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Comparing Family Sharing Behaviors in *BRCA* Carriers with *PALB2* Carriers

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## Table of Contents

List of Tables .....	iii
List of Figures .....	iv
Abstract .....	v
Introduction .....	1
Methods .....	6
Participants .....	6
Instrumentation .....	6
Procedures .....	9
Data Analysis .....	10
Results .....	12
Participant Characteristics .....	12
Themes Related to Family Sharing .....	12
Attitudes .....	14
Normative Influence .....	17
Perceived Control and Environmental Constraints .....	18
Self-Efficacy .....	20
Knowledge and Skills .....	23
Salience .....	25
Intention and Decision .....	27
Barriers and Facilitators to Family Sharing .....	29
Discussion .....	31
Theme: Health Protection .....	32
Theme: Anticipated Negative Emotions from Family Members .....	32
Theme: Family Reactions Range from Supportive to Not Supportive .....	33
Theme: Normative Influence from Providers and Family .....	33
Theme: Strong Family Relationships .....	33
Theme: Lack of Contact and Communication Barriers .....	34
Theme: Impact of Public Knowledge and Awareness of <i>BRCA</i> .....	34
Theme: Confident in Sharing Basics .....	35
Theme: Informational Resources Boost Self-Efficacy .....	36
Theme: Knowing What to Share and How to Share .....	37
Theme: Important When Risks Are High, Actionable and Relative is Prepared .....	37
Theme: High Rates of Family Sharing and Intention to Share .....	38

Study Limitations .....	39
Practice Implications .....	41
Research Recommendations .....	42
Conclusions .....	44
References .....	45

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## List of Tables

Table 1: Targeted IBM-Related Constructs .....	8
Table 2: Participant Demographics .....	13
Table 3: Theme: Health Protection .....	14
Table 4: Theme: Anticipated Negative Emotions from Family Members .....	15
Table 5: Theme: Family Reactions Range from Supportive to Not Supportive .....	16
Table 6: Theme: Normative Influence from Providers and Family .....	17
Table 7: Theme: Strong Family Relationships .....	19
Table 8: Theme: Lack of Contact and Communication Barriers .....	19
Table 9: Theme: Impact of Public Knowledge and Awareness of <i>BRCA</i> .....	20
Table 10: Theme: Confident in Sharing Basics .....	21
Table 11: Theme: Informational Resources Boost Self-Efficacy .....	22
Table 12: Theme: Knowing What to Share and How to Share .....	24
Table 13: Theme: Important When Risks Are High, Actionable and Relative is Prepared .....	26
Table 14: Theme: High Rates of Sharing and Intention to Share .....	29

## List of Figures

Figure 1: The Integrated Behavioral Model .....	7
Figure 2: Family Sharing for <i>BRCA</i> and <i>PALB2</i> Carriers Using IBM Framework .....	31
Figure 3: Practice Implications for Providers Flowchart .....	42

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## Abstract

Identifying individuals with hereditary cancer predisposition can improve health outcomes for patients and their family members through early cancer detection and prevention strategies. Prior research about family sharing of genetic test results among those with hereditary breast cancer has overwhelmingly been limited to the *BRCA1* and *BRCA2* genes. The present study sought to compare family sharing behaviors in women with pathogenic *BRCA* variants to women with pathogenic variants in the more recently identified and characterized *PALB2* gene. A total of 18 *BRCA* carriers and 13 *PALB2* carriers were interviewed about family sharing practices using a semi-structured guide based on the Integrated Behavioral Model. Barriers and facilitators to family sharing were similar for both *BRCA* and *PALB2* carriers, with logistical difficulties and emotional struggles related to anticipated negative reactions from relatives being the most salient barriers. The most important facilitators were: attitude that sharing enables health protection, provider recommendation, strong family relationships, confidence in sharing basic information, knowledge of what to share and how to share, and belief that sharing is highly important. Given similar attitudes, norms, and control beliefs related to family sharing, similar, but tailored interventions may be effective at increasing family disclosures among both groups. Such interventions should involve a discussion of patients' attitudes towards sharing with healthcare providers to strengthen motivations and address barriers and provision of informational resources to increase confidence and knowledge. Family sharing resources should clearly specify which relatives need to be informed, why sharing is important, and how at-risk relatives may benefit.

## Introduction

Breast cancer is the second most prevalent cancer in women with over 260,000 estimated new cases in the United States in 2018 (Surveillance, Epidemiology, and End Results Program, National Cancer Institute, 2018). Roughly 5-10% of female breast cancer patients have an inherited predisposition, most commonly due to *BRCA1* or *BRCA2* (American Cancer Society, 2017). The lifetime risk to develop breast cancer for women with pathogenic *BRCA* variants is 60-70% (Kuchenbaecker et al., 2017), compared to a 12.4% lifetime risk for average women (Surveillance, Epidemiology, and End Results Program, National Cancer Institute, 2018). In addition to *BRCA1* and *BRCA2*, there are other highly penetrant genes that also confer increased risks for breast cancer. The gene *PALB2* (partner and localizer of *BRCA2*) is estimated to account for 1-3% of hereditary breast cancers (Antoniou et al., 2014; Casadei et al., 2011; Couch et al., 2015; Cybulski et al., 2015; Thompson et al., 2016), with lifetime breast cancer risks ranging 33-58%, modified by family history of breast cancer (Antoniou et al., 2014; Couch et al., 2017).

