Previvors' Perspectives on Population-Based Genetic Screening in the Primary Care Setting

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A thesis
submitted in partial fulfillment of the
requirements for the degree of

Master of Science

University of Washington
2023

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Program Authorized to Offer Degree:
Public Health Genetics

University of Washington Abstract

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Advances in next-generation sequencing have led to the utilization of multigene cancer panel testing for assessing inherent cancer risk. Multigene cancer panels could be the key to unlocking multiple clinical results at once but may also open the door to potential uncertainties. The EDGE Study is a clinical implementation study that facilitated genetic testing using a population-based screening tool in the primary care setting. EDGE offered hereditary cancer genetic testing using a panel of 29 different genes, associated with hereditary risk for eight different cancers. Genetic variants ranged from low-, moderate-, and high-penetrance. Semi-structured interviews were conducted with previvors (those with a positive pathogenic test result) to evaluate experiences following receiving results from the genetic test. Thematic analysis was utilized to identify key themes and findings. We discovered that communication with providers and family was critical in understanding and processing results. Older previvors and those with low- and moderate-penetrance variants explained low utility. This study offers important insights into the implications of multigene cancer panel testing on genes of varying penetrance.

Introduction

Cancer is the second leading cause of death in the US[1]. Genetic testing for hereditary cancer can improve health outcomes through individualized recommendations for cancer prevention and early detection. By providing patients with information about their risk, individuals can make informed decisions about their health and future healthcare plans[2]. Genetic testing for hereditary cancer was first introduced for breast cancer patients, specifically testing for *BRCA1* and *BRCA2* (*BRCA1/2*) variants[3]. Understanding, if there is a hereditary link to a breast cancer diagnosis, enables family members to take preventative action: whether that be in the form of preventative surgery or increasing screening for early detection. Genetic testing has also proven helpful for individuals who have not received a cancer diagnosis but may be aware that cancer runs in their family[4].

The term previvor, coined by Sue Friedman, has been used to describe a person with a predisposition to cancer who has not yet been affected by the disease[2]. This term applies to those who have tested positive for a pathogenic variant associated with cancer. Previvors are often faced with difficult decisions about how to manage their cancer risk after receiving genetic test results[5-8]. Previous research on the experiences of previvors has been limited primarily to females with *BRCA1/2*-associated variants. Angelina Jolie's 2013 editorial disclosing her decision to undergo a preventative double mastectomy after finding out she carried a pathogenic *BRCA1* variant led to heightened awareness around BRCA1/2 variants[9,10]. *BRCA1/2* variants have been studied extensively and the knowledge surrounding these high-penetrance genes has grown significantly over the years. Investigators have worked to find answers related to risk-reducing surgeries[9], communication with families[11], and barriers to access[6]. Current testing panels, however, routinely include many cancer susceptibility genes in addition to *BRCA1* and *BRCA2*.

Advances in next-generation sequencing have led to the utilization of multigene cancer panel testing for assessing inherent cancer risk by genetic specialists and clinicians[12-16]. More widespread and comprehensive testing means there will be an increase in the detection of pathogenic variants as well as variant of uncertain significance (VUS). Cancer susceptibility genes in clinical panels include well-known high-penetrance genes such as *BRCA1/2* in addition to genes that are classified as low- or of moderate-penetrance[17]. There has been increased testing and knowledge around moderate-penetrance breast and ovarian cancer genes since the advent of multigene cancer panels[18-21]. Little is known about the psychological impact associated with receiving results for a pathogenic variant in a moderate-penetrance gene [13]. Some quantitative research has suggested that identification of pathogenic variants in moderate-penetrance genes led to patient confusion, uncertainty, and distress[14, 18, 21].

Genetic testing using multigene cancer panels can unlock multiple clinical results at once but may also open the door to potential harms such as overtreatment with risk-reducing surgeries[19,22]. Current guidelines suggest annual screenings and shared decision-making for any risk-reducing surgeries for pathogenic variants in moderate-penetrance genes[23,24]. However, clinical guidelines and risk estimates are constantly changing and expanding[13]. Uncertainty about appropriate recommendations for managing risk remains one of the largest concerns[25, 26]. To adequately address the implications of multigene cancer panel testing we need to know more about how patients manage and navigate pathogenic results for genes across all penetrance levels. More information is needed to understand the process previvors go through after receiving a positive result over the range of penetrance for different hereditary cancer genes in order to identify specific supports that are needed.

The Early Detection of GEnetic Risk (EDGE) Study is a clinical implementation study that facilitated genetic testing after identifying eligible individuals using a population-based screening tool in the primary care setting. The EDGE Study offered hereditary cancer genetic testing that consisted of a panel of 29 different genes, associated with eight different cancer types: breast, colon, skin, ovarian, pancreatic, prostate, stomach, and uterine cancers. Semi-structured interviews were conducted with a subset of individuals who completed the genetic testing and came back positive for a pathogenic variant to evaluate experiences immediately following genetic testing and receiving results. This project aims to evaluate reactions to a pathogenic variant including how they feel, who they take to talk to, what actions they take, and how they utilize their results moving forward. Learning more about how individuals process and utilize genetic information on variants of varying penetrance will help inform how we can better implement hereditary cancer genetic testing on a broader scale. Additionally, understanding how people feel, who they talk to, and what choices they make can help us better understand the type of support previvors need when facing difficult decisions regarding managing their cancer risk.

Project Aims

The overarching goal of this project is to assess how previvors of pathogenic variants of genes of varying penetrance, identified using a population-based screening tool in the primary care setting, react to receiving positive genetic test results. This includes characterizing how people feel, who they talk to, what actions they take, and how they utilize genetic information moving forward.

