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Research Report

Perceptions and care Recommendations from Previvors: Qualitative analysis of female BRCA1/2 mutation Carriers' experience with genetic testing and counseling

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ABSTRACT

Introduction: It is estimated that 12.5% of women will be diagnosed with breast cancer and 1.10% with ovarian cancer during their lifetime. Although less common, women with these mutations have a 11–72% increased risk of breast/ovarian cancers and are hereditary. Genetic testing/counseling presents the opportunity to identify carriers of *BRCA1/2* genetic mutations before a cancer diagnosis.

Methods: Thirty-four *BRCA1/2*-positive women (with and without histories of breast/ovarian cancers) were recruited through online national support groups to gain a better understanding of their genetic testing/counseling perceptions and experiences. After confirming eligibility, they were invited to participate in either a telephone or webcam interview. Interview transcripts were analyzed using qualitative thematic text analysis and descriptive coding techniques.

Results: Six major themes emerged, capturing the perceptions and experiences of genetic testing/counseling for these women: 1) Emotional Reactions to Results and Genetic Counseling, 2) Future Recommendations, 3) Family Solidarity and Support, 4) Experiences with the Healthcare System, 5) Preventive Concerns and Decisions, and 6) Sources Affecting Perceived Risk. Two subthemes also emerged within the first theme, which are termed "Previvor," and "Testing Intuition."

Conclusions: Participants indicated that genetic testing/counseling improvements would be helpful for women in this population surrounding quality care, including sensitivity training for healthcare professionals involved in testing/counseling, additional educational resources, and increased emotional and financial support. Although these recommendations may be beneficial, more widespread research with greater generalizability to disparate groups may be necessary prior to implementation.

1. Introduction

In the United States (US), 12.5% and 1.10% of women will develop breast and ovarian cancers, respectively, at some point during their lifetime. A minority of the US population have familial factors associated with breast/ovarian cancers and actually have hereditary *BRCA1* and/or *BRCA2* mutations (Suryavanshi et al., 2017; National Cancer Institute, 2022a). An estimated 72% women with a *BRCA1* mutation and 69% of women with a *BRCA2* mutation will develop breast cancer before the age of 80 (National Cancer Institute, 2022b). These numbers are lower in ovarian cases, where women with *BRCA1* mutations have a lifetime risk of 39–44% and those with *BRCA2* have a risk of 11–17% ([2]). Regardless of breast and ovarian cancer risk, female *BRCA1/2* mutation carriers have an increased risk of other malignancies including endometrial, uterine, and pancreatic cancers as well as an "elevated risk" for cutaneous and ocular melanomas (Petrucelli et al., 2016; Shu, et al., 2016). These women also experience related invasive recurrences and metastatic cancers (even more so among those with cancer histories) more often than women without these mutations (Song, 2020). These mutations occur among biologically-related family members and have adverse consequences for those affected, including an increased risk for breast/ovarian cancers, pancreatic and melanoma cancers, stress,

Abbreviations: BRCA, BReast Cancer; DTC, Direct-to-consumer; MRI, Magnetic resonance imaging; NCI, National Cancer Institute; PCP, Primary care physician; US, United States.

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anxiety, worry, and financial burden (Suryavanshi et al., 2017).

Genetic testing and counseling presents the opportunity to identify carriers of *BRCA1/2* mutations before a cancer diagnosis occurs, but consequently, introduces the potential of experiencing anxiety and fear. Genetic testing is primarily recommended for those individuals determined to have a high risk of a genetic mutation depending on various personal and clinical factors: having a previous cancer history, a family member test positive for *BRCA1/BRCA2*, family members who have been diagnosed with breast/ovarian cancer(s) at age 50 or younger, cancer in both breasts or ovaries, triple-negative breast cancer, and/or being of Ashkenazi Jewish heritage (Colombo et al., 2018; Daly et al., 2015; Riley et al., 2012; Niendorf et al., 2016). There are some experts, however, that argue that all women aged 35 and older should undergo genetic testing as a form of population-based breast and ovarian screening to reduce the economic and diagnostic burden of cancer incidence (Manchanda and Gaba, 2018). More recent literature has discussed this possibility but on a case-wise basis, suggesting that those with a 10% chance of having a *BRCA1/2* mutation (based on personal and family history) should be approved for testing (Ficarazzi et al., 2021).

Previous research outlining women's experiences with genetic testing and counseling has primarily focused on *BRCA1/2*-positive women who have received a breast/ovarian cancer diagnosis (Katapodi et al., 2004; Taber et al., 2015). Although some research has explored the relationship between receiving a *BRCA1/2* diagnosis *without first receiving a cancer diagnosis*, both areas of research have identified doctor/patient communication patterns involving treatment methods (i.e., surgery versus surveillance) as well as emotional and cancer-related outcomes of these treatment(s) (Hesse-Biber and An, 2016). Due to recruitment and availability issues, research measuring availability, satisfaction, and other components of genetic testing/counseling often recruit women who have undergone these processes but not necessarily those who have tested positive. Other research has highlighted women's attitudes towards genetic counseling, finding that genetic counselor thoroughness and emotional sensitivity determines the comfortability of patients, in addition to how genetic results are disclosed (e.g., phone, email, web-based application, mail) (Ormondroyd et al., 2012). A paucity of research has found that genetic mutations alter the dynamic of family systems and relationships. As outlined in past qualitative research, the nature of *BRCA1/2* genetic mutations often suggests an impending cancer diagnosis at some point in one's life (Dean, 2016; Hesse-Biber, 2018), which in turn can negatively impact solidarity of family relationships and communicative patterns (Dean et al., 2017; Dean and Rauscher, 2018). While genetic testing/counseling is growing in popularity and accessibility, past literature has overlooked the impact, both negative and positive, that genetic testing/counseling may have on women at risk for these mutations. Therefore, they may be unable to provide a firsthand glance into what it means to undergo testing/counseling and test positive for *BRCA1/2* mutations.

Therefore, the primary aim of this study was to further explore and understand the *lived experiences* and *preferences* of *BRCA1/2*-positive women who have completed genetic testing or counseling, focusing on the process of testing/counseling and what happens after testing positive. Although research on *BRCA1/2* genetic testing and counseling is growing, unanswered questions remain surrounding women's attitudes toward being tested and its tertiary outcomes. This research conducted in-depth interviews with women who had completed *BRCA1/2* genetic testing/counseling within the past five years regardless of cancer history to better understand their lived experiences, attitudes, and preferences regarding the entire genetic testing/counseling process.

