many of the other family history questionnaires that they were asked to fill out for other medical visits. Participants often expressed frustration at completing a seemingly redundant form.

> *"Sometimes I think 'I just did this last week,' or 'Do you have to know this again?' That's the thing I can say about it that's a negative…It's not difficult [but] it takes time."*
> - Participant who did not complete the screener

In terms of receiving the genetic test, participants did not always realize that the recommendation was tied to their family history and an increased personal risk for certain cancers; participants suggested that understanding this link may have increased their motivation to receive the test.

> *"I think that [knowing the connection between the screener and a genetic test recommendation] would change [my willingness to complete the screener] tremendously because it's like you're using your family history to figure out if it's a possibility that that relates to you."*
> - Participant who did not complete the screener

> *"You know, when I did the genetic testing, it was basically like, 'Do you wanna do it?' …There wasn't, you know, 'This is gonna be very helpful.' Of course, it's helpful, but the bottom line was, 'You can do genetic testing if you're interested.' So I was, because of all that history that I had on my dad's side, thinking, you know, that has to be it."*
> - Participant who received test

Many of the interviewees who engaged in the testing were motivated by being able to share their results with their family members. This benefit was not always clearly understood by interviewees who did not complete the screener or take the test; a better understanding of the connection may have increased engagement.

> *"Although, you know, I was thinking too, it's like I just had a grandchild born, so I think probably genetics is like more important to me now, now that there's, you know, another generation that's maybe a motivation as well."*
> - Participant who did not complete the screener

Information and Knowledge

A large proportion of interviewees had prior exposure to some kind of genetic testing through genetic testing associated with pregnancy, direct-to-consumer genetic testing, or genetic testing after a personal cancer diagnosis. Despite this previous exposure, some participants were still unsure what the recommended genetic test entailed, including whether it involved a blood test or biopsy or required a hospital stay.

> *"I'm not even sure what entails genetic testing. Is it a blood test? Is it, you know, a biopsy kind of thing?"*
> - Participant who did not receive the recommended test

One person who chose not to get the genetic test expressed confusion between the genetic tumor testing the participant had already received when diagnosed with breast cancer, and the recommendation to undergo genetic testing to assess cancer risk. Participants who had not completed the screener gave similar responses. Some indicated that they would want more information about the specifics around the test, if it were recommended, including both general information about the test (e.g., how the test is done and how long it takes) and whether there were any medical risks (e.g., one participant who was concerned because they were "*allergic to everything*").

"I thought it was saying that if you have cancer, you know, if I'm already sick, would I be interested in it. And that was probably in my mind, too. Like 'No, I'm fine, I don't need this" …So I just didn't understand it."

- Participant who did not receive the recommended test

None of the participants (either those who had not received the screener or who did not receive the test) were fully opposed to the idea of genetic testing for cancer risk. Participants who were more hesitant about genetic testing wanted to know the personal benefit and expressed concerns about cost, but none of the interviewees had overriding objections to knowing about their genetic cancer predisposition because they felt that knowledge was power ("ignorance is bliss, right? But I feel like I'd rather in control of some risks as opposed to it surprising me"). A few interviewees mentioned that they had encountered people (including family members they reached out to after receiving a test) who "*put their head in the sand…like an ostrich, they just don't want to know*" but most of the participants who received the testing recommendation already had a heightened awareness of their risk given their family and sometimes personal history with cancer.

"That website form it said something about genetic testing and have I had it? No. Would you be interested? Yes. And the reason why I wanted to do it was because my father, mother, brother, and sister have all had cancer. I have not, and three of those have passed away due to cancer. And my sister had breast cancer…So I was just interested in how their cancer…pertained to me."

- Participant who received the testing recommendation but was unable to complete the test

Importance of a Trusted Provider

The provider making the recommendation also mattered to some participants. A number of participants expressed confusion based on their providers' level of emphasis on the testing recommendation, as well. Interviewees described the recommendation as a "suggestion" from their mammogram technicians that was not accompanied by a sense of urgency, which made it difficult for them to judge the importance of the recommendation for testing. One participant who eventually received genetic testing did so because when their primary care provider recommended the test, they emphasized how important it was for the participant to be tested given their family history. Interviewees felt that the recommendation from the CARE program could be communicated more strongly, and one interviewee suggested that receiving the recommendation from their doctor (as opposed to a technician or other staff) would make it appear more important.

"It wasn't like, "Oh, we think this would be good for you." […] versus, "If you wanna do it, you can, it doesn't matter." You know, it was just a matter of fact, "You can have it done if you want."

- Participant who received the genetic test

"I'm not sure that coming from a tech, it would carry a lot of weight…if [my oncologist] suggested it to me, I would probably do it."

- Participant who did not receive the genetic test

Many participants across the CARE program pathway described strong, trusting relationship with at least one provider – usually a primary care provider or oncologist – whose recommendations they were most likely to follow; none identified mammography staff as that trusted source of information. Participants

that received a genetic test and tested positive for at least one genetic marker typically communicated a positive relationship with the genetic counselor.

> *"[The genetic counselor] was very thorough. It went really well. She was very, um, kind and understanding and you know, I'm sure she gets various reactions of people when they go in there. So, she was very comforting and very reassuring."*
> - Patient who tested positive

Participants were given the option to see the counselor before receiving the test, however, most opted to take the test first, for some because of the convenience of taking the test quickly. Some participants who later tested positive wished that they had been directed more strongly toward the option of meeting the genetic counselor prior to testing, so that they could receive more information about possible life insurance effects or additional explanation around possible test results.

> *"I didn't know what was going on…so I think it was just like a big surprise. And I mean granted part of it is I chose not to meet with the genetic counselor first. And looking back I think I would've, but I just didn't know and I was going with whatever seemed easiest.*
>
> *I wasn't aware that I should have life insurance before I do genetic testing in case something shows up…which is a huge factor…I think part of it is I didn't ask questions and I wasn't really proactive in this care. Because I honestly never believed I had a genetic mutation."*
> - Patient who tested positive

Repetition and Timing

Interviews with participants who did and did not receive the genetic test suggested that, while there may not be immediate uptake by those who receive the recommendation, hearing about genetic testing multiple times and in multiple ways has a long-term effect of encouraging appropriate patients to receive testing. Many of the participants who were eventually successful at moving through the CARE program pathway (i.e., completed the screener and took the genetic test) had received genetic testing recommendations before. Their decision to receive the test at this point, rather than when it was suggested first, was based on a confluence of contextual factors, including changing family and personal cancer experiences and changes in the cost of the testing, etc. As mentioned previously, some participants received the test after getting the recommendation from multiple types of providers.

> *"I was just worn out...and I'd been through so much. I think it was at one of those points where I just felt like I just needed to like curl up and lick my wounds and recover and kind of hide for a while."*
> - Participant who did not receive genetic testing after initial recommendation but received it after a subsequent recommendation

Similarly, participants who did not complete the screener described being busy with family and work obligations, but none expressed a complete refusal to share the information or fill out the forms. They described generally completing documentation of information like family history prior to a medical appointment but were unable to do so for their most recent mammography appointment.

"And then this year...I can't remember what was going on…I was like, I don't have time to do this, so I just didn't do it. I figured I'll do it next year."

- Participant who did not complete the screener

Table 2. Themes and exemplar quotations

Communication of benefits	*"So I went in for my first mammogram and I think you do a risk factor analysis that like gives you a number of your like lifetime risk of breast cancer, right? And my number was really high. And so I remember I did the mammogram and then they said, 'Do you wanna do genetic testing?' And I said, 'No, I don't need to know.' And it was just the ultrasound tech, he was offering it and I really didn't have any understanding or-or like, I had no idea like the extent that it could provide information.* *What ended up happening is I got a new younger lady as a PCP and I went to see her…and I was sharing my history and she said, 'You absolutely need genetic testing.' She's like, 'You can't get to the bottom of breast cancer. Like there's too many factors involved. Like you need to do genetic testing. I'm sending you to see a genetic counselor. Like you need to get this done.' And she was the first person who ever like flat out told me to my face that like, I need to get it done."* - Participant who did not receive genetic testing after initial recommendation but received it after a subsequent recommendation
Information and knowledge	*"I didn't know what it entailed. I think if I would have known more of what it entailed…there would be no fear or anything like that. [But the recommendation] is just in with all the other stuff…it was just real quick. I don't remember them talking about it in particular."* - Participant who did not receive the genetic test *"I probably wouldn't have asked had my doctor not recommended it. 'cause I just assumed…insurance wouldn't cover it, but, if they just made it easier to get as far as saying, 'Yes…your insurance will cover this, do you want it?' I think a lot more people would get it."* - Participant who received the genetic test *"Well, I'd wanna know, what do they do? How do they do it? How long it takes."* - Participant who did not complete the screener *"I asked if it was gonna be, um, like a blood test or saliva 'cause I wasn't sure."* - Participant who tested positive
Importance of a trusted provider	*"The only interaction I have is with the techs and ... if they threw it out there, it was very casual, but no one has ever sat down with me and said, 'Let's talk about doing some genetic counseling.' It's never been that formal. And it's never been with a doctor. It's only-- I've only met with techs... You know, I think it would mean more coming from a doctor."* - Participant who did not receive the genetic test *"Don't send me a letter telling me that you're recommending genetic testing. I mean, you know, that's just gonna freak me out. It's like, 'Okay, well what are you seeing? You know, why is this a concern all of a sudden? You know, why do you*