ERC/IRB approval(s)

The University of Washington IRB approved this research project on 4/2/2020 (IRB ID: STUDY00009476).

Methods

Study Design

We conducted a qualitative study with individuals who tested positive for a genetic variant associated with hereditary cancer risk. Thematic analysis was utilized to identify key themes and findings from the semi-structured interviews. A combination of inductive and deductive approaches were used in the design and analysis of the interviews.

Study Population

The study population consisted of patients over the age of 25, that were comfortable with reading/writing in English, registered at one of 12 primary care clinics, and had an annual appointment during the study's recruitment period (Point of Care (POC) arm: April 2021-April/May/June 2022 and Direct Patient Engagement (DPE) arm: September 2021 December 2022). The 12 primary care clinics can be broken into two larger Federally Qualified Health centers (FQHCs). MultiCare Health System based out of Washington had six participating clinics, all classified as urban. Billings Health Care System is based out of Montana and Wyoming with three participating clinics classified as urban and another three clinics that are classified as rural. All participants included in this analysis completed hereditary cancer genetic testing through the study and received a positive test result for a pathogenic variant between November 2021-September 2022.

Hereditary Cancer Test

The genetic testing was done through Color Health using sequencing of 29 genes associated with eight cancers. The panel consisted of the following genes: *BRCA1, BRCA2, MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH, MITF, BAP1, CDKN2A, CDK4, TP53, PTEN, STK11, CDH1, BMPR1A, SMAD4, GREM1, POLD1, POLE, PALB2, CHEK2, ATM, BARD1, BRIP1, RAD51C, RAD51D.* Associated cancers included: breast, colon, skin, ovarian, pancreatic, prostate, stomach, and uterine cancers[27].

Analysis Team

All follow-up interviews were conducted by the first author, Faith Beers. Faith identifies as female and uses she/her pronouns. She is a master's student in Genetic Epidemiology at the University of Washington. She had been working on the larger EDGE project as a graduate research assistant since November 2021 and was involved with patient outreach, preparation of patient reports, data management, and design of smaller projects within the larger study. Faith was trained on qualitative research methods and interviewing techniques by Sue Trinidad prior to conducting any interviews. Faith completed interview outreach for all previvor interviews and became acquainted with interviewees through a series of outreach attempts and two interviews in the span of one year. Little information was shared with interviewees about Faith aside from her degree, position, and interest in the overall study. Faith also served as the first coder on the follow-up transcripts.

Sue Trinidad was a Ph.D. candidate who later became an assistant professor at the University of Washington. Sue identifies as female and uses she/her pronouns. As a senior researcher, Sue served as a qualitative research expert for the project. She consulted the research team in the study design, methods, and analysis of all qualitative projects.

Heather Harris served as the second coder on the follow-up interviews. Heather identifies as female and uses she/her pronouns. Heather is the data manager for the larger EDGE project.

Data Collection

Participant Recruitment

Participants were approached through their healthcare system via email, phone, or in person at their appointment (depending on the clinic and study arm). Those who completed the family history screener and were identified as eligible for free genetic testing through the study were then contacted for genetic testing by the study's research assistants. All participants who were eligible for testing and had a working phone number were called at least once. If the participant did not answer a voicemail was left. For participants that did not have a working phone number or those that the research assistants were unable to leave voicemails for, an email was sent. Those who moved forward with the testing and received a positive test for a pathogenic variant between December 2021- September 2022 (86 participants) were contacted for a baseline interview. Those who completed a baseline interview (56 participants) were contacted again six-nine months after to complete a follow-up interview (32 participants). All interview outreach was conducted by the lead author. Interview outreach consisted of two phone calls (typically one week to one month apart) and one email before the participant was deemed "unable to contact".

Data Sources and Collection Processes

Semi-structured interviews were conducted to collect information about the participant's thoughts and experiences following genetic testing in the primary care setting. The interview guide was developed by the research team and was later reviewed by the study's PI, investigators, and qualitative research expert. The follow-up guide included repeat questions from the baseline guide as well as new questions that focused on responses following the receipt of their test result. The follow-up guide contained questions probing at conversations participants had with their providers, family members, genetic counselors, and any other relevant parties. Additionally, the guide contained questions related to participants' feelings and reactions to their results, views on their overall health, changes in their behaviors and lifestyle, and concerns about their cancer risk. Closed-ended questions were also included to collect quantitative data about the decision participants made to enroll in the study, how they felt about their provider, the amount of time they spent on follow-up activities, and the frequency of their thoughts related to cancer risk. The follow-up guide was piloted by the lead author and the same study participant that piloted the baseline interview. Following the pilot, revisions to the guide were made and the final interview guide was completed.

All interviews were conducted in English via Zoom. For those that were unable to log in and join the meeting with the Zoom app or website, a dial-in option was available. All interviews were recorded with audio and video, except for those who dialed in and/or preferred no video. Interviews ranged from 22 minutes to one hour and 10 minutes, averaging about 43 mins per interview. Recordings were saved to the research team's shared drive. Field notes highlighting the main takeaways and summary of each interview were completed and saved to the drive immediately following each interview. As needed, the project manager uploaded the interview recordings to REV for transcription services, and transcripts were saved in the shared drive. Transcripts were deidentified and cleaned before being uploaded to the ATLAS.ti (the qualitative coding software program used).

Data Logistics and Storage

Project data is stored in Redcap, the Color Health website, and the research team's shared drive. Most of the data from Redcap and Color can additionally be found in reports on the shared drive. All qualitative data is saved on the shared drive. Original and cleaned transcripts are saved in the same project under separate folders. Two study ID forms were utilized: the patient's Redcap ID, assigned when the participant completes the initial screener, and the patient's requisition number (req#), assigned when a genetic test is ordered for the patient.