2. Materials & methods

2.1. Study design and participant recruitment

This research was the second phase of a larger two-phase mixed methods research project funded by a grant from the Connecticut Breast Health Initiative, Inc. (CTBHI). Participants were recruited for Phase 1, beginning in 2019, through three national, online support groups housed on Facebook: 1) *BRCA1* or *BRCA2* Genetic Ovarian and Breast Cancer Gene Group (~6,800 members), 2) *BRCA* Genetic Sisters Group (~2,700 members), and 3) *BRCA* Strong Group (~1,300 members). One study recruitment post was posted per day on each support group's website (*BRCA* Strong Group only allowed one post per week), with prior written permission obtained from each group's moderators. The post consisted of a brief announcement introducing the study, eligibility criteria, compensation offer for participation, and a link to an anonymous screener via Qualtrics©. At the completion of Phase 1, participants were invited to further participate in a follow-up interview with the study coordinator (Phase 2). If agreeable, participants were able to schedule the date and time of either a phone or webcam interview through a built-in scheduling option on the Facebook study webpage. Participants were eligible if they were 18 years or older, female, lived in the US, and tested positive for *BRCA1* and/or *BRCA2* genetic mutations within the last five years. Participants were ineligible if they were male, could not speak and/or read English, were currently undergoing genetic testing/counseling for *BRCA1/2* genetic mutations, and/or did not reside within the US (obtained by Qualtrics© geolocation). The current study was reviewed and approved by the University of Connecticut Institutional Review Board (IRB#H18-173).

2.2. Interview protocol

A semi-structured interview guide was developed and modeled after an interview script constructed by Augestad and colleagues (Augestad et al., 2017). Interview questions focused on several areas of interest: perceptions and experiences of genetic testing/counseling; delivery of genetic test results (e.g., in-person, web-based, telephone, mail), emotional reactions and responses to test results; and how genetic testing/counseling can be improved (see Supplement 1 for interview guide questions). Before beginning the interview, verbal consent was obtained to record the participants' voices and/or likeness. Interviews lasted between 30 and 90 min. Adequate sample size was contingent on data saturation, suggesting between 20 and 40 participants (Morgan and Nica, 2020; Vogel et al., 2018; Braun and Clarke, 2006; Braun and Clarke, 2019). Saturation occurred after the 28th interview; however, all 34 interviews were included and analyzed.

2.3. Data analysis

Interviews were transcribed and analyzed using the standard procedures of qualitative thematic text analysis (Morgan and Nica, 2020; Braun and Clarke, 2006; Braun and Clarke, 2019). Two members from the research team independently coded interview transcriptions with the goal of identifying similar meanings and patterns within and across the interviews. Simultaneously, memoing was conducted by each research member to capture their reflective notes (Cicero et al., 2017). Upon completion, coders met to organize the meanings and patterns recorded into broad themes based on interview questions and study goals. After identifying broad themes, each coder independently conducted an analysis using grounded descriptive coding techniques (Morgan and Nica, 2020; Braun and Clarke, 2006; Braun and Clarke, 2019). This ground-up approach utilized an ongoing process in which initial insights influence the direction of subsequent questions and interview topics (Ormondroyd et al., 2012; Dean and Rauscher, 2018). Across several meetings, results were compared and thoroughly discussed for consistency with the goal of identifying the overarching (major) themes and subthemes. The last activity involved re-reading through coded transcripts to choose representative quotes for identified themes and subthemes.

3. Results

A total of 34 *BRCA1/2*-positive women completed in-depth semi-structured interviews. They ranged in age from 28 to 40 years, averaging 33.3 years of age. The majority were white ($n = 32$), non-Hispanic ($n = 34$), married ($n = 25$), employed full-time ($n = 24$), and earned a college degree ($n = 29$). The average number of years since genetic testing/counseling was 2.29 (ranging from 3 months to 4 years), and all women had medical insurance at the time of testing or counseling. Genetic test results were split, with half testing positive for *BRCA1* and the remainder for *BRCA2*. A total of eight women reported having *BRCA1/2*-related cancers prior to completing genetic testing/counseling.

3.1. Major themes

Six major overarching themes emerged describing *BRCA1/2*-positive women's experiences with genetic testing/counseling: 1) Emotional Reactions to Genetic Results and Counseling; 2) Future Recommendations; 3) Family Solidarity and Support; 4) Experiences with the Healthcare System; 5) Preventive Concerns and Decisions; and 6) Sources Affecting Perceived Risk. In addition to these themes, two subthemes were identified within the first theme, "Emotional Reactions to Genetic Results and Counseling", highlighting common thoughts the participants experienced throughout the genetic testing/counseling process: 1a) "Pre-vivor", and 1b) "Testing Intuition". The following will discuss and detail each of these themes, from the most to least impactful. Please see Fig. 1 for a pictorial representation of the themes identified, where they are represented in mindmap format with how many women introduced a specific theme (or subtheme) in their interview. Table 2 shows participant quotes from each major theme and subtheme.

3.2. Major theme 1. Emotional reactions to genetic results and counseling

Emotional reactions were mostly negative throughout the genetic testing/counseling process, mostly relating to *BRCA1/2*-positive women's genetic test results. Women reported feeling shocked and overwhelmed by their results, the push for undergoing prophylactic surgeries, and their impact on personal and familial relationships. They noted feeling "floored", "stunned", "knocked off-kilter", "angry", "devastated", and "frustrated" when genetic test results were positive, and they knew having such a mutation was out of their control.

> *"I was so overwhelmed. I was sitting there, a perfectly healthy woman who, at 33, knew she was going to lose her breasts. It took me weeks, if not months, to come to terms with."*

"The news that I was positive shook me to the core. I literally felt like I was punched in the stomach, and that feeling did not go away.".

A subset of these women experienced depression and loss, especially regarding post-test result recommendations for prophylactic surgeries.

> *"I started therapy after my surgery [on my own accord], but I wish I started it in advance. Because I think people don't really prepare you for the loss. People don't give you a space to talk about how it is a loss and a painful situation."*

When participants were asked if they experienced positive emotions during genetic counseling, empowerment and self-advocacy were most common.