	care?" Because my first go-to is to panic that, 'Oh my gosh, something's, you know, something's back and you saw something on the mammogram.' So I'd prefer not to be freaked out in the process." - Participant who did not receive the genetic test
Repetition and timing	*"And then this year...I can't remember what was going on. I think my husband had a…surgery and my son, his [school] team was competing. I was like, I don't have time to do this, so I just didn't do it. I figured I'll do it next year. But yeah...it takes a really, really long time to do those forms."* - Participant who did not complete the screener *"I found that…a lot of the stuff I just didn't wanna know about, you know. I was just pretty focused on getting rid of the cancer and jumping through the hoops that the doctor wanted me to do. So all the other stuff, it's like, 'Okay, you just do your thing and-and I'm just gonna try not to die during chemo.'"* - Participant who did not receive the genetic test initially

DISCUSSION

Population-based genetic screening and testing programs can help identify patients at an increased risk of cancer and direct them toward enhanced cancer screening services (e.g., mammograms, colonoscopies, ultrasounds, magnetic resonance imaging etc.) or other recommendations. However, for these programs to be successful, patients must be effectively engaged at each step of the program pathway. Although study participants had varying degrees of engagement in the CARE program pathway, four themes emerged highlighting barriers and facilitators to participation that cut across the different groups: the benefits around clarity of communication, the need for increased information and knowledge, the importance of a trusted provider, and the effects of repetition and timing.

The themes we identified largely indicate that the reasons participants have for not engaging in testing are potentially mutable, and that patients are willing to engage in genetic screening and testing when they have a clear understanding of the specific purpose and procedures at each point in the pathway. Similar to prior research, our study found that patients are open to hearing about their genetic risks [11] but are most likely to support testing when there are clear medical benefits, and when testing is for specific diagnostic and predictive purposes [12,20,21]. These findings reiterate that a genetic risk assessment program, such as CARE, needs to clearly articulate to patients the diagnostic and predictive purposes of genetic screening and testing and how the testing would impact their future healthcare. Programs could consider opportunities to integrate patient communication and education during appointments, such as when mammogram patients are waiting to be seen.

Interestingly, while a limited number of individuals suggested that "not wanting to know" factored into their thoughts around receiving a genetic test, the fear of emotional and psychosocial effects from genetic testing results that have been cited in other literature [20,22,23] was not frequently identified by participants in this study. In fact, most participants felt that "knowledge was power" and having more information could help in their care. These findings may suggest a growing acceptance of genetic testing, with knowledge and provider relationships as the main factors affecting engagement.

Other barriers to genetic testing identified by patients in the study included effects on insurance and potential costs, which aligns with findings from previous studies [1,13,24]. Interviewees also expressed being frustrated by redundant requests for information that did not feel relevant to a given appointment or were not addressed by their healthcare provider during appointments. This highlights a need for improved portability and visibility of patient-provided history, particularly for patients who return year over year to the same center or see multiple providers within the same network. Moreover, patients at each stage in the pathway described their own limited capacity, in terms of time and attention, to prioritize, navigate, and complete each request and recommendation offered by the healthcare system.

Communication from a trusted provider helped counteract barriers to genetic testing and was identified as a key facilitator to moving forward in the process for many participants. Previous research has also underscored the importance of hearing the recommendation for genetic testing from a trusted provider, especially among racially and ethnically diverse patients [25–27]. In addition, patient navigators have been shown to improve patient follow-through on genetic testing [28]. Despite patients' desire to hear this information from a provider, few studies have evaluated the impact of educational training programs designed to improve health care providers' communication skills related to genetic testing and cancer risk on the uptake of genetic testing [29,30]. This highlights the need for additional research to identify strategies to effective support non-genetic clinicians in the provision of this type of patient counseling. Our findings also suggest that patients who opt to get genetic testing do not always choose to talk to a genetic counselor prior to testing; patient navigators may also be able to provide information about talking to a genetic counselor before getting testing, and helping a patient determine if that is the best option for them.

The findings highlight the importance of supporting the coordination of health care team around genetic testing and risk assessment. Improved coordination of the health care team may help ensure that multiple providers address the recommendation with patients at multiple time points and increase the feeling of importance around the test. Additionally, expanding the languages the screener is available in is an important way to support equitable access to testing, especially in linguistically diverse regions like Southern California.

Limitations

As mentioned before, self-selection bias was a limitation in this study; participants who were completely opposed to genetic testing or had the largest concerns around privacy issues or distrust of the healthcare system may not have chosen to participate in the qualitative interviews. This limitation was mitigated by performing outreach in three separate categories – including those who did not fill out the family history screening and those who did not receive a test after a recommendation – which ensured that the study included patients that stopped engaging early in the pathway. Additionally, these participants likely reflect to some extent a patient population that would be willing to consider screening and testing, if aspects of the program were improved; this study gives insight into what those considerations and changes might be.

Another limitation was in identifying participants from each of the three groups. The CARE patient portal was used for outreach; however, this portal was designed to support patient care rather than research purposes, and patient experiences and history with genetic testing was not always included. For example, of the seven interviews conducted with participants categorized in the program data as Group

2 (i.e., did not receive genetic screening after the recommendation), only four had never actually received a genetic test for cancer risk in their lifetime. The other three had received genetic testing for cancer risk years prior to the CARE recommendation, received the genetic test in another country, or received the test after the CARE recommendation when they were referred again by their primary care provider. We addressed this limitation by adjusting interview guides based on a participant's actual experiences, rather than our initial categorization.

There are also limitations around the number of participants that were interviewed for the study, which limits the generalizability of the findings. In addition, participants largely identified as white, and with English as a preferred language; the program was implemented in a number of Southern California clinics, which also limits the geographic scope and relevance of the study. Participants also overwhelming identified as women, with an average age of 59 years. While this expected, since the program was implemented in a mammogram setting, it affects how much the findings can be generalized to other genders and age groups. Additionally, the study only included participants who were scheduled to receive or recently received a mammogram. Individuals who do not receive this kind of screening, including those that are generally not engaged with the health care system, are not represented in our findings. This population would likely face different barriers to receiving genetic screening or testing recommendations. Future research could focus on the equity considerations of engaging missing populations in these genetic risk programs.

CONCLUSION

Interviews with participants at multiple stages of a genetic screening and testing program pathway suggest that patients face similar barriers at each step in the program, as they try to understand and prioritize the relative importance of screening and testing for their personal health and healthcare. Given the growth of genetic risk assessment in outpatient care settings, the results of study provide helpful insights to improve patient engagement with risk assessment and uptake of genetic testing. Patient education around the benefits of the program to patients and their families, as well as clear communication about what testing entails may help patients engage with the program. Strategies aimed at increasing coordination among a patient's healthcare team can help present information to patients in multiple ways, from multiple providers, to help ensure that recommendations come from trusted sources and support the uptake of genetic testing.

Data Availability

The data generated and analyzed in this study were derived from in-depth interviews and are not publicly available due to restrictions, namely the contain information that could compromise the privacy of research participants.

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Competing Interests

The authors have no competing interests to declare that are relevant to the content of this article.

Ethics Statement

The protocol for this study was approved by the Providence Institutional Review Board (IRB # 2023000128). Participants provided verbal informed consent prior to enrollment in the study.

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Authors' Contributions

The author contributions were as follows: All authors (CD, KE, KC, SB, OG, SR) contributed to the study conception and design. Material preparation, data collection and analysis were performed by CD and SR. KE, KC, SB, and OG aided in the interpretation of results and implications for practice. The first draft of the manuscript was written by CD and SR and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript. SR is responsible for the overall content as guarantor.