Data Analysis

Overall Plan

Thematic analysis was utilized to identify key themes and findings from 16 semi-structured interviews conducted with patients who tested positive for a pathogenic variant associated with hereditary cancer (previvors). Only the first 16 interviews were used for analysis to meet deadline requirements while interviews were ongoing. The additional interviews will be coded and integrated into a future analysis

Coding Description Overview

All transcripts were coded by two study team members. Both coders met the inclusion criteria for participation in the study, however, neither were patients at either of the participating clinics (MultiCare and Billings Clinic). Neither had genetic testing for hereditary cancer and were therefore unaware of their hereditary cancer gene status. The timeline for coding was January-March 2023. Codebook development began in December of 2022. Transcripts were coded between January-March 2023.

Codebook development began with deductively creating codes based on the interview guide to highlight the key sections of the interview. From there, a set of transcripts were used to test the existing codes and inductively identify new codes that came out of the transcripts. The codebook was reviewed by the study team's qualitative research expert. The next step in codebook development was three rounds of coding and reconciliation between the two coders. The codebook was updated as needed to add codes that were missed, split codes to add granularity and merge similar codes that were being used interchangeably. The final codebook consisted of 43 codes and eight code groups (appendix).

The two coders coded the first two transcripts independently and then met for a reconciliation meeting. Inter-coder agreement (ICA) was run to pinpoint discernible differences in how codes were being applied. From there the two coders discussed how they were applying the codes, what worked well, what did not work well, and so on. Updates to the codebook were made based on the result of their conversations. Additionally, the two coders reconciled the two transcripts by going through each of them together, discussing the quotation sizes and codes applied, and coming to a consensus on a single coding for each transcript. Consensus coding consisted of keeping only one quotation and set of codes if the two had been coded the exact same or discussing the discrepancies and updating the coding with the consensus that resulted from the discussion. This process was repeated a total of three times so that all six transcripts were coded by both coders independently and then reconciled through discussion and consensus.

To aid in the reconciliation process, there was a code called “for discussion” that the coders would add to any quotation they wanted to highlight and discuss in the reconciliation meeting. Additionally, both coders kept methodical memos of each round of coding to detail their process through independent coding and reconciliation.

Late-stage Analysis/Interpretation

The process of identifying themes first started with recognizing patterns. Coders kept thematic memos of each transcript to record key information coming out of the data such as recurring and novel views/ideas. Recurring responses/behaviors were later identified and grouped as patterns. Within-category patterns were considered to identify if patients had similar experiences when it came to receiving results, talking with providers, genetic counselors, and family members, and choosing to participate in follow-up care. Between category, patterns were assessed to evaluate if having similar experiences lead to similar outcomes, for example, if people who had positive conversations with their provider and family members did, they also pursue follow-up care related to their test results. Novel ideas were noted as they often represented contradicting or extreme views. We also considered values and rationale influencing the responses/behaviors and further identified patterns from there. For example, those with a pathogenic variant in a high-penetrance gene variant may respond differently than those with a pathogenic variant in a low-penetrance gene or those who have had a personal history of cancer may respond differently than those who have not experienced cancer. Taking all the above into consideration, we worked to pull out major and minor themes. The study’s qualitative research expert participated in the early stages of pattern recognition and later stages of theme identification. Additionally, the qualitative research expert reviewed all themes that were found at the end of the analysis. Participants did not provide feedback on findings/results. ATLAS.ti was utilized to code interviews, generate query reports and occurrence tables, and visually display data that were used in the analysis.

Results - Population

Participant Demographics

56 participants were contacted for a follow-up interview, 32 of which completed the interview (Table 2). The first 16 interviews were analyzed. Nine of the participants were female, and seven were male (Table 3). Of the participants included in analysis, 37.5% were over age 70, 37.5% were between the ages of 46-70 and 25% were between the ages of 25-45 (Table 3). All participants belonged to either MultiCare Health System based out of Washington (56%), or Billings Clinic based out of Montana and Wyoming (44%) (Table 3). All participants identified as white non-Hispanic noting there was missing data on race/ethnicity for four participants. Six (37.5%) of the participants had a personal history of cancer and two received a cancer diagnosis following genetic testing. Genes were classified by penetrance (Table 4). Actionability of the genes was also accounted for twelve (75%) participants tested positive for an actionable gene and four (25%) participants tested positive for a non-actionable gene. Three participants had participated in genetic testing prior to participating in the EDGE Study.

Table 2. Outreach Table

	Contacted	**Interviewed**	**Refused**	**Lost to follow-up**
Baseline	86	56	14	12
Follow Up	56	32	4	20

Table 3. Demographic Table (N=16)

Characteristic		**N (%)**
Gender	Female	9 (56%)
	Male	7 (44%)
Age	25-45	4 (25%)
	46-70	6 (37.5%)
	70+	6 (37.5%)
Race/Ethnicity	White, non-Hispanic	12 (75%)
	Missing	4 (25%)
Clinic	Billings Clinic	7 (44%)
	MultiCare	9 (56%)
Personal History of Cancer	Prior to EDGE	6 (37.5%)
	Following EDGE	2 (12.5%)
Prior Gx Testing	Completed	3 (19%)

Table 4. Genetic Variant Demographics

Gene	**Penetrance**	**N = 16**
PMS2	High	2
MSH6	High	1
APC	High	2
BRCA1	High	1
BRCA2	High	2
CHEK2	Moderate	4
ATM	Moderate	1
*MUTYH**	Low	4

*Non-actionable variant as cancer risk only increased for individuals with biallelic pathogenic variants