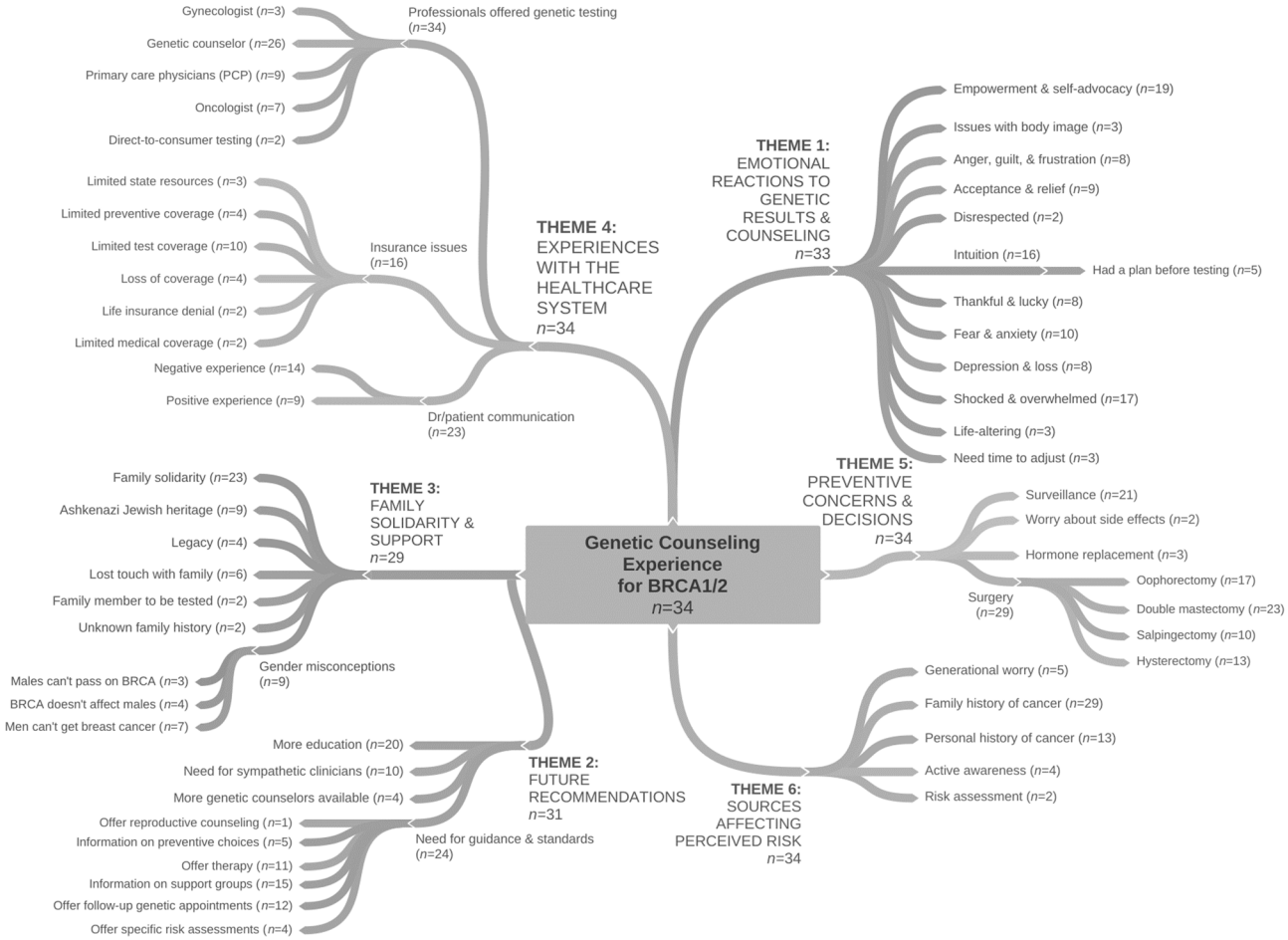

Fig. 1. Diagram of interview themes and important topics (N = 34).

Table 2
Exemplary participant quotes from each major theme and subtheme (N = 34).