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Figure Legend:

Figure 1 – CARE Program workflow

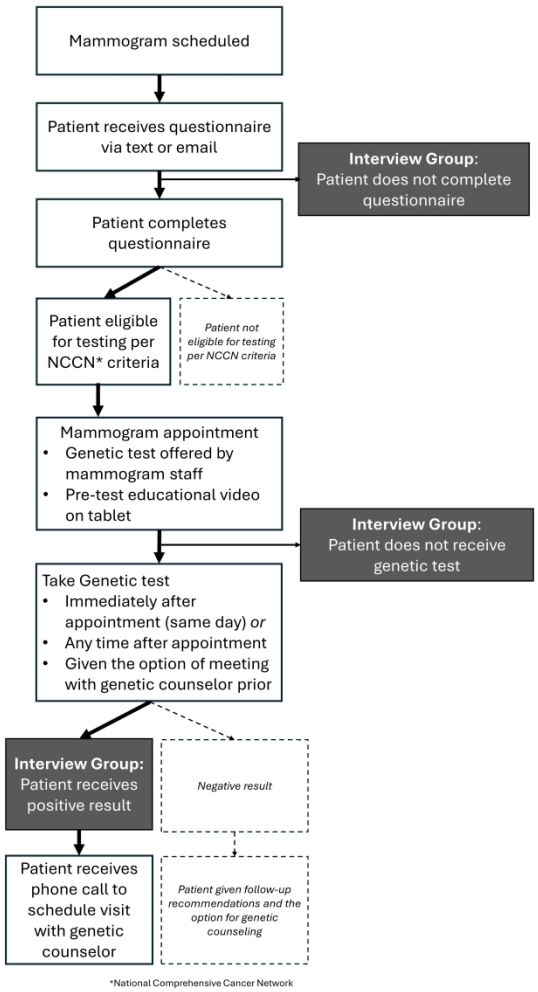

Figure 1. CARE Program workflow

254x508mm (300 x 300 DPI)

Supplemental Table 1: Semi-structured Interview Guides

Group 1: Received questionnaire, didn't complete

- Can you describe your experience getting ready for your mammogram through Providence?
  - *What do you remember around: scheduling, reminders, staff, communication with PCP, etc.*

- Part of getting a mammogram is about understanding your cancer risk. How important is it to you to understand your cancer risk?
  - *Do you feel like you are at particularly high risk of cancer?*

- What do you remember about receiving an email or text to complete a questionnaire prior to your mammogram?
  - *Can you describe your experience with receiving the questionnaire?*
    - *Do you recall if you opened or started the questionnaire? If so, why did you stop?*
    - *Do you recall why you chose not to fill it out ahead of the appointment? If so, can you explain why?*
    - *Do you recall the front desk staff asking you about completing the family history questionnaire? If so, can you describe experience?*

- What sort of things make it difficult to fill out healthcare questionnaires received via text or email?
  - *To what extent do you feel you have sufficient time during the day to fill out the forms?*
  - *To what extent does the information requested seem important enough to fill out?*
  - *How easy or difficult are the family history questions easy for you to fill out?*
  - *To what extent is the language on the forms a barrier to completing the questionnaires?*

- What's your preferred method of receiving health or family history questionnaires prior to an appointment?
  - *Are you comfortable with receiving them as a text? Email?*
    Would you prefer someone ask you the questions directly during your appointment?

- What would make it easier for you to fill out those kinds of questionnaires?
  - *Examples: right before the appointment, same day as the appointment, during the appointment*
  - *Would reminders to fill out the form be helpful?*

- How important does it feel to share information on your family medical history with your healthcare providers?
  - *How do you expect the information will be used?*

- In general, what concerns do you have, if any, around sharing information like family and personal medical history of cancer with your healthcare provider prior to your healthcare appointments?

- *Examples: Mistrust of the healthcare system, don't see the importance of sharing the information, overly personal information, not sure about family history.*
- *Are you more comfortable sharing this information with your primary care doctor versus the provider giving you a mammogram, with the Providence health system, etc.*
- *What would make it easier for you to share your personal health information like family history with your healthcare providers?*

- [*For those that recall receiving the questionnaires*] Were you aware that the questionnaire would be used to determine if a genetic test would be recommended to help assess cancer risk as part of your healthcare?

- To what extent would knowing that the answers to the questionnaire were used as part of a recommendation for genetic testing have made it seem more important?
  - *Would you be more likely to answer it?*
  - *Why or why not?*
  - *How important is it to you to know if you have a genetic predisposition for cancer?*

- If your healthcare provider recommended that you have a genetic test, what would your reaction be?
  - *Would you be willing to do so? Why or why not?*
  - *What concerns would you have?*
  - *Have you ever received genetic testing through someone else?*
    - *What made you decide to do the genetic testing?*

- Is there anything I didn't ask about around understanding your cancer risk, genetic testing, sharing family history information that you'd like to mention?

Group 2: Complete questionnaire, didn't complete testing

- Can you describe your experience with receiving the recommendation for genetic testing?
  - *Who told you about your results?*
  - *How did you feel?*
  - *What questions did you have?*
  - *What were you told were the next steps?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *How important did it seem to your health care for you to take those next steps?*
  - *Are there other ways that would have been more useful to receive the testing recommendation? (i.e., in person, multiple times)*

- How important is it to you to understand your cancer risk?
  - *Do you feel like you are at particularly high risk of cancer?*
    How important is it to you to know if you have a genetic predisposition for cancer?

- What concerns (if any) did you have around genetic testing?
  - *How concerned were you about the results you might receive?*
  - *How worried were you that results might affect your health insurance?*
  - *How worried are you that results might affect your employment?*
  - *How much do you trust the healthcare system when it comes to genetic testing?*

- Did you discuss any of your concerns with a healthcare provider?
  - *If yes, how did that conversation go?*
    - *What kinds of questions or concerns did you raise with your provider?*
    - *To what extent did you provider answer your questions?*
    - *To what extent did your provider address your concerns?*
    - *Were you able to receive the information that you needed from your provider? If not, why not?*
    - *Is there anything else the provider could have done to address your concerns?*
  - *If not, what would have made it easier to talk to a healthcare provider about your concerns?*

- Do you feel like you had enough information in deciding whether or not to get a genetic test?
  - *Is there anything that your healthcare provider could have communicated that would have encouraged you to get the test?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *What other information would have been helpful to have?*
  - *Did you get information or advice from someone other than a healthcare provider or in some other way?*

- What steps, if any, did you take toward getting a genetic test after receiving the recommendation?
- What things did you experience that made getting the test difficult?
  - *To what extent did you have any concerns about the cost of the test?*
  - *To what extent did you feel you had enough time to get the test?*

- *To what extent was getting transportation or getting to and from the test a barrier?*
- *What difficulties, if any, did you around scheduling a test?*
- *Were there language barriers that made receiving the recommendation or getting the test difficult? Please explain.*
- *Was there anything else that made getting the test difficult?*

- Overall, what was the primary reason you chose not to get a test?

- What changes could your provider or insurer make that would encourage you to get a genetic test?
  - *Examples: financial assistance, additional information, etc.*

- Have you ever received genetic testing another way, other than this program?
  - *Did you receive this testing prior to getting the recommendation from this program?*
    - *If yes, did you share the results of that test with your provider?*
      - *How did that conversation go?*
    - *If not, what encouraged you to get the other test?*
      - *Was there anything that made that test easier to get?*

- Other than the genetic testing, do you remember getting a recommendation for additional screening based on your family history?
  - *What was your experience receiving the recommendation?*
  - *Did your primary care doctor follow up about the recommendations with you?*
    - *If yes, please explain.*
    - *If not, to what extent would that have been helpful?*
  - *Were the recommendations for additional screening communicated clearly?*
  - *What has your experience been with getting additional screening?*
    - *Did you experience any barriers in receiving follow-up care?*
    - *How are you using the information?*

- Is there anything I didn't ask about around understanding your cancer risk, genetic testing, sharing family history information that you'd like to mention?

Group 3: Completed testing

- Can you describe your experience with receiving the recommendation for genetic testing?
  - *Who told you about your results?*
  - *How did you feel?*
  - *What questions did you have?*
  - *What were you told were the next steps?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *How important did it seem to your health care for you to take those next steps?*
  - *Are there other ways that would have been more useful to receive the testing recommendation? (i.e., in person, multiple times)*

- How important is it to you to understand your cancer risk?
  - *Do you feel like you are at particularly high risk of cancer?*

How important is it to you to know if you have a genetic predisposition for cancer?