Results – Thematic Description

General Overview

All major and minor themes were organized into one of two categories: emotional or behavioral response. Themes categorized as emotional responses referred to how participants reacted and processed receiving their results. Themes categorized as behavioral responses referred to what participants actually did with the information. Overall, the two categories helped explain how participants handle receiving hereditary cancer genetic test results for genes with varying penetrance and how they go on to use those results in their conversations with family and medical providers. Identified themes were irrespective of gene penetrance and spoke to commonalities found between participants with results of varying penetrance. We found that people are not overly worried about their positive genetic variation/increased risk for hereditary cancer. People are interested in having the information so that they have the knowledge to take preventative action and are not typically overwhelmed by the results or what they mean. Generally, people did not feel as though their increased risk was something that warranted any drastic changes. People instead talked about having a greater awareness of their overall health and being proactive when it came to routine testing/screening for hereditary cancers and living healthy lifestyles all around. Higher importance was often placed on other things over one’s own personal risk for hereditary cancer. For example, other health problems that people were actively dealing with were described as the priority while the genetic test result was less relevant. Additionally, many people expressed concern for their family members' risk. Many felt as though younger family members would benefit from the information more and worried about future generations passing the pathogenic/likely pathogenic variant down. We also found that the results did not come as a surprise to most previvors as they are familiar with their family history and have considered themselves at higher risk for hereditary cancers based on the fact that they have had and often witnessed family members battle cancer.

Key Themes/Domains

Emotional

1. Receiving a positive test result for a genetic variant associated with hereditary cancers often does not come as a surprise.

The majority of participants stated that they were not surprised by their positive result, and many said they were expecting a positive result based on the fact that they have a family history of cancer. One participant that had a personal history of cancer (PH) explained "*Well, I wasn't surprised because we have such a family history, although only one niece had also had this variant of those that were tested. So basically, I wasn't surprised.*" (F, 81, PH, *ATM*). Those who had lost relatives to cancer seemed to identify themselves more closely as people who were at higher risk for hereditary cancer prior to receiving the results and therefore expected the results to confirm what they had already been thinking. One participant stated *"I was expecting that the result that I had, the susceptibility, because of the fact that both that my father had colon cancer, and my brother also had died of different type of cancer. The susceptibility is there, or you believe the susceptibility is there."* (M, 71, *MUTYH*).

1.1 Aware of family history, not the first time they are made aware of their potential risk for hereditary cancer

Within the major theme of being "unsurprised", it was found that most individuals had already internalized a higher propensity for hereditary cancer based on their family history alone.

Participants with a family history consider themselves at higher risk from cancer so they are not surprised by results when it is confirmed to run in their genetics. When asked how they felt about their results one participant responded with *"I must say I just was kind of, "Oh, okay." Because what do you say you? You've seen it in your mom and your dad, so.*"(F, 69, PH, *PMS2*). This could speak to the value that people place on family history as a tool for evaluating risk.

1.2 Though not surprised there is still a negative impact

A sub-theme that emerged highlighted that despite the lack of surprise, getting a positive result still evoked thoughts and feelings that were more negative. One participant explained "*I was not really surprised, because I knew I had a susceptibility, because of family history. But, it's one of those things that when you see it in writing, it makes it look a little different. It was scary, but expected."* (M, 71, *MUTYH).* They also explained their view of getting the results by saying "*It's one of those things that you know is there, but you don't think about every day. It was like, "Hey, yeah, thanks for reminding me." It would be the same as you have something like you're overweight, and you know it. Then somebody says, "Boy, you look like you gained weight." It's a reminder of something you already know.*" (M, 71, *MUTYH*). These quotes outline the negative thoughts that are associated with receiving this kind of information, indicating that there is a potential negative impact even when the results may be expected.

1.3 Relief to know there is a genetic component

For some participants, there was a sense of relief when they found out they tested positive for a pathogenic variant associated with hereditary cancer. This subtheme was found among participants with both a personal and family history of cancer. In these cases, the positive genetic test result gave them an explanation for their personal and family history of cancer. One participant said that they were *"Somewhat relieved to know that there is a genetic component to it, because that's what I've always felt…"* after receiving the results. The participant further explained that in the past they had seen specialists who told them there was not a genetic component to their cancer and went on to say *"So actually a relief to know that it's there and maybe certain things make it come up. Relief.*" (F, 71, PH, *MUTYH*).

1.4 Results were surprising when the results did not match expectations

A contradicting sub-theme arose in individuals who received a result that was different from what they were expecting. Specifically, participants found the results surprising when they differed from the expected results. Some participants expected to have a result associated with a particular cancer that they had a family history of but got results for something else. For example, when asked how they felt about the result one participant said "*Actually surprised, simply because I think it was a small chance of colon cancer and I think I've never had a family member have issues with that, at least not that to my knowledge. I was actually kind of surprised about it.*" (Female, 31, *MUTYH).* In some cases, the participants were not expecting a positive result of any kind. Either because they had been told in the past that there was no genetic link to their cancer "*I had two nieces who both had cancer several times and one passed away. They had the testing back some years ago and they were told it wasn't genetic, it was environmental. I suppose I was a bit surprised because of their results, but I know things have progressed, like medicine, everything, where they know more now"* (F, 81, PH, *ATM*) or because they have remained cancer-free their entire lives. When asked about their feelings one participant said "*I was shocked. Although I have a history of my mother having had breast cancer, I just wasn't* *suspecting that I would be at a high risk as well. I had not avoided doctors. I had plenty of medical care and I had routine screenings like any 70-year-old plus person should have, and I thought I was really up-to-date. This really took me by surprise.*" (M, 74, PH, *BRCA2, APC*).

2. Initial fear fades as more information becomes available

Another major theme that came out of this analysis was that previvors become less stressed, scared, and overwhelmed as they get more information, support, and answers.