	Theme	Exemplary Quotes
1.	Emotional Reactions to Genetic Results & Counseling (n = 33)	"I was so overwhelmed. I was sitting there, a perfectly healthy woman who, at 33, knew she was going to lose her breasts. It took me weeks, if not months, to come to terms with." "The news that I was positive shook me to the core. I literally felt like I was punched in the stomach, and that feeling did not go away." "I've been very vocal about my results and my risk has helped other people learn, you know. My mom died from breast cancer, my grandmother died from breast cancer, and both aunts had ovarian cancer, and so, where do I start? I'm glad that I have a pushing off point to share my positive experiences and willing to share information, being there to help other people."
1a.	Pre-vivors (n = 9)	"It's a hard thing to explain to people. It's like, 'I don't have cancer, I just have a really, really high chance of getting cancer in the future, so I'm going to have this really radical surgery.' That was one challenge - I didn't know how or what to tell people." "I just don't want to live my life waiting for cancer. There were many days at the beginning that I was despondent, cried a lot. I would go in the shower, taking about half an hour, and sob because I didn't want my kids to know what I was crying about. It affects them too."
1b.	Testing Intuition (n = 16)	"When it came time to get my results, I pretty much already knew. I think I've known for a long time and was able to come to terms with it quickly." "When I went in for the testing, I felt really comfortable with the time, and comfortable with what I would do if I received a positive result – I had a plan."
2.	Future Recommendations (n = 31)	"After what I had just gone through, I needed a doctor or medical staff or whoever to understand what had just happened – what this meant for me. I desperately wanted someone to know where I was coming from, and how fearful I was. There was nothing like that for me." "That's how I knew I had it – from a freaking webpage. I was sitting there at this computer, I was at work, and I just started bawling! It's a moment I'll never forget, looking at the screen and it said, 'BRCA2' and I was like, 'holy shit.'" "I think follow-up genetic [counseling] appointments are a great idea. You get all this information, and not to mention, life-changing news, and you're supposed to be ready to ask questions after? I just sat there and shut down. Having that time to process would have been great for me." "I know doctors who are ignorant who tell patients, 'Oh there's always this sort of risk; men can't pass it on…I mean, doctors are telling patients this, and to me, it seems bizarre.'" "You should be hooked up with someone that walks you through the process and you know, follows you until the end."
3.	Family Solidarity & Support (n = 29)	"Between my two sisters and I, who are positive, I didn't want to be left out. I can't believe I'm saying this, but I would have felt left out if I didn't have BRCA." "My older brother is positive too, and he kind of looks at it more along the lines of, 'Well, we know how we're going to die.' I keep talking to him about it, telling him it's not a guarantee…I'm pretty positive about it and I'm glad to know that I'm able to do things and try to look on the bright side." "You find out your genetics are bad, you've got to wrap your head around that and think about my children, my grandchildren, the family line." "Years after my aunt was tested for BRCA, I find out from our family Facebook chat that she was positive. One of her sons had it, then two out of three of his kids had it, but she never told anyone. When my cousin reported to my sister that everyone on that side was tested in the early 2000 s but never told anyone, we're all like, don't you think we should've known back then?" "I had two cousins who were my mother's nieces both tested for BRCA2 in like, 1997. They never told anyone in their extended family. I think a lot would've been different in my life if I had received a 'dear family' letter from them, or even a phone call just to discuss this. I don't want to be like, 'Oh you know, we haven't talked for 30 years, and here I am. I'm going to talk to you about my mutant cancer gene. No, I didn't want to do that.'
4.	Experiences with the Healthcare System (n = 34)	"This technician was really like hell on wheels. I left that mammogram very aware of my own mortality like I'm going to die tomorrow. She took it upon herself to scare me with statements, telling me to immediately get a hysterectomy before age 40, a double mastectomy, and find an oncologist to get me on tamoxifen right now. She also told me to put all my paperwork [regarding BRCA] in a fireproof safe so that when I'm gone, the information doesn't die with me." "My genetic counselor got me in for active surveillance the day after my genetic test results – I kid you not – until I could get in to see a surgeon. She probably saved my life." "I had to wait a year to get tested because my parents were in-between insurance coverages. I took the time to come up with a plan if I was positive. At the end, I was comfortable with my plan." "At the time [I was going to be tested] I didn't have insurance. I didn't want to pay out-of-pocket to go to the doctor and have them tell me my results, so I contacted the company that ran my sample to get the results myself." "At first, my mammogram and reconstructive surgeries weren't covered by insurance, but my gynecologist [who did the genetic testing] wrote a letter to the insurance company and called them until they approved my claim wasn't just 'cosmetic.' She went above and beyond to help me and take the time to help me."

(*continued on next page*)

Table 2 (*continued*)

	Theme	Exemplary Quotes
5.	Preventive Concerns & Decisions ($n = 34$)	"I was recommended to have an oophorectomy and then a bilateral mastectomy two days later. I chose not to because I'm so young, but I am starting colon and pancreatic cancer screenings because those run in my family too. Endoscopic ultrasounds twice or once per year don't sound fun." "I've had it in my head since day one that if I'm positive I'm going through all of the surgeries to give myself a fighting chance." "It's taking the bull by the horns…if it were just me, I might have a totally different approach, but I've got a six-year-old daughter and eight-year-old son, and I need to be around to help if either of them have to go through something like this. There's no question for me that getting [the surgery] done is the right thing. My initial response was that I didn't even blink as soon as my doctor told me what the results were, I said 'gut me and get them off.'" "I didn't want to go into menopause that way…I wanted to go naturally. At first, I was afraid of side effects from surgery, of a hysterectomy and so forth, then the whole thought of having a mastectomy - just felt like amputations." "I've been very positive and vocal about my results and my own risk. I help other people this way. I don't think a lot of people know where to start and if they have someone who has had positive experiences and is willing to share information, it's empowering."
6.	Sources Affecting Perceived Risk ($n = 34$)	"I was diagnosed with breast cancer, my father had prostate and melanoma cancers, and his mother had ovarian cancer. I started to wonder if maybe this was relevant to me and my cancer risk." "My mother had breast cancer twice; she's a survivor. Her sister had it twice, my aunt, but the second time it killed her. Their mother passed away from ovarian cancer at an early age, and I was starting to think there's something wrong with the women in my family." "I do what I can to be proactive – I don't want the same fate. I especially don't want my children to have to see their mom go through cancer treatment if I have choices to prevent it." "I've opted for preventive maintenance, like alternate between mammograms and MRIs every six or eight months in addition to tamoxifen to prevent cancer, since I've had it in the past."

"I feel empowered by the information that I found by myself…I can take control of my own destiny really. I chose to take the head-on approach."

"Now I look at this test result as a blessing. I get to do something about my risk of cancer.".

"I've been very vocal about my results and my risk has helped other people learn, you know. My mom died from breast cancer, my grandmother died from breast cancer, and both aunts had ovarian cancer, and so, where do I start? I'm glad that I have a pushing off point to share my positive experiences and willing to share information, being there to help other people."

"It's made me more aware and bolder because now I'm not afraid to talk to people about it. If I hear someone has a relative with cancer, I'm like, 'we need to talk.' I try to at least let people ask questions if they want and act as a resource for them, which makes me feel better, in return."

Within this major theme, two subthemes emerged from the interviews: 1a) "Pre-vivors", and 1b) Testing Intuition, both of which were unexpected emotions and will be discussed below.

Subtheme 1a. "Pre-vivors". Negative emotions were commonly discussed by a subset of women who identified themselves as *"pre-vivors"* because they did not fit "cleanly" into cancer patient or survivor categories, not experiencing either. *BRCA1/2*-positive women reported experiencing anger because they had difficulty explaining their hereditary risk to others, including family members. Being a "pre-vivor" left women not being able to identify a place or define their experience in a more understandable way.

"It's a hard thing to explain to people. It's like, 'I don't have cancer, I just have a really, really high chance of getting cancer in the future, so I'm going to have this really radical surgery.' That was one challenge - I didn't know how or what to tell people."

"I just don't want to live my life waiting for cancer. There were many days at the beginning that I was despondent, cried a lot. I would go in the shower, taking about half an hour, and sob because I didn't want my kids to know what I was crying about. It affects them too."