- What concerns (if any) did you have around genetic testing?
  - *How concerned were you about the results you might receive?*
  - *How worried were you that results might affect your health insurance?*
  - *How worried are you that results might affect your employment?*
  - *How much do you trust the healthcare system when it comes to genetic testing?*

- Did you discuss any of your concerns with a healthcare provider?
  - *If yes, how did that conversation go?*
    - *What kinds of questions or concerns did you raise with your provider?*
    - *To what extent did you provider answer your questions?*
    - *To what extent did your provider address your concerns?*
    - *Were you able to receive the information that you needed from your provider? If not, why not?*
    - *Is there anything else the provider could have done to address your concerns?*
  - *If not, what would have made it easier to talk to a healthcare provider about your concerns?*

- Do you feel like you had enough information in deciding whether or not to get a genetic test?
  - *What other information would have been helpful to have?*
  - *Is there anything that your healthcare provider could have communicated that would have encouraged you to get the test?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *Did you get information or advice from someone other than a healthcare provider or in some other way?*
  - *What did you find the most helpful to know in making the decision to get the test?*

- What was the primary reason you decided to get testing?
- Can you describe your experience with getting the test itself?
  - *Was there anything that made that experience difficult?*
  - *Is there anything that could have changed to make the process easier?*

- Were there any factors that helped you get the test?
  - *Was the full cost of the test covered by insurance?*
  - *What difficulties, if any, did you have scheduling the test?*
  - *Did you have any assistance getting to and from the testing center?*
  - *Did anything else make getting the test easier?*
  - *After getting the test, was there anything else that you would have wanted to know beforehand? Please explain.*

- Can you describe your experience receiving the results of the test?
  - *Who gave you the results of your test?*
  - *What kind of information were you given when you received your results?*
    - *Was the information you received sufficient? Why or why not?*

- *What, if anything, would you have changed about the process or experience of receiving the results?*

- What was your experience receiving any follow-up recommendation based on your test results?
  - *What was your experience receiving the recommendation?*
  - *Did your primary care doctor follow-up about the recommendations with you?*
    - *If yes, please explain.*
    - *If not, to what extent would that have been helpful?*
  - *Were the recommendations for any additional screening or treatment communicated clearly?*

- What has your experience been with getting any additional screening or care?
  - *Did you experience any barriers in receiving follow-up care?*
  - *How are you using the information?*

- How has your knowledge or awareness of your genetic results changed your care?
  - *Has it prompted any other changes?*

- Is there anything I didn't ask about around understanding your cancer risk, genetic testing, or follow-up care that you'd like to mention?

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Experiences across a genetic screening and testing program pathway: a qualitative study of mammogram patient perspectives

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TITLE: Experiences across a genetic screening and testing program pathway: a qualitative study of mammogram patient perspectives

ABSTRACT

Background: Population-based genetic screening and testing programs have substantial potential to improve cancer-related outcomes through early detection and cancer prevention. Yet, genetic testing for cancer risk remains largely underutilized. This study aimed to describe barriers and facilitators to patient engagement at each stage of a California-based genetic screening program, from completing the electronic screener to receiving the test, and to identify potential improvements that could support precision medicine-based approaches to patient care.

Methods: We conducted 26 semi-structured interviews among program participants who did not complete the screener (n=9), those who did not receive the recommended test (n=7), and those who received a genetic test (n=10). Interviewees were selected from patients who recently received a mammogram through one of the participating Southern California clinics. Interviews were transcribed and coded using Atlas.ti. The study used a qualitative descriptive approach to identify similar and contrasting themes among the participant groups.

Results: This study found that barriers and facilitators to engagement were largely the same regardless of how far participants had moved through the process toward getting a genetic test. We identified four overarching themes: participants wanted clear communication of personal benefits at each stage; participants needed additional information and knowledge to navigate genetic screening and testing; a trusted provider could be instrumental in participants following a recommendation; and repetition and timing strongly impacted participants' likelihood to engage.

Conclusions: Providing education around the benefits of genetic screening and testing to patients and their families, as well as clear communication about what each step entails may help patients engage with similar programs. Strategies aimed at increasing coordination among a patient's healthcare team can also help ensure information reaches patients in multiple ways, from multiple providers, to increase the likelihood that recommendations for testing come from trusted sources, which supports the uptake of genetic testing.

Strengths and Limitations of this Study:

- In-depth interviews with patients expand our understanding of patients' experiences and preferences for a population-based genetic screening program.
- This study offers insights on how to promote engagement at multiple stages of a genetic screening and testing pathway.
- The majority of interviewees identified as white, women, with English as a preferred language, which limits generalizability.
- The study was limited to participants already engaged in healthcare and does not address overall barriers and access to care.

INTRODUCTION

A growing number of genetic screening programs have been implemented in multiple settings (e.g., primary care, specialty ambulatory care like cardiology, etc.) to identify high-risk patients in need of additional preventive treatment and connect them to needed follow up care [1,2]. Programs that offer population genetic screening to assess cancer risk are increasingly viewed as a key means for supporting population health overall because of their potential to identify patients in need of enhanced monitoring or who could benefit from risk-reducing treatments to prevent or mitigate symptoms [3,4]. Indeed, conducting additional cancer-risk assessment during routine screening appointments such as mammograms has become a recommended standard of care [5] which creates a critical need for these centers to implement efficient, scalable tools and workflows to support the risk assessment and genetic screening of their entire served population. Population-based genetic screening programs can also help mitigate some of the barriers that have been identified in more standard genetic service delivery models, such as long wait-times, limited genetic counselors, and overall accessibility. However, genetic screening and testing for cancer risk remains largely underutilized among patients [6,7], raising questions about patient perceptions of these programs, including benefits and barriers, to understand how to encourage participation.

Leveraging genetic testing to assess cancer risk has substantial potential to improve cancer-related outcomes through early detection and cancer prevention [8–10]. Yet, for these programs to be effective, patients must be actively engaged through a multi-step process. This includes the completion of a personal and family history screener to qualify for testing, the completion of a genetic test if they meet the recommended risk threshold, and connection to appropriate cancer screening and prevention care if patients are identified to have elevated cancer risk.

Despite population-based cancer risk genetic testing programs requiring multiple steps, previous studies tend to assess general perceptions of genetic testing from individuals who were not being offered a genetic test [11–13] or post-test reactions of participants who completed the genetic testing [1,14–16]. Few studies have focused on learning about the barriers and facilitators to engagement from patients who drop out of the program at each stage – including those who do not complete the risk assessment and those who do not receive a recommended genetic test. For these programs to be ultimately successful it is essential to understand the factors that shape patients' uptake and engagement in genetic testing at each stage of the process.

As genetic screening programs become more common, it is important to understand patient experiences of these programs, and what may motivate patients to complete the full genetic screening and testing process. This qualitative study used interviews with patients who had engaged to various degrees with a cancer screening program in a clinical setting to identify barriers and facilitators to patient engagement at each stage in the process, patient perspectives on the program, and what could be improved to support precision medicine-based approaches to patient care.

MATERIALS & METHODS

Setting

In 2020, the Providence Clinical Genetics and Genomics team in Southern California launched the Comprehensive Assessment, Risk, Education developed by Ambry Genetics (The Ambry CARE Program®), a population-based approach designed to increase cancer-related genetic testing for preventive purposes [17]. The program uses a text or email-based link that directs patients to an

electronic screener to gather information about personal and family cancer history from patients who are due to receive a mammogram through one of six participating clinics (See Figure 1). Patients are sent the screener one week prior to their scheduled mammogram and reminders one to three days prior. Patients who meet the National Comprehensive Cancer Network (NCCN) criteria of "high risk" [18] are recommended to receive a multi-gene "panel" genetic testing for alterations in more than 80 genes associated with hereditary cancer risks across a broad spectrum of tumor sites in addition to breast (uterine, ovarian, colorectal, pancreatic, etc.). After being identified as meeting NCCN criteria, the electronic platform provides patients with their recommendation for genetic testing and a pre-test educational video. Patients receive the screening and/or genetic testing recommendation at their mammogram appointment, and have the opportunity to ask questions, provide consent for testing, and collect their sample on the same day of their visit; they may also elect to schedule a traditional pre-test genetic counseling appointment with a genetic counselor if they wish. For eligible patients who do not present for genetic testing on the day of their mammogram, the genetics clinic follows up to offer sample collection through alternate means, such as returning to a testing center on a different day, sending a kit to the patient's home, or scheduling a genetic counseling appointment to discuss further. Patients who undergo genetic testing and test positive for a pathogenic variant in any of the genes are offered genetic counseling and receive detailed risk reduction, prevention, and management recommendations. Program data on which patients completed the screener, which received the testing recommendation but did not receive genetic testing, and which completed the full process are collected as part of routine care and program oversight.