2.1 Talking with the provider and creating a plan

Talking with a provider, confidence in their provider, and provider support played the biggest roles in making patients feel more at ease with their positive results. One previvor explained "*I had a little fear and felt unprepared to know how to proceed, but I was certain I had the proper people to let me know how to go. And in fact, as I recall, my primary care doctor was the one who first contacted me and let me know about this and had a plan of further screening tests for me to do. He made a very rapid appointment in person just a couple of days later for us to discuss it."* (M, 74, PH, *BRCA2, APC*). Previvors felt better about the results after getting a follow-up care plan in place.

2.2 Talking with a genetic counselor and getting more specific information

Meeting with a genetic counselor was another major support for the previvors. Participants often spoke of genetic counseling appointments as the place where they learned about their true risk of developing cancer. One participant said, "*I think it got better when I chatted with one of the providers on your guys' end, and she said the chances were low for it, and* *so that kind of made it go, "Okay, well, I don't really need to worry about it as much." And so I felt better over time."* (F, 31, *MUTYH*). Previvors often felt less stressed about their risk after meeting with a genetic counselor and receiving more information about their result.

2.3 Talking with family and getting support

The family was another large support for previvors when navigating feelings about results. Most previvors reported spending the most time talking with their family members about their results, compared to providers and genetic counselors. One previvor explained *"Well, I sat down with the test, and showed it to my wife, the results. We went over it and we discussed it and said, "Well reality wise, what's going to happen is going to happen." But we just keep trying to do the normal things, what you can do in one's life to try to prevent."* (M, 71, *MUTYH*). Another previvor talked about his family's experience with pathogenic test results *"My mom and one of my sisters also have tested positive for the MSH6, so we're talking more from a supportive... Everybody's supportive… It's just something I guess that binds us. I mean, we share information. If one of us finds, sees a study about other things..., and we had a thread where we were sharing*" (Male, 51, *MSH6*). Another previvor shared a laugh over their conversations/relationship with their siblings "*You'd have to see my siblings and I. We get together and we go, Oh my gosh, look at my arm…. We just get together and laugh and go, Hey, look at my back. I couldn't wait to go down and show him my lip and say, Look at my lip. Yeah. Yeah. We joke about it*." (Female, 71, PH, *MUTYH*). Family played a huge role in helping previvors process the results and decide what steps needed to come next.

2.4 Appropriate next steps

Previvors were reassured by the additional information they received about their results. Information was obtained from a range of different sources including those discussed above (providers, specialists, genetic counselors). They felt empowered to take the necessary next steps that came along with receiving the genetic test results. After being asked about their feelings, one previvor said "*First, a little bit of excitement, well, kind of. But after I educated myself a little bit so I calmed down*." (M, 69, PH, *CHEK2*). Again, reiterating the idea that initial emotions and reactions tend to fade as more information becomes available.

3. Previvors are often more concerned about their family members' genetic cancer risk than their own

Concerns for family members' genetic risk for cancer often outweighed the previvors' concern for their own risk. While some previvors did not feel much concern about their own risk and others were concerned, uniformly all previvors spoke of their concern for their relatives. When asked if they were concerned for family members one previvor said "Well, of course, with so much in the family, even though the other ones apparently are not of the same gene. But obviously, the family's not doing well when it comes to cancer." (F, 81, PH, ATM).

3.1 Age- Those that are older find it more relevant for family, those that are younger consider the chances of passing it on if they decide to have children of their own

Just over a third of participants in this sample were over the age of 70 and many previvors felt that younger relatives could benefit greatly from the genetic testing results. When asked about their feelings one previvor said they were *"Concerned. I was concerned for my future, and I have a daughter and I worry about her."* (M, 71, *MUTYH*).

Another said *"Well, I have five granddaughters so I was concerned, thinking of passing this on to them wasn't something I wanted to pass on. So, I was just praying that it stopped with me.*" (F, 81, PH, *ATM*). Concern for family members was seen in the younger participants of childbearing age as well. One shared details about what was discussed with the genetic counselor stating *"We talked about my potential risk was low, whether or not it could be something that my kid could inherit. We talked about that as well."* (F, 31, *MUTYH*). This indicated that no matter the stage in life that previvors are in there is a great concern for family members.

3.2 Sex- Perceived implications of the variant depending on sex

A reoccurring perception of risk for female relatives compared to male previvors was also found. In particular, men who tested positive for variants classically known to be associated with cancers in the female organs were relatively unconcerned about the possible implications the result would have for them while they were very concerned about their female relatives. One previvor explained "*I mean, maybe a minor amount of concern, but not anything that was particularly impactful to me. I thought, honestly, with my specific results, the mentioning of female members of my family and the more significant impact it could have on them in terms of potential breast cancer, I think, was more of a concern for me than my own increased risk.*" (M, 41, *CHEK2*). The idea that it is a greater concern for females also came up when talking about family members getting genetic testing. One previvor said "*I have two sons, but they have yet to be, I keep reminding them, you really need to go and be tested because they both have daughters* *so far. So I told both of the daughters, my granddaughters, I said, you might want to get tested because your dads probably aren't.*" (F, 81, PH, *ATM*).

3.3 Communication with family and encouragement of cascade testing

Some previvors showed greater concern for their family members because they felt that they were already doing everything they needed to for themselves. Previvors urged their family member to get tested so they could start taking preventative action if needed as well. One previvor said "*...I keep reminding them and I want them to get tested. So we've discussed all the ins and outs of it and so hopefully when she's back home now she'll go and get that done plus my other granddaughters and family.*" (F, 81, PH, *ATM*). Many previvors spoke of reminding and encouraging their family members to get genetic testing after finding out their results.