Subtheme 1b. Testing Intuition. Some participants reported feeling a sense of "testing intuition" before they received their results, knowing the test would come back positive. These women even reported having a plan ahead of time (i.e., prophylactic surgery, chemoprevention, ongoing surveillance) for what they would do in response to a positive genetic test:

"When it came time to get my results, I pretty much already knew. I think I've known for a long time and was able to come to terms with it quickly."

"When I went in for the testing, I felt really comfortable with the time, and comfortable with what I would do if I received a positive result – I had a plan."

3.3. Major theme 2. Future Recommendations

This theme captures what *BRCA1/2*-positive women believed would improve the genetic testing/counseling experience. Recommendations fell into four general areas (in order of importance) and are depicted in Table 1.

"After what I had just gone through, I needed a doctor or medical staff or whoever to understand what had just happened – what this meant for me. I desperately wanted someone to know where I was coming from, and how fearful I was. There was nothing like that for me."

"That's how I knew I had it – from a freaking webpage. I was sitting there at this computer, I was at work, and I just started bawling! It's a moment I'll never forget, looking at the screen and it said, 'BRCA2' and I was like, 'holy shit.'"

"I think follow-up genetic [counseling] appointments are a great idea. You get all this information, and not to mention, life-changing news, and you're supposed to be ready to ask questions after? I just sat there and shut down. Having that time to process would have been great for me."

"I know doctors who are ignorant who tell patients, 'Oh there's always this sort of risk; men can't pass it on…I mean, doctors are telling patients this, and to me, it seems bizarre.'"

Overall, women suggested mandatory support services (e.g., follow-up appointments, mental health counseling, family counseling), no matter positive or negative genetic test result, to assist them and their families in processing their experiences and what this may mean for future healthcare trajectory and testing.

"You should be hooked up with someone that walks you through the process and you know, follows you until the end."

Table 1
Demographic characteristics of the interviewed sample (N = 34).

	M	*SD*
Age at survey	43.3	9.4
Years since genetic counseling	1.87	2.29
	n	%
Education		
Less than undergraduate	5	14.7
Undergraduate or above	29	85.3
Ethnicity		
Non-Hispanic	34	100.0
Hispanic	0	0.0
Race		
White	32	94.1
African American/Black	2	5.9
Marital status		
Not married	9	26.5
Married	25	73.5
Employment status		
Not working full-time	10	29.4
Working full-time	24	70.6
Region		
Northeast	8	23.5
Southeast	6	17.6
Southwest	2	5.9
Midwest	11	32.4
West	7	20.6
Health insurance at time of genetic testing/counseling		
No	0	0.0
Yes	34	100.0
Previous cancer diagnoses		
No cancer diagnoses	26	76.5
Breast cancer	4	11.8
Ovarian cancer	3	8.8
Another type of cancer	0	0.0
Multiple cancers	1	2.9
Recurrences		
Not applicable	28	82.4
None	3	8.8
1 or 2	3	8.8
Avenue for genetic counseling		
Private genetic counseling office	8	23.5
Hospital	15	44.1
Primary care physician (PCP)	9	26.5
Direct-to-consumer (DTC)	2	5.9
Missing	0	0.0
Preferred approach of genetic counseling		
Individualized	13	38.2
Family-based	21	61.8
Genetic counseling result		
BRCA1	16	47.1
BRCA2	16	47.1
Both BRCA1 & BRCA2	0	0.0
BRCA & CHEK	2	5.9
BRCA & unrelated mutations	0	0.0
Mental health therapy offered		
No therapy offered	23	67.6
Therapy offered	11	32.4

Note. $p < .05$*; $p < .01$**; Pearson chi-square analyses were not conducted for the ethnicity, health insurance at time of genetic testing/counseling, and preferred approach of genetic counseling because there was no variability.

> *"I like the knowledge is power standpoint. What can I do now, knowing this information, and be proactive?"*

3.4. Major theme 3. Family Solidarity and support

Participants felt a sense of family solidarity that they had not felt prior to testing/counseling. Family was also identified as one of the main sources of support *BRCA1/2*-positive women noted after a positive test.

> "*Between my two sisters and I, who are positive, I didn't want to be left out. I can't believe I'm saying this, but I would have felt left out if I didn't have BRCA*."

"I have a brother and sister, who are both positive, so that makes all of us. It's our Ashkenazi heritage.".

> "*My older brother is positive too, and he kind of looks at it more along the lines of, 'Well, we know how we're going to die.' I keep talking to him about it, telling him it's not a guarantee…I'm pretty positive about it and I'm glad to know that I'm able to do things and try to look on the bright side."*

Noting the sanctity of familial "legacy" was introduced by a small, but persistent group of women. They did not want *BRCA1/2* mutations to be known as the "family curse" to their children, grandchildren, and others.

> "*You find out your genetics are bad, you've got to wrap your head around that and think about my children, my grandchildren, the family line*."

Lastly, participants discussed losing touch and the lack of communication between family members and moreover, the impact this had on their genetic testing/counseling experience. Being left in the dark and feeling like they could be bearers of the "test results" added even more anxiety to an already stressful situation.

> *"Years after my aunt was tested for BRCA, I find out from our family Facebook chat that she was positive. One of her sons had it, then two out of three of his kids had it, but she never told anyone. When my cousin reported to my sister that everyone on that side was tested in the early 2000s but never told anyone, we're all like, don't you think we should've known back then?"*.
>
> *"I had two cousins who were my mother's nieces both tested for BRCA2 in like, 1997. They never told anyone in their extended family. I think a lot would've been different in my life if I had received a 'dear family' letter from them, or even a phone call just to discuss this. I don't want to be like, 'Oh you know, we haven't talked for 30 years, and here I am. I'm going to talk to you about my mutant cancer gene. No, I didn't want to do that.'"*.

3.5. Major theme 4. Experiences with the healthcare System

Varying experiences with healthcare systems were discussed by all women with *BRCA1/2* mutations. All made it a point to share different ways they received genetic testing/counseling: genetic counselors, PCPs, oncologists, gynecologists or direct-to-consumer testing (DTC, e.g., websites offering testing like Color and 23andMe). Although most women completed genetic counseling through a private licensed genetic counselor, PCPs (including gynecologists) and oncologists were also found to be common. Participants reflected on the quality of physician/counselor communication. Such experiences ranged from "sympathetic", "knowledgeable", and "conscientious" to "fear-inducing", "ignorant", "rushed", and "rude." Most positive experiences were with PCPs and DTC testing. The most negative experiences were with gynecologists and oncologists, as shown below.