At the time of the study, around 150,000 electronic screeners had been sent to patients, with an approximately 70% completion rate. A little less than 30% of these patients met NCCN criteria and received a testing recommendation. Of these, about 15% of patients eligible to receive testing go on to complete genetic testing. The institutional review board of Providence Health & Services (Oregon) approved the study (#2023000128).

Participant Selection

In May through June 2023, we conducted qualitative interviews with patients who had recently received or were due to receive a mammogram through one of the participating clinics. We used program data to identify potential interviewees and to classify them according to where they were along the workflow of receiving the electronic screener, completing the information, receiving the recommendation that they meet guidelines for hereditary risk assessment, and receiving the genetic test. Of note, these classifications were based on the patient's point along the workflow for their current appointment rather than any past appointments in which they may have been exposed to the electronic screener. As described in more detail in the limitations, a greater proportion of participants had previously received genetic testing than anticipated during recruitment. Email invitations were sent to all patients who met the study criteria; outreach materials and patient interviews were conducted in English. Participants reviewed consent forms with the interviewer and gave verbal consent separately to the interview procedure and to electronic health records (EHR) release.

Public and Patient Involvement

Patients or the public were not involved in the design, conduct, analysis, or dissemination of the research study. However, patients were central to the study in determining the relevant barriers and facilitators to genetic testing. In addition, based on the results of patient interviews and what we

learned about patient misconceptions and areas of interest, we provided feedback to program staff, who updated a Frequently Asked Questions document that was distributed to all participants in the study.

Data Collection

Three semi-structured interview guides were developed for the groups: (1) those who had received the screener but had declined to fill out the information or had not yet completed it, (2) those who met NCCN criteria for high risk but declined testing or had not yet decided to receive testing, and (3) those who received a positive genetic test result (i.e., pathogenic variant(s) detected). While patients who tested negative would have insight into what encouraged them to take the genetic test, our study was limited to patients who tested positive for a variant so that interviews could explore patients' experiences receiving genetic counseling and risk reduction recommendations. In addition, the experiences of patients who had a variant were of the most interest to the program team to help ensure quality improvement across all stages as individuals who test negative for a variant are standardly offered genetic counseling as part of the program for tailored empiric risk assessment, whereas it is a requirement for individuals who test positive.

Participants were asked open-ended questions about their views on genetic testing, what could or did motivate them to receive a genetic test (including their history of prior testing), and any barriers or facilitators they encountered during the process. Interview domains were developed after a literature search on barriers and facilitators to genetic testing and review with the Providence Clinical Genetics and Genomics team (See Supplemental Table 1). Participant experiences with genetic testing did not always align with data captured by the program data; interview questions were adjusted based on this information.

We aimed to contact patients in the first two groups who had completed their mammograms within a month of study outreach. For patients in the tested-positive group, we conducted outreach with those who had tested positive at least five months prior. Interviews were between 30 and 60 minutes and were conducted either virtually or via the phone, based on participant preference. Interviews were recorded and transcribed verbatim by a transcription service and reviewed for accuracy by the research team.

Data Analysis

Interview data were analyzed using a qualitative descriptive approach, which stays closer to the data than other qualitative approaches, provides in-depth content from the participants' perspective, and is well-suited to health care research [19]. Initial codebooks were developed from the interview guide and revised iteratively when emergent codes were identified. To maintain methodological rigor, three of the interviews were double coded. After each double-coded interview, the coding team met to discuss codes until consensus was reach. The team reviewed the content of the codes to develop a set of central themes. Illustrative quotes are presented in the results section to support the themes and recommendations. All analyses were completed in Atlas.ti version 23. Additionally, information from electronic health record data was pulled to demographically describe the sample.

RESULTS

A total of 26 interviews were conducted. Participants were primarily White and English as a preferred language. Table 1.

Table 1. Participant Demographics

Characteristic	Number or Mean
Participant Categorization	26
Did not complete screener	9
Did not receive genetic test	7
Tested positive	10
Age in years (mean [standard deviation])	59.2 [11.6]
Gender	
Women	23
Preferred Language	
English	23
Race/Ethnicity	
White	21
Asian	2
Hispanic/Latina	1
Insurance Type	
Commercial	16
Medicare	6
Unknown/Uninsured	1

Data do not sum to 26 (i.e., 100%) due to absence of data. Three participants did not consent to have their interview data linked to their electronic health record data.

Four themes emerged from interviews with all participants, regardless of how far they had moved through the process toward completing a genetic test. These included: (1) the desire for clear communication of benefits, (2) the need for more information and knowledge, (3) the importance of a trusted provider, and (4) the advantages of repetition and timing. Additional exemplar quotes found in Table 2.

Communication of Benefits

At each stage of the CARE program pathway, from filling out the electronic screener to receiving the genetic test, participants were motivated to complete the tasks based on their understanding of the importance of the steps. Many of the participants did not understand the purpose of the screener when they received it; nearly all of participants who did not complete the screener mentioned that they would have been more likely to complete it or find the task important had they known that it would be used to determine if they should receive a genetic test or otherwise inform the type of healthcare they would receive. Participants did not differentiate the electronic screener from other pre-appointment paperwork and forms; to most, it appeared to be the same as many of the other family history questionnaires that they were asked to fill out for other medical visits. Participants often expressed frustration at completing a seemingly redundant form.

> *"Sometimes I think 'I just did this last week,' or 'Do you have to know this again?' That's the thing I can say about it that's a negative…It's not difficult [but] it takes time."*
> - Participant who did not complete the screener

In terms of receiving the genetic test, participants did not always realize that the recommendation was tied to their family history and an increased personal risk for certain cancers; participants suggested that understanding this link may have increased their motivation to receive the test.

> *"I think that [knowing the connection between the screener and a genetic test recommendation] would change [my willingness to complete the screener] tremendously because it's like you're using your family history to figure out if it's a possibility that that relates to you."*
> - Participant who did not complete the screener

> *"You know, when I did the genetic testing, it was basically like, 'Do you wanna do it?' …There wasn't, you know, 'This is gonna be very helpful.' Of course, it's helpful, but the bottom line was, 'You can do genetic testing if you're interested.' So I was, because of all that history that I had on my dad's side, thinking, you know, that has to be it."*
> - Participant who received test

Many of the interviewees who engaged in the testing were motivated by being able to share their results with their family members. This benefit was not always clearly understood by interviewees who did not complete the screener or take the test; a better understanding of the connection may have increased engagement.

> *"Although, you know, I was thinking too, it's like I just had a grandchild born, so I think probably genetics is like more important to me now, now that there's, you know, another generation that's maybe a motivation as well."*
> - Participant who did not complete the screener

Information and Knowledge

A large proportion of interviewees had prior exposure to some kind of genetic testing through genetic testing associated with pregnancy, direct-to-consumer genetic testing, or genetic testing after a personal cancer diagnosis. Despite this previous exposure, some participants were still unsure what the recommended genetic test entailed, including whether it involved a blood test or biopsy or required a hospital stay.

> *"I'm not even sure what entails genetic testing. Is it a blood test? Is it, you know, a biopsy kind of thing?"*
> - Participant who did not receive the recommended test

One person who chose not to get the genetic test expressed confusion between the genetic tumor testing the participant had already received when diagnosed with breast cancer, and the recommendation to undergo genetic testing to assess cancer risk. Participants who had not completed the screener gave similar responses. Some indicated that they would want more information about the specifics around the test, if it were recommended, including both general information about the test (e.g., how the test is done and how long it takes) and whether there were any medical risks (e.g., one participant who was concerned because they were "*allergic to everything*").

"I thought it was saying that if you have cancer, you know, if I'm already sick, would I be interested in it. And that was probably in my mind, too. Like 'No, I'm fine, I don't need this" …So I just didn't understand it."