4. Previvors have many competing health priorities aside from predisposition to hereditary cancer, other health priorities often have greater relevance in their day-to-day lives

Competing health priorities make the potential for developing hereditary cancer less of a concern. Previvors have too many other things to worry about to be caught up on their predisposition for hereditary cancer

4.1 Having cancer or getting cancer generally

Some previvors talked about having gone through cancer prior to participation in the study. Previvors reiterated that the concern was around the immediate implications of having cancer or having been through it and less about the potential for it. One previvor argued "*The matter is that I was diagnosed with prostate cancer, so I'm not a previvor already. I'm on active* *surveillance already. I don't know. I'm not a previvor."* (M, 69, PH, *CHEK2*). Additionally, some previvors highlighted their concern for cancer generally and explained that it was not specific to the genetic test results or where it may come from. When asked if they were concerned, they responded, "*Not genetic cancer risk probably, just more cancer risk."* (F, 31, *MUTYH*).

4.2 Other health issues

Previvors also brought up several additional health priorities that hold their focus on the day-to-day. One previvor shared how their mindset shifted after having a heart attack, saying *"Plus recently I had a heart attack, so prostate cancer is on the back of my head but not in the front. My major concern is different right now. Previously, I was all about my cancer. Now it's more since, yeah, it's reduced my life expectancy pretty severely, so I'm more about a heart problem rather than a cancer problem."* (M, 69, PH, *CHEK2*). This was just one of many alternative health conditions brought up when explaining that their predisposition to cancer often rests in the back of their minds.

4.3 Older age

Health concerns become more abundant as people age. Previvors who were older often explained that worrying about genetic cancer risk was not at the top of their mind. They explained that at their age things are as they are. One previvor said, "*In reality, I look at it and I say, "Well, I'm 72 years old." It would be different if I was your age, if you understand what I'm trying to say*." (M,71, *MUTYH*). They expressed less fear since they've already lived so long without getting cancer. As people get older, they have an increasing number of health concerns. The idea of not worrying about what you cannot control came up when talking to older previvors about their results and the feelings they had. This idea also came up when discussing decisions of taking action.

Behavioral

5. Accepting and awareness

Overall previvors were very accepting of their results. Those who felt it necessary to take action felt that they were empowered to do so and started to follow through with those steps. One previvor explained “*I think that now this is a reality, knowing it's part of my awareness of just general health and what is out there. So, I think if anything, it's affected my behavior [more] than feelings. I've accepted it and now if there's anything... Whether there's [anything] I can actually do to control it, I'm doing what I can. What's within my control, I'm doing.*” (M, 51, *MSH6*).

5.1 It is what it is, deal with it and move on

Previvors were realistic about what the results meant. They understood what actions they could take in response to the results and moved forward from there. One previvor said “I kind of think I just dealt with it and moved on... You kind of deal with it and move and just move on and say, "Okay, let's get it taken care of and then get on down the road." (F, 69, PH, *PMS2*). This highlighted the idea that previvors accepted the steps that could be taken and were ready to take them. This behavioral theme was related to the emotional theme (2), initial fear fades as more information becomes available. Previvors feel better about results when they know what their next steps are and describe having greater self-efficacy for pursuing preventative measures.

6. Age plays a large role in whether people feel that taking action is needed

There were varying perceptions on the relevance/necessity of acting in response to the results. The spread of responses was strongly attributed to the ages of previvors.

6.1 Those who are older may find taking preventative action less relevant/useful

Those around age 70 and older saw their results less relevant to their future care. One previvor stated "*At my age, it's different than if I were a really young person, and had a whole lifetime ahead of me to worry about these things.*" (F, 79, *PMS2*) Another explained "*Life is short anyway, so and especially my age, you've got to be focused on more important thing in life than just your.... Health is important. Of course. Yeah. Because it relates to your mental state and how you view the world, but it's not all that life has to offer*." (M, 69, PH, *CHEK2*) This theme was closely related to the emotional theme of concern for family members. Older previvors saw the results as useful information for informing younger family members as opposed to information to inform their future care.

6.2 Those who are younger may think it is too early to start engaging in preventative action

On the other side, younger providers see the results as information that they can utilize in the future but do not see the immediate relevance. When asked if they would have continued with genetic testing if it was not offered for free, one previvor said "Probably not. Not now. It maybe would've been something I would've done later." (F, 31, *MUTYH*).

7. Provider communication and confidence in providers can lead to informed decision making

People who had conversations with their providers displayed higher confidence in informed decision-making. Whether the decision was to increase screenings, undergo preventative surgery, or leave their follow-up care plan as is, those that expressed confidence in their providers also displayed the highest confidence in moving forward with their results. One previvor detailed their conversation with a provider explaining "*...the geneticist looks at it, she said, "This is how you should look at it. You got a 30% chance of getting it, but you got a 70% chance of not getting it." But that depends on whatever it depends on. She said, "Do you want to take a risk at 30% or do you want to just take your chances at 70%?" That's really what it boils down to. Which is kind of interesting, looking at from somebody that's not really a gambler*." (F, 69, PH, PMS2). The previvor illustrated a strong understanding of their results and the potential implications associated with the results.

7.1 Communication with providers can lead to appropriate outcomes for non-actionable variants

Not all previvors felt that using their genetic test results moving forward was important. When asked why they chose not to take the recommended next steps one previvor responded with "I think the only reason why I may not is because they said the chances are so low. That might be... I don't want to do unnecessary procedures and unnecessary if there's no, like the odds are not against me, I guess." (F, 31, *MUTYH*). When they received appropriate communication about their variant, previvors were both content and confident with the next steps.