> *"This technician was really like hell on wheels. I left that mammogram very aware of my own mortality like I'm going to die tomorrow. She took it upon herself to scare me with statements, telling me to immediately get a hysterectomy before age 40, a double mastectomy, and find an oncologist to get me on tamoxifen right now. She also told me to put all my paperwork [regarding BRCA] in a fireproof safe so that when I'm gone, the information doesn't die with me."*
>
> *"My genetic counselor got me in for active surveillance the day after my genetic test results – I kid you not – until I could get in to see a surgeon. She probably saved my life."*

Insurance-related issues were also identified regarding the genetic testing/counseling experience. The most widely cited issue was the limited amount of surveillance methods and genetic testing/counseling covered by healthcare insurance, despite women's *BRCA1/2*-positive status.

"I had to wait a year to get tested because my parents were in-between insurance coverages. I took the time to come up with a plan if I was positive. At the end, I was comfortable with my plan."

"At the time [I was going to be tested] I didn't have insurance. I didn't want to pay out-of-pocket to go to the doctor and have them tell me my results, so I contacted the company that ran my sample to get the results myself."

"At first, my mammogram and reconstructive surgeries weren't covered by insurance, but my gynecologist [who did the genetic testing] wrote a letter to the insurance company and called them until they approved my claim wasn't just 'cosmetic.' She went above and beyond to help me and take the time to help me."

A small group reported that some biannual surveillance methods were not covered by their health insurance, even though these individuals had tested positive for *BRCA1/2* mutations. One woman described having reconstruction surgery after a double mastectomy but was denied by insurance because she did not have active cancer.

3.6. Major theme 5. Preventive Concerns and Decisions

Women with *BRCA1/2* mutations discussed their prophylactic treatment options (i.e., specific surgeries and/or medications), and their plans if they received a positive *BRCA1/2* test result. Decisions for opting in or out of various prophylactic methods were strongly influenced by age, potential side effects, views of disfigurement, and/or potential for causing infertility. The influence of these factors sometimes outweighed prophylactic recommendations of providers. Even so, it became apparent that over time they would have to undergo all risk-reduction surgeries eventually to fully reduce their risks.

"*I was recommended to have an oophorectomy and then a bilateral mastectomy two days later. I chose not to because I'm so young, but I am starting colon and pancreatic cancer screenings because those run in my family too. Endoscopic ultrasounds twice or once per year don't sound fun*."

When considering recommendations for immediate prophylactic surgery such as double mastectomy, total hysterectomy, and/or bilateral salpingo-oophorectomy, the standard of care for women testing *BRCA1/2*-positive, depends on numerous factors including age, menopausal status, subtle differences between *BRCA1* and *BRCA2* risk, and cancer history. Participants explained they felt overwhelmed and needed time to reflect, however in the back of their minds, there was no question about having the surgery.

"I've had it in my head since day one that if I'm positive I'm going through all of the surgeries to give myself a fighting chance.".

"It's taking the bull by the horns…if it were just me, I might have a totally different approach, but I've got a six-year-old daughter and eight-year-old son, and I need to be around to help if either of them have to go through something like this. There's no question for me that getting [the surgery] done is the right thing. My initial response was that I didn't even blink as soon as my doctor told me what the results were, I said 'gut me and get them off.'"

Several women experienced both negative and positive reactions consecutively. Common words used to describe each were "disrespected and dehumanized" and "empowered and strength".

"I didn't want to go into menopause that way…I wanted to go naturally. At first, I was afraid of side effects from surgery, of a hysterectomy and so forth, then the whole thought of having a mastectomy - just felt like amputations."

"I've been very positive and vocal about my results and my own risk. I help other people this way. I don't think a lot of people know where to start and if they have someone who has had positive experiences and is willing to share information, it's empowering."

3.7. Major theme 6. Sources Affecting perceived risk

The last theme encompassed how *BRCA1/2*-positive women perceived the risk of breast and/or ovarian cancer diagnosis for themselves and family members. Perceived risk increased as the number of affected family members increased (e.g., multiple familial histories of cancer and having a personal history of cancer).

"I was diagnosed with breast cancer, my father had prostate and melanoma cancers, and his mother had ovarian cancer. I started to wonder if maybe this was relevant to me and my cancer risk."

"My mother had breast cancer twice; she's a survivor. Her sister had it twice, my aunt, but the second time it killed her. Their mother passed away from ovarian cancer at an early age, and I was starting to think there's something wrong with the women in my family."

However, for some of the women, sources affecting perceived risk seemed to be mitigated by active awareness and surveillance of cancer (e.g., active surveillance biannual mammograms, magnetic resonance imaging [MRI], gynecologic exams, and transvaginal ultrasounds). Additionally, some women felt less perceived risk of cancer when actively aware of their risk coupled with support from genetic counselors and oncologists.

"I do what I can to be proactive – I don't want the same fate. I especially don't want my children to have to see their mom go through cancer treatment if I have choices to prevent it."

"I've opted for preventive maintenance, like alternate between mammograms and MRIs every six or eight months in addition to tamoxifen to prevent cancer, since I've had it in the past."

4. Discussion

The purpose of the current study was to describe the lived experiences of women who have received genetic testing/counseling and tested positive for *BRCA1/2* mutations to better understand what it means to undergo genetic testing/counseling. Six main themes and two minor themes emerged, including: 1) Emotional Reactions to Genetic Results and Counseling, 2) Future Recommendations, 3) Family Solidarity and Support, 4) Experiences with the Healthcare System, 5) Preventive Concerns and Decisions, and 6) Sources Affecting Perceived Risk. Theme 1 contained two subthemes: (subtheme 1a: Pre-vivors; subtheme 1b: Testing Intuition).