- Participant who did not receive the recommended test

None of the participants (either those who had not received the screener or who did not receive the test) were fully opposed to the idea of genetic testing for cancer risk. Participants who were more hesitant about genetic testing wanted to know the personal benefit and expressed concerns about cost, but none of the interviewees had overriding objections to knowing about their genetic cancer predisposition because they felt that knowledge was power ("ignorance is bliss, right? But I feel like I'd rather in control of some risks as opposed to it surprising me"). A few interviewees mentioned that they had encountered people (including family members they reached out to after receiving a test) who "*put their head in the sand…like an ostrich, they just don't want to know*" but most of the participants who received the testing recommendation already had a heightened awareness of their risk given their family and sometimes personal history with cancer.

"That website form it said something about genetic testing and have I had it? No. Would you be interested? Yes. And the reason why I wanted to do it was because my father, mother, brother, and sister have all had cancer. I have not, and three of those have passed away due to cancer. And my sister had breast cancer…So I was just interested in how their cancer…pertained to me."

- Participant who received the testing recommendation but was unable to complete the test

Importance of a Trusted Provider

The provider making the recommendation also mattered to some participants. A number of participants expressed confusion based on their providers' level of emphasis on the testing recommendation, as well. Interviewees described the recommendation as a "suggestion" from their mammogram technicians that was not accompanied by a sense of urgency, which made it difficult for them to judge the importance of the recommendation for testing. One participant who eventually received genetic testing did so because when their primary care provider recommended the test, they emphasized how important it was for the participant to be tested given their family history. Interviewees felt that the recommendation from the CARE program could be communicated more strongly, and one interviewee suggested that receiving the recommendation from their doctor (as opposed to a technician or other staff) would make it appear more important.

"It wasn't like, "Oh, we think this would be good for you." […] versus, "If you wanna do it, you can, it doesn't matter." You know, it was just a matter of fact, "You can have it done if you want."

- Participant who received the genetic test

"I'm not sure that coming from a tech, it would carry a lot of weight…if [my oncologist] suggested it to me, I would probably do it."

- Participant who did not receive the genetic test

Many participants across the CARE program pathway described strong, trusting relationship with at least one provider – usually a primary care provider or oncologist – whose recommendations they were most likely to follow; none identified mammography staff as that trusted source of information. Participants

that received a genetic test and tested positive for at least one genetic marker typically communicated a positive relationship with the genetic counselor.

> *"[The genetic counselor] was very thorough. It went really well. She was very, um, kind and understanding and you know, I'm sure she gets various reactions of people when they go in there. So, she was very comforting and very reassuring."*
> - Patient who tested positive

Participants were given the option to see the counselor before receiving the test, however, most opted to take the test first, for some because of the convenience of taking the test quickly. Some participants who later tested positive wished that they had been directed more strongly toward the option of meeting the genetic counselor prior to testing, so that they could receive more information about possible life insurance effects or additional explanation around possible test results.

> *"I didn't know what was going on…so I think it was just like a big surprise. And I mean granted part of it is I chose not to meet with the genetic counselor first. And looking back I think I would've, but I just didn't know and I was going with whatever seemed easiest.*
>
> *I wasn't aware that I should have life insurance before I do genetic testing in case something shows up…which is a huge factor…I think part of it is I didn't ask questions and I wasn't really proactive in this care. Because I honestly never believed I had a genetic mutation."*
> - Patient who tested positive

Repetition and Timing

Interviews with participants who did and did not receive the genetic test suggested that, while there may not be immediate uptake by those who receive the recommendation, hearing about genetic testing multiple times and in multiple ways has a long-term effect of encouraging appropriate patients to receive testing. Many of the participants who were eventually successful at moving through the CARE program pathway (i.e., completed the screener and took the genetic test) had received genetic testing recommendations before. Their decision to receive the test at this point, rather than when it was suggested first, was based on a confluence of contextual factors, including changing family and personal cancer experiences and changes in the cost of the testing, etc. As mentioned previously, some participants received the test after getting the recommendation from multiple types of providers.

> *"I was just worn out...and I'd been through so much. I think it was at one of those points where I just felt like I just needed to like curl up and lick my wounds and recover and kind of hide for a while."*
> - Participant who did not receive genetic testing after initial recommendation but received it after a subsequent recommendation

Similarly, participants who did not complete the screener described being busy with family and work obligations, but none expressed a complete refusal to share the information or fill out the forms. They described generally completing documentation of information like family history prior to a medical appointment but were unable to do so for their most recent mammography appointment.

"And then this year...I can't remember what was going on…I was like, I don't have time to do this, so I just didn't do it. I figured I'll do it next year."

- Participant who did not complete the screener

Table 2. Themes and exemplar quotations

Communication of benefits	*"So I went in for my first mammogram and I think you do a risk factor analysis that like gives you a number of your like lifetime risk of breast cancer, right? And my number was really high. And so I remember I did the mammogram and then they said, 'Do you wanna do genetic testing?' And I said, 'No, I don't need to know.' And it was just the ultrasound tech, he was offering it and I really didn't have any understanding or-or like, I had no idea like the extent that it could provide information.* *What ended up happening is I got a new younger lady as a PCP and I went to see her…and I was sharing my history and she said, 'You absolutely need genetic testing.' She's like, 'You can't get to the bottom of breast cancer. Like there's too many factors involved. Like you need to do genetic testing. I'm sending you to see a genetic counselor. Like you need to get this done.' And she was the first person who ever like flat out told me to my face that like, I need to get it done."* - Participant who did not receive genetic testing after initial recommendation but received it after a subsequent recommendation
Information and knowledge	*"I didn't know what it entailed. I think if I would have known more of what it entailed…there would be no fear or anything like that. [But the recommendation] is just in with all the other stuff…it was just real quick. I don't remember them talking about it in particular."* - Participant who did not receive the genetic test *"I probably wouldn't have asked had my doctor not recommended it. 'cause I just assumed…insurance wouldn't cover it, but, if they just made it easier to get as far as saying, 'Yes…your insurance will cover this, do you want it?' I think a lot more people would get it."* - Participant who received the genetic test *"Well, I'd wanna know, what do they do? How do they do it? How long it takes."* - Participant who did not complete the screener *"I asked if it was gonna be, um, like a blood test or saliva 'cause I wasn't sure."* - Participant who tested positive
Importance of a trusted provider	*"The only interaction I have is with the techs and ... if they threw it out there, it was very casual, but no one has ever sat down with me and said, 'Let's talk about doing some genetic counseling.' It's never been that formal. And it's never been with a doctor. It's only-- I've only met with techs... You know, I think it would mean more coming from a doctor."* - Participant who did not receive the genetic test *"Don't send me a letter telling me that you're recommending genetic testing. I mean, you know, that's just gonna freak me out. It's like, 'Okay, well what are you seeing? You know, why is this a concern all of a sudden? You know, why do you*

	care?" Because my first go-to is to panic that, 'Oh my gosh, something's, you know, something's back and you saw something on the mammogram.' So I'd prefer not to be freaked out in the process." - Participant who did not receive the genetic test
Repetition and timing	*"And then this year...I can't remember what was going on. I think my husband had a…surgery and my son, his [school] team was competing. I was like, I don't have time to do this, so I just didn't do it. I figured I'll do it next year. But yeah...it takes a really, really long time to do those forms."* - Participant who did not complete the screener *"I found that…a lot of the stuff I just didn't wanna know about, you know. I was just pretty focused on getting rid of the cancer and jumping through the hoops that the doctor wanted me to do. So all the other stuff, it's like, 'Okay, you just do your thing and-and I'm just gonna try not to die during chemo.'"* - Participant who did not receive the genetic test initially

DISCUSSION

Population-based genetic screening and testing programs can help identify patients at an increased risk of cancer and direct them toward enhanced cancer screening services (e.g., mammograms, colonoscopies, ultrasounds, magnetic resonance imaging etc.) or other recommendations. However, for these programs to be successful, patients must be effectively engaged at each step of the program pathway. Although study participants had varying degrees of engagement in the CARE program pathway, four themes emerged highlighting barriers and facilitators to participation that cut across the different groups: the benefits around clarity of communication, the need for increased information and knowledge, the importance of a trusted provider, and the effects of repetition and timing.

The themes we identified largely indicate that the reasons participants have for not engaging in testing are potentially mutable, and that patients are willing to engage in genetic screening and testing when they have a clear understanding of the specific purpose and procedures at each point in the pathway. Similar to prior research, our study found that patients are open to hearing about their genetic risks [11] but are most likely to support testing when there are clear medical benefits, and when testing is for specific diagnostic and predictive purposes [12,20,21]. These findings reiterate that a genetic risk assessment program, such as CARE, needs to clearly articulate to patients the diagnostic and predictive purposes of genetic screening and testing and how the testing would impact their future healthcare. Programs could consider opportunities to integrate patient communication and education during appointments, such as when mammogram patients are waiting to be seen.