8. Genetic testing leads to better health outcomes

Overall previvors voiced the importance of genetic testing and its ability to improve the health and wellbeing of individuals. When asked about their thoughts related to genetic testing in the primary care setting one previvor said "*I think it is a wonderful idea. I think, in my opinion, I think that more people should be aware that it is available, and the study and the research that you are doing, and that you have shared with me, has made a significant difference.*" (M, 71, *MUTYH*)

8.1 Value of genetic testing for hereditary cancer

For some previvors, the genetic test results lead to additional screenings which ultimately lead to a cancer discovery/diagnosis. The previvors were incredibly grateful that they did the testing because without it they would have had no idea that something was wrong at the time. One previvor said "*But without having had the EDGE study participation, I would still probably have no symptoms whatsoever until I would've had stage IV disease and my prognosis would've been terrible. So without having done this genomic study, I think I would've a year or two from now be faced with a very horrible prognosis. And as of today, my prognosis is for, you know, and I'm in remission and hopefully will remain so for the rest of my life.*" (M, 74, PH, *BRCA2, APC*)

Discussion

This study set out to evaluate experiences following receiving positive test results from a multigene cancer panel, including how people feel, who they talk to, and what choices they make in order to better understand the type of support previvors need when facing difficult decisions regarding cancer risk management. Overall, previvors conveyed that having a family history of cancer had prepared them to receive positive results when undergoing cancer genetic testing. This highlights the value that previvors place on family history as a tool for evaluating risk. Additionally, this helps explain why previvors with a family history are often not surprised and do not have very strong emotional reactions to their results. Our findings were consistent with the current literature suggesting that patients experience a wide range of feelings that can be both positive and negative[28]. Our data suggest that previvors feel better about their results after receiving more detailed information about what the results mean and having conversations about what the next steps will be. These conversations can take many different forms, whether it be between a patient and provider, patient and specialist, or amongst trusted friends and family members. The root of all these conversations was confidence and support. Previvors felt more at ease with the information when they had confidence in their providers and felt supported by friends and family.

Across the board, previvors' greatest concerns were those regarding their family members. This not only indicated the possible lack of relevance the results may have had for the previvor but also highlighted the perceived importance of information sharing and cascade testing. In contrast to what was found by James et al., that cascade testing is not frequently encouraged, we found that cascade testing was often encouraged by relatives. This could speak to our study population's mean age being older and therefore the perception was that younger family members may be more likely to benefit from testing[18]. Previvors also discussed competing health priorities as justification as to why their pathogenic results were not a top priority for them. For those who had other more pressing health issues, the potential increased risk for developing hereditary cancer was just another piece of information for people to be aware of. This result may have been skewed due to our slightly older sample population which is more likely to have other health issues/concerns.

When it came to making decisions about how to move forward with the results previvors often conveyed that they were taking the appropriate steps that they saw fit. Age played a large role in determining the perceived need to take action in response to the results. Older individuals tended to be on the side that it was a little late in life to worry about making changes or felt that they had already been taking all the necessary steps. Those who were younger leaned more towards the idea that it was too early to take steps now, but they would plan to use the information in the future when the timing was more appropriate. Communication was the first step for the majority of previvors in creating a follow-up care plan. Talking through their results and risk management strategies led to subsequent informed decision-making about future risk management plans. There was a shared understanding and acknowledgment of hereditary cancer genetic testing and its utility for previvors and their families.

Implications

The findings from the EDGE Study highlight the importance of communication and support systems after receiving multigene cancer panel testing. Communication among primary care providers is vital in this model because not all patients underwent genetic counseling despite all being contacted for free services through Color Health. This emphasizes the need for provider knowledge and confidence when having risk management conversations regarding all levels of gene penetrance. Our findings suggest that confidence in providers and communication lead to trust in information and informed decision making. It is well known that providers are already overburdened and lack the time to integrate sharing of genetic information into routine clinical practice[29,30]. Future research should look to implement supports within primary care that could aid in the communication of genetic test results within clinics. Patients and providers may benefit from having genetic counseling appointments scheduled automatically when genetic testing results are available. Communication is critical following testing so having appointments to discuss results scheduled automatically could help ensure that these conversations happen. The use of AI for interpreting results and communicating appropriate risk management strategies should also be explored as a possibility for removing the responsibility from providers while still ensuring adequate communication of results.

Previvors expressed great concern for their family members in the population-based screening implementation model. Communication with family members regarding cascade testing should also be explored at length in future research. Specifically, assessment on information accuracy could tell us more about the accuracy of family communication surrounding genetic test results and risk estimates. Risk communication accuracy continues to be a challenge in the implementation of routine genetic testing[31-33]. Population-based screening models like EDGE paired with cascade testing could alleviate the burden of communicating risk on individuals as more people are routinely tested in primary care and through cascade testing following identification of a pathogenic variant in a biological relative[34-36].

Future research should also further explore perceptions in younger and middle-aged previvors from multigene cancer panel testing. Our findings suggested that older previvors may find lower utility in the genetic test results which could mean there is greater utility in the results for younger previvors. This previvor perception appears to align with the greater public health benefit (i.e., potential greater number of life years saved) of provide genetic risk assessment to younger adults. The utility of results should be assessed in a greater sample of middle-aged participants to confirm these findings.

Limitations

One clear limitation of this study was the selection bias of our study sample. To qualify for testing, participants had to have an annual appointment with their primary care provider, indicating that they are involved in routine healthcare visits. Participants with a family history of cancer who consider themselves at higher risk are also more likely to fill out screenings related to family history. Additionally, these participants made the decision to enroll in the study and receive the results from genetic testing which could select participants who are more likely to engage in information-seeking activities than the general population. It is also important to note that those who think they will be unable to handle results do not proceed with genetic testing. Another weakness of this study is the homogeneity of our study sample with most participants self-identifying as white non-Hispanic.