This study extends previous research surrounding genetic testing/counseling experiences of women with *BRCA1/2* genetic mutations, contributing additional information pertaining to lived hardships, benefits, and overall insight into how women view the genetic testing/counseling process, including what comes next. As found in previous research, women noted how family and personal histories of cancer were significant sources affecting perceived cancer risk (Katapodi et al., 2004; Taber et al., 2015). However, more detail was provided surrounding the difficulties involved in prophylactic decision-making and disseminating genetic test results with other at-risk family members (Finch et al., 2011; Mau and Untch, 2017; Metcalfe et al., 2015; Saskova), possibly to begin cascade testing, or follow-up genetic testing for other at-risk biological family members. Specifically, those opting for ongoing surveillance reported pressure from medical professionals to undergo prophylactic surgeries. Although this immediacy was noted by Myklebust, Gjengedal, and Stromsvik (Myklebust et al., 2016), few studies have discussed surgery-related pressure by provider type (Caiata-Zufferey et al., 2015). This can be explained, in part, by reproductive age, as women of childbearing age would be more likely to undergo ongoing surveillance and after childbearing, complete prophylactic surgeries. Doctor-patient communication also surfaced in these decisions in combination with health insurance coverage (for genetic counseling itself and related surveillance and surgeries) and provided support services. The majority

of women reported that providers were not as empathetic and helpful as they could have been. Although this finding has been discussed in previous research (Dean, 2016; Dean et al., 2017), the current study expands on this by capturing varied experiences (positive, negative) and associated emotional reactions stratified by provider type (i.e., genetic counselor, PCP, gynecologist, oncologist).

When receiving a positive genetic test result, women's emotions ranged from extremely negative (e.g., depression/loss, anger, guilt, frustration) to extremely positive (e.g., empowerment and self-advocacy). Prior research has also found similar emotional ranges in this population (Hesse-Biber, 2018; Cicero et al., 2017; Gonzalez-Ramirez et al., 2017;15(13):28943989.; Oberguggenberger et al., 2016; Wenzel et al., 2012), mostly describing negative emotions, whereas the current study found they coexisted in relation to risk status (Peshkin et al., 2016). Also consistent with previous research, participants described themselves as "pre-vivors", not fitting into their own cancer-oriented category (patient or survivor) (Dean, 2016; Dean et al., 2017; Dean and Rauscher, 2017; Getachew-Smith et al., 2019; Herndl, 2014).

Hereditary mutations, such as *BRCA1/2* mutations, impact more than just the individual but family members as well, who can serve as support systems imperative for mental health (Mendes et al., 2010). However, the present research found that women voiced a need for assistance in disseminating results for the possibility of cascade testing, especially among family members they have lost touch with who may be at risk for *BRCA1/2* mutations themselves.

5. Clinical Recommendations for the future of genetic Testing/Counseling

Despite many improvements made thus far in the field of genetic testing/counseling, it is obvious that further advancements are necessary for the sustained care of *BRCA1/2*-positive women. This section outlines and expands on recommendations suggested by *BRCA1/2*-positive women within the current study to better provide and care for women testing for these mutations in the future.

The genetic testing/counseling field is changing rapidly, allowing medical and testing advancements to dominate over patient knowledge and awareness. Women in this study recommended genetic counseling procedural changes, such as the implementation of mandatory follow-up referral standards that could benefit all women undergoing genetic counseling in the future. These findings support previous literature, including access to genetic counselors (Markens, 2017), finding applicable reproductive options and specialists (Petrucelli et al., 2016; Rojas et al., 2019) and support groups (Dean and Rauscher, 2017), referrals for post-result family therapy (Godino et al., 2016), and scheduling follow-up genetic counseling appointments after results were presented to ask additional questions (Markens, 2017). Women specifically felt ongoing education and sensitivity training should be necessary for all who counsel *BRCA1/2*-positive women, what a positive test result truly means for women at different ages, how to properly (and accurately) reduce cancer risk, and how to present results to patients and families appropriately, as suggested by Roter and colleagues (Roter et al., 2008). Similarly, Mays and colleagues (Mays et al., 2012) conducted an intervention that offered personalized cancer risk assessments to low-income women with *BRCA1/2* mutations, finding that those who received assessments utilized preventive measures (e.g., surgery, surveillance, chemoprevention) more often than those who did not receive an assessment. Similar research has been conducted by Hanoch and colleagues (Hanoch et al., 2014), who found that by offering online training opportunities to genetic counselors on how to present test results, negative emotional reactions were reduced among patients.

The reception of genetic test results also mattered, and clinically, it may be beneficial for *BRCA1/2*-positive women to have a choice on how they receive results (i.e., in-person, email, mail, phone, webcam). It also may be beneficial for families to be included in these communicative processes, as identified in the current study and past literature (Elrick et al., 2017). Genetic counselors could offer to help disseminate test results to all family members deemed at-risk, so that no family member would be left unaware, and the burden of disclosure would not be placed on one individual. Montgomery and colleagues (Montgomery et al., 2014) offer a 3-step framework for providing information to individuals testing positive for *BRCA1/2* mutations on how they can disseminate test results to family members: 1) identify relatives who could benefit from the information; 2) choose a communication format (e.g., phone, in-person, email, text, web-based video); and 3) assess how much families might want (or do not want) to know. Findings here support testing family-based counseling approaches both during and after genetic counseling or providing additional guidance to individuals and families on how to disseminate such information as to mitigate negative emotional reactions. Similarly, clinicians could potentially use positive emotional aspects (e.g., empowerment, control) to support women through the common and expected negative emotions (Scherr et al., 2015).

The participants in this study believed that referrals to mental health professionals and therapists specifically trained to support women who test positive for *BRCA1/2* mutations should be mandatory and standard practice within genetic testing/counseling programs. Thus, the introduction of family therapy (Hoskins and Gotlieb, 2017) and other mental health resources (e.g., support groups, educational programs, mentor programs) (Landsbergen et al., 2010) may mitigate stress and anxiety (Hesse-Biber, 2018) after test results are given and when making preventive decisions.

Additionally, genetic counseling seemed to be limited geographically. Given the COVID-19 pandemic which began in 2019–20, the availability of tele-genetic counseling and other telehealth services have been increasing (Trepanier et al., 2020). Accessibility can be expanded by applying telemedicine or tele-genetic counseling that allow secure, video or text chats with counselors, which may be especially important for women living in rural areas of the US or those that cannot travel (Evans and Manchanda, 2020; Zhao et al., 2022). Recent tele-genetic counseling interventions have applied genetic counseling for *BRCA1/2*, finding that such applications were used mainly by younger individuals (<60 years) (Goldstein et al., 2018). Another manuscript currently under review described an intervention that occurred before and during the COVID-19 pandemic, called "Consultagene". Researchers created a virtual platform for tele-genetic counseling and associated education to connect patient questions to expert answers, finding that the on-demand aspect of the platform was favorable and convenient (Worley, et al., in press). These practices had the ability to reduce geographical barriers for those living in rural areas as well as provide an increasingly-available alternative option for patients (Zhao et al., 2022). Tele-genetic counseling is beneficial despite limitations, including challenges in establishing rapport, responding to verbal cues, and technical issues (Gorrie et al., 2021).