Interestingly, while a limited number of individuals suggested that "not wanting to know" factored into their thoughts around receiving a genetic test, the fear of emotional and psychosocial effects from genetic testing results that have been cited in other literature [20,22,23] was not frequently identified by participants in this study. In fact, most participants felt that "knowledge was power" and having more information could help in their care. These findings may suggest a growing acceptance of genetic testing, with knowledge and provider relationships as the main factors affecting engagement.

Other barriers to genetic testing identified by patients in the study included effects on insurance and potential costs, which aligns with findings from previous studies [1,13,24]. Interviewees also expressed being frustrated by redundant requests for information that did not feel relevant to a given appointment or were not addressed by their healthcare provider during appointments. This highlights a need for improved portability and visibility of patient-provided history, particularly for patients who return year over year to the same center or see multiple providers within the same network. Moreover, patients at each stage in the pathway described their own limited capacity, in terms of time and attention, to prioritize, navigate, and complete each request and recommendation offered by the healthcare system.

Communication from a trusted provider helped counteract barriers to genetic testing and was identified as a key facilitator to moving forward in the process for many participants. Previous research has also underscored the importance of hearing the recommendation for genetic testing from a trusted provider, especially among racially and ethnically diverse patients [25–27]. In addition, patient navigators have been shown to improve patient follow-through on genetic testing [28]. Despite patients' desire to hear this information from a provider, few studies have evaluated the impact of educational training programs designed to improve health care providers' communication skills related to genetic testing and cancer risk on the uptake of genetic testing [29,30]. This highlights the need for additional research to identify strategies to effective support non-genetic clinicians in the provision of this type of patient counseling. Our findings also suggest that patients who opt to get genetic testing do not always choose to talk to a genetic counselor prior to testing; patient navigators may also be able to provide information about talking to a genetic counselor before getting testing, and helping a patient determine if that is the best option for them.

The findings highlight the importance of supporting the coordination of health care team around genetic testing and risk assessment. Improved coordination of the health care team may help ensure that multiple providers address the recommendation with patients at multiple time points and increase the feeling of importance around the test. Additionally, expanding the languages the screener is available in is an important way to support equitable access to testing, especially in linguistically diverse regions like Southern California.

Limitations

As mentioned before, self-selection bias was a limitation in this study; participants who were completely opposed to genetic testing or had the largest concerns around privacy issues or distrust of the healthcare system may not have chosen to participate in the qualitative interviews. This limitation was mitigated by performing outreach in three separate categories – including those who did not fill out the family history screening and those who did not receive a test after a recommendation – which ensured that the study included patients that stopped engaging early in the pathway. Additionally, these participants likely reflect to some extent a patient population that would be willing to consider screening and testing, if aspects of the program were improved; this study gives insight into what those considerations and changes might be.

Another limitation was in identifying participants from each of the three groups. The CARE patient portal was used for outreach; however, this portal was designed to support patient care rather than research purposes, and patient experiences and history with genetic testing was not always included. For example, of the seven interviews conducted with participants categorized in the program data as Group

2 (i.e., did not receive genetic screening after the recommendation), only four had never actually received a genetic test for cancer risk in their lifetime. The other three had received genetic testing for cancer risk years prior to the CARE recommendation, received the genetic test in another country, or received the test after the CARE recommendation when they were referred again by their primary care provider. We addressed this limitation by adjusting interview guides based on a participant's actual experiences, rather than our initial categorization.

There are also limitations around the number of participants that were interviewed for the study, which limits the generalizability of the findings. In addition, participants largely identified as white, and with English as a preferred language; the program was implemented in a number of Southern California clinics, which also limits the geographic scope and relevance of the study. Participants also overwhelming identified as women, with an average age of 59 years. While this expected, since the program was implemented in a mammogram setting, it affects how much the findings can be generalized to other genders and age groups. Additionally, the study only included participants who were scheduled to receive or recently received a mammogram. Individuals who do not receive this kind of screening, including those that are generally not engaged with the health care system, are not represented in our findings. This population would likely face different barriers to receiving genetic screening or testing recommendations. Future research could focus on the equity considerations of engaging missing populations in these genetic risk programs.

CONCLUSION

Interviews with participants at multiple stages of a genetic screening and testing program pathway suggest that patients face similar barriers at each step in the program, as they try to understand and prioritize the relative importance of screening and testing for their personal health and healthcare. Given the growth of genetic risk assessment in outpatient care settings, the results of study provide helpful insights to improve patient engagement with risk assessment and uptake of genetic testing. Patient education around the benefits of the program to patients and their families, as well as clear communication about what testing entails may help patients engage with the program. Strategies aimed at increasing coordination among a patient's healthcare team can help present information to patients in multiple ways, from multiple providers, to help ensure that recommendations come from trusted sources and support the uptake of genetic testing.

Data Availability

The data generated and analyzed in this study were derived from in-depth interviews and are not publicly available due to restrictions, namely the contain information that could compromise the privacy of research participants.

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Competing Interests

The authors have no competing interests to declare that are relevant to the content of this article.

Ethics Statement

The protocol for this study was approved by the Providence Institutional Review Board (IRB # 2023000128). Participants provided verbal informed consent prior to enrollment in the study.

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Authors' Contributions

The author contributions were as follows: All authors (CD, KE, KC, SB, OG, SR) contributed to the study conception and design. Material preparation, data collection and analysis were performed by CD and SR. KE, KC, SB, and OG aided in the interpretation of results and implications for practice. The first draft of the manuscript was written by CD and SR and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript. SR is responsible for the overall content as guarantor.

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Figure Legend:

Figure 1 – CARE Program workflow

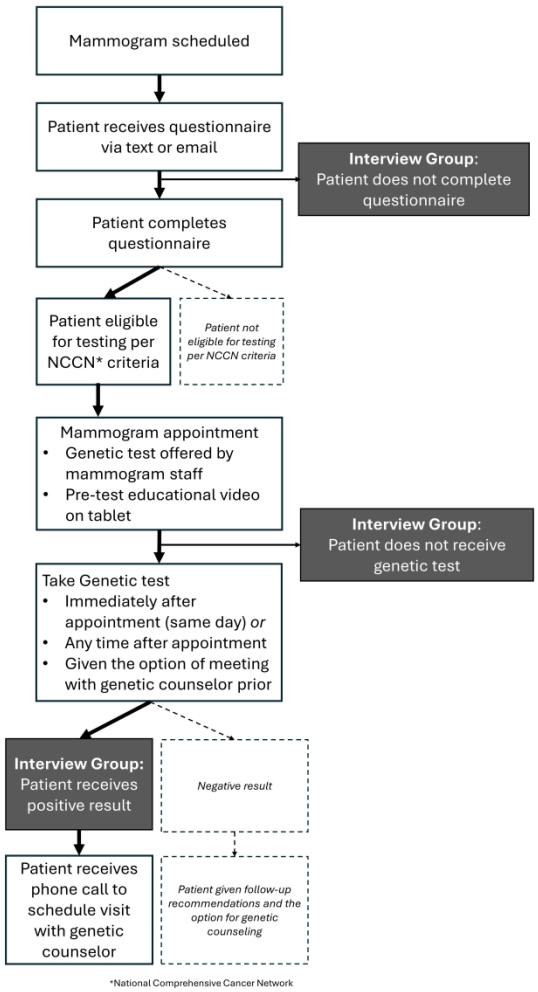

Figure 1. CARE Program workflow

254x508mm (300 x 300 DPI)

Supplemental Table 1: Semi-structured Interview Guides

Group 1: Received questionnaire, didn't complete

- Can you describe your experience getting ready for your mammogram through Providence?
  - *What do you remember around: scheduling, reminders, staff, communication with PCP, etc.*

- Part of getting a mammogram is about understanding your cancer risk. How important is it to you to understand your cancer risk?
  - *Do you feel like you are at particularly high risk of cancer?*

- What do you remember about receiving an email or text to complete a questionnaire prior to your mammogram?
  - *Can you describe your experience with receiving the questionnaire?*
    - *Do you recall if you opened or started the questionnaire? If so, why did you stop?*
    - *Do you recall why you chose not to fill it out ahead of the appointment? If so, can you explain why?*
    - *Do you recall the front desk staff asking you about completing the family history questionnaire? If so, can you describe experience?*

- What sort of things make it difficult to fill out healthcare questionnaires received via text or email?
  - *To what extent do you feel you have sufficient time during the day to fill out the forms?*
  - *To what extent does the information requested seem important enough to fill out?*
  - *How easy or difficult are the family history questions easy for you to fill out?*
  - *To what extent is the language on the forms a barrier to completing the questionnaires?*

- What's your preferred method of receiving health or family history questionnaires prior to an appointment?
  - *Are you comfortable with receiving them as a text? Email?*
    Would you prefer someone ask you the questions directly during your appointment?