Conclusions

The primary aim of this study was to evaluate previvor experiences following multigene cancer panel testing. Understanding how people receive the results and utilize their genetic information is critical in addressing the gaps in support needed to move genetic testing for hereditary cancer into the primary care setting. The study findings offer insights into how previvors feel initially versus after they receive more information, pinpointing the need for increased communication efficacy in patient-provider relationships as well as family systems. Information regarding the timing in life of cancer genetic testing could be further explored to increase utility. This study identified a lack of perceived utility in older participants and therefore future research could benefit from studying younger and middle-aged populations. Genetic testing is gaining traction and acceptance as a clinical tool, and previvors are expressing more awareness and understanding of their results. Future work is needed to understand the utility of multigene cancer panel testing and how it translates to adherence to follow-up cancer risk management plans.

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Appendix: codebook

Code Name	**Category**	**Code Description**	**Inclusion/Exclusion Criteria**
Already doing it	Personal	Talk about things that they were already doing that are good and that they will continue to do because they are things that would be recommended	
Changes in health behavior	Personal	Any mentions about changes or lack thereof after receiving test result, any reasons behind why they did or did not decide to make changes	Code if changes were made OR comments about why they did NOT make changes
Cons of GX	Personal	Things that people think are bad about genetic testing. OR Reasons they don't want to have genetic testing.	
Cost Considerations	Cost	Things people balance when considering cost of gx testing	
Cost of Gx	Cost	Comments, feelings, and thoughts related to the cost of genetic testing	
Delivery method of results	Study	How patients received test results	
EDGE Impact	Study	What role did participating in EDGE and receiving the results of the genetic test have on the patient's life. Only code if there was an impact of some sort.	
Expectations	Emotions	If people were surprised by the result or got something different than they expected (different type of mutation, etc).	
Family	Family	Any comments related to family, could be about conversations with family, concern for family / family risk, family plans for genetic testing, etc. If the patient did NOT speak to their family, this should be coded as "family - no discussion"	Do NOT code if they talk about not speaking with family and reasons why.
Family- no discussion	Family	Comments about not speaking to family and reasons for choosing not to discuss with family	Code only if they did not speak with family members
FH	Family	Family History of Cancer, includes any relatives with cancer.	Do not include personal history here
Follow-up Care Costs	Cost	The costs associated with follow-up care, copays, out-of-pocket costs, etc.	
For discussion	Other	Marker for something we may want to come back to and discuss	
Gene penetrance	Personal	Comments about level of risk based on variant	Include comment about if risk is high or low, if they are worried about it or not
Genetic counselor	Healthcare	Comments about genetic counseling appointments or lack thereof.	Code if they saw a genetic counselor OR if they talk about not seeing a genetic counselor and reasons why
Health Assessment	Personal	How people feel about their health, why people view their health the way they do, what factors people consider when assessing their health, how the gx result factors into their view of their health	
Insurance	Cost	Do they have insurance, how they feel about their coverage, what they think is covered and what is not by insurance when it comes to genetic testing and follow-up care	Code if they have insurance or if there are mentions of not having insurance
Max cost	Cost	Numeric value reported	
Other support	Other	Support received outside of family members and	Do not code if they say they did not

		providers/gx counselors. Only code if other support was actually received, not if they said "no" when asked.	seek support
Other thoughts	Other	Additional comments, thoughts, experiences shared	
PH Cancer	Personal	Personal history of cancer; experience with cancer (including diagnoses made after the testing)	
Plans moving forward	Personal	Follow up care plans, plans for discussions (family, providers, etc.), what people are planning to do with this information, including no plans to use this information	
Preventative surgery	Healthcare	Comments about having preventative surgery, planning to have, weighing the decisions to, not wanting to, etc.	Include comments about getting surgery, not getting surgery considering surgery or not considering surgery
Previvor Report	Study	Comments about receiving the previvor report, if it was useful or not, how they felt about having it	
Previvorship	Personal	How people feel about the term previvor, do they identify with it, do they think it is good or bad, how they think it may be helpful or harmful if they have any thoughts about the term	
Prior gx testing	Personal	Anything about gx testing prior to the EDGE study	
Provider	Healthcare	Comments about meeting with a provider, discussions with provider; could be about testing, test results, follow up care, confidence in provider, etc.	
Provider- no follow-up	Healthcare	Patient has NOT met with their provider to talk about their results	Only if the patient has not met with provider
Provider response/ provider learning	Healthcare	The provider's experience. What the provider did, how they reacted to getting the results (emotions, actions, learning more about genetics).	
Quotable	Other	Good quotes that we want to mark	
Reactions to results	Emotions	How people reacted to receiving a positive result; emotions, feelings, actions, etc.	Positive or negative
Recommendations for EDGE	Study	Recommendations to improve study/ things that patients expressed would have been helpful/ things that were needed but were missing, etc	
Referral	Healthcare	Referrals given to patients after receiving their results	
Risk - Family	Family	Risk for family members; whether the patient OR the family members themselves consider them to be at risk	Include patient's feelings about their family members risk or the patient talking about how their family member feels about their risk
Risk - Personal	Personal	Patients perceived risk	
Routine tests	Healthcare	Comments about tests such as colonoscopy, mammogram, etc.	
Specialist	Healthcare	Comments about seeing a specialist, could be referred, already have a specialist, conversations, recommendations made by specialists, etc.	Only code if they see, speak, or are referred to a specialist
Study feedback	Study	Comments about the experience in participating in EDGE, what worked, what didn't work, and how they liked it or not	