Due to the sensitive nature of genetic testing/counseling, resources and changes should be implemented to the genetic testing/counseling framework regarding individual and familial post-testing support. Resources should be provided to patients and at-risk family members to help understand their results. New online resources, such as AssessYourRisk.org (AssessYourRisk.org. Assessing your risk., 2022), have provided women with a personalized prevention plan, showing them their risk (average, increased, or high risk of breast/ovarian cancers), key risk factors, how to lower risk, and steps they can take to protect themselves in the future. Prophylactic decision-making has remained an important aspect of risk management. It is important to support women and their families during the decision-making process. Resources involving prophylactic decision-making tools may be beneficial; these could include online or in-person support groups that allow for open discussion of preventive decision-making, as suggested by Hoskins and Gotlieb (Hoskins and Gotlieb, 2017). Such decision-making tools have existed, as published by Jabaley, Underhill-Blazey, and Berry (Jabaley et al.,

2019) and Krassuski and colleagues (Krassuski et al., 2019) and are currently being tested via clinical trials by the NCI (Institute, 2022). They could be implemented within this population, when approved by the NCI or other governing body, to possibly ease decision-making processes. With additional information readily available to these populations, these individuals and their families can help manage cancer worry by having more accurate risk appraisals. Considering the emotional reaction that immediately follows a genetic test result, resources such as infographic tools outlining possible questions for genetic counselors for next steps may assist women in making decisions despite feeling overwhelmed. Similar to information published by the Illinois Department of Public Health (Illinois Department of Public Health. What to ask your genetic counselor., 2022), Facing Our Risk of Cancer Empowered (FORCE) (Risk, 2022) and beBRCAware.org (BeBRCAware.org. BRCA by the numbers., 2022), infographic tools could present possible questions and prompts for women who plan (or are) undergoing genetic testing/counseling or have tested positive for *BRCA1/2* mutations. Assistance for disseminating test results could be given in a brochure or interactive webpage format to provide examples of how to begin conversations with family members about *BRCA1/2* risk, as suggested by FORCE (Risk, 2022), BeBRCAware.org (BeBRCAware.org. BRCA by the numbers., 2022), and NCI (Institute, 2022) resource websites. Additionally, resources can include genetic testing-to-preventive care roadmaps which may have the ability to guide women from genetic testing/counseling to continuing care and beyond, even considering a cancer diagnosis. There are online resources that offer portions of roadmaps but are not continuous across types of care (e.g., testing/counseling, ongoing surveillance, prophylactic surgery, etc.). Therefore, more detailed interventions are needed to provide a tangible research-based roadmap to assist *BRCA1/2*-positive women navigate their care, possibly containing specialist and patient education, insurance and cost-related resources, and the implementation of family therapy or familial communication resources.

5.1. Future research implications

Future research should focus on understanding the qualitative, lived experiences and patterns outlined within the current study at a more in-depth level, with a larger, more representative sample. Research may also want to explore differences in preventive experiences by prophylactic decision and/or by surgery type to further inform resources for patients, their families, and doctors alike. Research could gain insight into understanding the role of intuition on perceived risk and subsequent psychosocial health outcomes. Although emotional reactions were not mutually exclusive, and could co-occur, it was imperative that medical professionals and researchers working with and researching this population, be aware of this distinction. Thus, effective doctor-patient communication requires further understanding, focusing on the emotional needs of women who test positive for these mutations. Physically and psychosocially, genetic testing for *BRCA1/2* mutations impacts the lives of at-risk women, in addition to a variety of other groups such as family members, health providers, and policymakers.

5.2. Limitations

It is important to interpret these findings considering its limitations. Recall bias may be an issue, as participants were asked to recall information up to five years earlier regarding their genetic testing/counseling experience. Although being tested for *BRCA1/2* mutations is becoming more common, the number of women who test positive remains relatively limited, so a widened five-year limit was set to ensure data saturation was met. Generalization is limited due to the number of participants interviewed, specifically educated, mostly insured, non-Hispanic white women are more commonly tested for *BRCA1/2* genetic mutations within the US. Also, generalizability could be limited to women who participate in support groups, and in this case, online support groups. These participants may introduce bias by being more open and willing to share experiences than others not in support groups, Future research should replicate this study using a larger, more representative, and diverse population. Lastly, follow-up interviews could have been included so that researchers could gain clarification and/or ask additional questions that surfaced during the analyses.

6. Conclusions

This study provides novel and insightful information about women's experiences during and after the genetic testing/counseling process. Several themes emerged, some new and some consistent with previous literature. Women discussed the importance of family in the counseling process as well as identifying areas that could use improvement. Recommendations involving post-counseling referrals (e.g., oncology/surgical, support groups, behavioral therapy), additional provider education, follow-up genetic appointments, and risk assessments can serve to inform future intervention programs and future research. Although genetic testing/counseling may only be conducted for a minority of the general population, it is becoming increasingly popular and widely available in the US for many hereditary cancers. These results can utilized to inform future research and interventions on a greater scale as well as increasing patient/physician care education, especially for those with mutations associated with predispositions to other cancers.

CRediT authorship contribution statement

Kate E. Dibble: Conceptualization, Methodology, Formal analysis, Investigation, Resources, Writing – original draft, Writing – review & editing, Visualization, Project administration, Funding acquisition. **Laura K.M. Donorfio:** Conceptualization, Methodology, Formal analysis, Resources, Writing – original draft, Writing – review & editing, Visualization, Supervision. **Preston A. Britner:** Methodology, Resources, Writing – original draft, Writing – review & editing. **Keith M. Bellizzi:** Conceptualization, Methodology, Resources, Writing – original draft, Writing – review & editing, Supervision, Funding acquisition.

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Appendix A. Supplementary material

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