- What would make it easier for you to fill out those kinds of questionnaires?
  - *Examples: right before the appointment, same day as the appointment, during the appointment*
  - *Would reminders to fill out the form be helpful?*

- How important does it feel to share information on your family medical history with your healthcare providers?
  - *How do you expect the information will be used?*

- In general, what concerns do you have, if any, around sharing information like family and personal medical history of cancer with your healthcare provider prior to your healthcare appointments?

- *Examples: Mistrust of the healthcare system, don't see the importance of sharing the information, overly personal information, not sure about family history.*
- *Are you more comfortable sharing this information with your primary care doctor versus the provider giving you a mammogram, with the Providence health system, etc.*
- *What would make it easier for you to share your personal health information like family history with your healthcare providers?*

- [*For those that recall receiving the questionnaires*] Were you aware that the questionnaire would be used to determine if a genetic test would be recommended to help assess cancer risk as part of your healthcare?

- To what extent would knowing that the answers to the questionnaire were used as part of a recommendation for genetic testing have made it seem more important?
  - *Would you be more likely to answer it?*
  - *Why or why not?*
  - *How important is it to you to know if you have a genetic predisposition for cancer?*

- If your healthcare provider recommended that you have a genetic test, what would your reaction be?
  - *Would you be willing to do so? Why or why not?*
  - *What concerns would you have?*
  - *Have you ever received genetic testing through someone else?*
    - *What made you decide to do the genetic testing?*

- Is there anything I didn't ask about around understanding your cancer risk, genetic testing, sharing family history information that you'd like to mention?

Group 2: Complete questionnaire, didn't complete testing

- Can you describe your experience with receiving the recommendation for genetic testing?
  - *Who told you about your results?*
  - *How did you feel?*
  - *What questions did you have?*
  - *What were you told were the next steps?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *How important did it seem to your health care for you to take those next steps?*
  - *Are there other ways that would have been more useful to receive the testing recommendation? (i.e., in person, multiple times)*

- How important is it to you to understand your cancer risk?
  - *Do you feel like you are at particularly high risk of cancer?*
    How important is it to you to know if you have a genetic predisposition for cancer?

- What concerns (if any) did you have around genetic testing?
  - *How concerned were you about the results you might receive?*
  - *How worried were you that results might affect your health insurance?*
  - *How worried are you that results might affect your employment?*
  - *How much do you trust the healthcare system when it comes to genetic testing?*

- Did you discuss any of your concerns with a healthcare provider?
  - *If yes, how did that conversation go?*
    - *What kinds of questions or concerns did you raise with your provider?*
    - *To what extent did you provider answer your questions?*
    - *To what extent did your provider address your concerns?*
    - *Were you able to receive the information that you needed from your provider? If not, why not?*
    - *Is there anything else the provider could have done to address your concerns?*
  - *If not, what would have made it easier to talk to a healthcare provider about your concerns?*

- Do you feel like you had enough information in deciding whether or not to get a genetic test?
  - *Is there anything that your healthcare provider could have communicated that would have encouraged you to get the test?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *What other information would have been helpful to have?*
  - *Did you get information or advice from someone other than a healthcare provider or in some other way?*

- What steps, if any, did you take toward getting a genetic test after receiving the recommendation?
- What things did you experience that made getting the test difficult?
  - *To what extent did you have any concerns about the cost of the test?*
  - *To what extent did you feel you had enough time to get the test?*

- *To what extent was getting transportation or getting to and from the test a barrier?*
- *What difficulties, if any, did you around scheduling a test?*
- *Were there language barriers that made receiving the recommendation or getting the test difficult? Please explain.*
- *Was there anything else that made getting the test difficult?*

- Overall, what was the primary reason you chose not to get a test?

- What changes could your provider or insurer make that would encourage you to get a genetic test?
  - *Examples: financial assistance, additional information, etc.*

- Have you ever received genetic testing another way, other than this program?
  - *Did you receive this testing prior to getting the recommendation from this program?*
    - *If yes, did you share the results of that test with your provider?*
      - *How did that conversation go?*
    - *If not, what encouraged you to get the other test?*
      - *Was there anything that made that test easier to get?*

- Other than the genetic testing, do you remember getting a recommendation for additional screening based on your family history?
  - *What was your experience receiving the recommendation?*
  - *Did your primary care doctor follow up about the recommendations with you?*
    - *If yes, please explain.*
    - *If not, to what extent would that have been helpful?*
  - *Were the recommendations for additional screening communicated clearly?*
  - *What has your experience been with getting additional screening?*
    - *Did you experience any barriers in receiving follow-up care?*
    - *How are you using the information?*

- Is there anything I didn't ask about around understanding your cancer risk, genetic testing, sharing family history information that you'd like to mention?

Group 3: Completed testing

- Can you describe your experience with receiving the recommendation for genetic testing?
  - *Who told you about your results?*
  - *How did you feel?*
  - *What questions did you have?*
  - *What were you told were the next steps?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *How important did it seem to your health care for you to take those next steps?*
  - *Are there other ways that would have been more useful to receive the testing recommendation? (i.e., in person, multiple times)*

- How important is it to you to understand your cancer risk?
  - *Do you feel like you are at particularly high risk of cancer?*

How important is it to you to know if you have a genetic predisposition for cancer?

- What concerns (if any) did you have around genetic testing?
  - *How concerned were you about the results you might receive?*
  - *How worried were you that results might affect your health insurance?*
  - *How worried are you that results might affect your employment?*
  - *How much do you trust the healthcare system when it comes to genetic testing?*

- Did you discuss any of your concerns with a healthcare provider?
  - *If yes, how did that conversation go?*
    - *What kinds of questions or concerns did you raise with your provider?*
    - *To what extent did you provider answer your questions?*
    - *To what extent did your provider address your concerns?*
    - *Were you able to receive the information that you needed from your provider? If not, why not?*
    - *Is there anything else the provider could have done to address your concerns?*
  - *If not, what would have made it easier to talk to a healthcare provider about your concerns?*

- Do you feel like you had enough information in deciding whether or not to get a genetic test?
  - *What other information would have been helpful to have?*
  - *Is there anything that your healthcare provider could have communicated that would have encouraged you to get the test?*
  - *Was the information about the test communicated in a way that was easy for you to understand?*
  - *Did you get information or advice from someone other than a healthcare provider or in some other way?*
  - *What did you find the most helpful to know in making the decision to get the test?*

- What was the primary reason you decided to get testing?
- Can you describe your experience with getting the test itself?
  - *Was there anything that made that experience difficult?*
  - *Is there anything that could have changed to make the process easier?*

- Were there any factors that helped you get the test?
  - *Was the full cost of the test covered by insurance?*
  - *What difficulties, if any, did you have scheduling the test?*
  - *Did you have any assistance getting to and from the testing center?*
  - *Did anything else make getting the test easier?*
  - *After getting the test, was there anything else that you would have wanted to know beforehand? Please explain.*

- Can you describe your experience receiving the results of the test?
  - *Who gave you the results of your test?*
  - *What kind of information were you given when you received your results?*
    - *Was the information you received sufficient? Why or why not?*

- *What, if anything, would you have changed about the process or experience of receiving the results?*

- What was your experience receiving any follow-up recommendation based on your test results?
  - *What was your experience receiving the recommendation?*
  - *Did your primary care doctor follow-up about the recommendations with you?*
    - *If yes, please explain.*
    - *If not, to what extent would that have been helpful?*
  - *Were the recommendations for any additional screening or treatment communicated clearly?*

- What has your experience been with getting any additional screening or care?
  - *Did you experience any barriers in receiving follow-up care?*
  - *How are you using the information?*

- How has your knowledge or awareness of your genetic results changed your care?
  - *Has it prompted any other changes?*

- Is there anything I didn't ask about around understanding your cancer risk, genetic testing, or follow-up care that you'd like to mention?