Next-generation sequencing and use of multi-gene panels has reduced costs and increased efficiency of clinical genetic testing for hereditary breast cancer, thereby increasing identification of high-risk individuals – particularly those with pathogenic variants in breast cancer genes besides *BRCA* (Antoniou et al., 2014; Ricker et al., 2018). Identifying hereditary predisposition to breast cancer is an important step for enabling early detection, prevention, and risk management strategies and for guiding cancer treatment (Black, McClellan, Avar, & Knoppers, 2013; Katapodi, Northouse, Milliron, Liu, & Merajver, 2013; Ricker et al., 2018). Given the higher risk and earlier onset of disease, women with pathogenic variants in *BRCA1*, *BRCA2*,

*PALB2*, and other breast cancer genes are eligible for increased surveillance and other preventive measures starting at younger ages (Couch et al., 2017; Ricker et al., 2018). According to national guidelines, high-risk screening and/or consideration of risk-reducing options may begin as early as age 25 (sometimes younger) for *BRCA* carriers and age 30 (sometimes younger) for *PALB2* carriers (National Comprehensive Cancer Network, 2018). High-risk screening can diagnose breast cancer at an earlier, more treatable stage, thus prolonging survival, whereas prophylactic mastectomy and salpingo-oophorectomy (when appropriate) can effectively reduce breast cancer risks (Domchek et al., 2010; Lauby-Secretan et al., 2015; Nelson et al., 2014). For mutation carriers who have already developed cancer, the benefit of identifying hereditary predisposition is focused on preventing a second primary cancer and informing treatment decisions (Ricker et al., 2018).

Identification of women with a cancer-predisposing variant confers health implications for their family members, as well (McCarthy & Armstrong, 2014; Nelson et al., 2014). Relatives of a *BRCA* or *PALB2* carrier may have up to a 50% chance to harbor the same gene mutation and associated cancer risks (Antoniou et al., 2014; Cheung, Olson, Yu, Han, & Beattie, 2010). For this reason, women found to carry a pathogenic variant in one of these cancer-predisposing genes are encouraged to notify their relatives of the result and the availability of genetic testing and risk management (Dancyger, Smith, Jacobs, Wallace, & Michie, 2010). Relatives who choose to pursue genetic testing for themselves may be able to clarify their own cancer risks and determine optimal risk management strategies (Daly, Montgomery, Bingler, & Ruth, 2016; Fehniger, Lin, Beattie, Joseph, & Kaplan, 2013; Katapodi et al., 2017). If relatives are determined to have inherited cancer predisposition, they can then execute health protective behaviors that may reduce breast cancer morbidity and mortality.

Family sharing (also called family communication or intra-familial communication or disclosure in the literature) is a complex yet critical step within the cancer control continuum (Daly et al., 2016; Derbez, 2018; Division of Cancer Control and Population Sciences, National Cancer Institute, 2017; Peters et al., 2011). Currently, it is the responsibility of the individual tested to notify their relatives of any potential risks (Daly, 2015; Kardashian, Fehniger, Creasman, Cheung, & Beattie, 2012). Studies have shown that rates of family sharing among *BRCA* carriers are relatively high, ranging from approximately 73% to 96% (Daly et al., 2016; Fehniger et al., 2013; Finlay et al., 2008; Hughes et al., 2002; Ricker et al., 2018). The most important reasons cited for sharing genetic results include making relatives aware of risk, suggesting they undergo genetic testing, and fulfilling a perceived responsibility to inform (Hughes et al., 2002; McGivern et al., 2004). Seeking emotional support and advice about management decisions have also been reported as motivators for sharing (Hamilton, Bowers, & Williams, 2005; Hughes et al., 2002). Despite the importance of sharing, rates of disclosure previously reported indicate that some at-risk relatives remain uninformed and unaware of potential cancer risks (Black et al., 2013; Daly et al., 2016). Furthermore, testing rates among family members remain low, ranging from roughly 15-50%, even when results are shared (Blandy, Chabal, Stoppa-Lyonnet, & Julian-Reynier, 2003; Lieberman et al., 2018).

The relationship between family sharing and various individual, familial, and sociocultural factors has been documented regarding disclosure of *BRCA* results (Nycum, Avard, & Knoppers, 2009). Personal feelings and perceptions of risk, relatives' attitudes, knowledge, and finding "the right time" may impact the decision to share genetic results with family members (Blandy et al., 2003; Cheung et al., 2010; Daly et al., 2016; Dean & Rauscher, 2018; Derbez, 2018; Hamilton et al., 2005; Lafrenière, Bouchard, Godard, Simard, & Dorval, 2013;

Lapointe et al., 2013; Nycum et al., 2009). Prior studies have shown that first-degree family members are most likely to receive genetic risk information, suggesting that more distant family members who may also benefit are often excluded (Blandy et al., 2003; Elrick et al., 2017; Kardashian et al., 2012; Katapodi et al., 2013; MacDonald et al., 2007; McGivern et al., 2004). Family communication styles, traditions, religious beliefs, and norms have also been shown to influence the decision to share (Etchegary, Potter, Perrier, & Wilson, 2013; Katapodi et al., 2013; Koehly et al., 2009; Lafrenière et al., 2013; Peters et al., 2011). Minimal contact and/or emotionally distant relationships with relatives have also been implicated as barriers to disclosure of results (Daly et al., 2016; Elrick et al., 2017; Etchegary et al., 2013; Hughes et al., 2002; Kardashian et al., 2012; MacDonald et al., 2007; McGivern et al., 2004). Those with a strong family history of *BRCA*-related cancers are more likely to share compared to families with a less striking history of cancer (Dean & Rauscher, 2018; Kardashian et al., 2012). Women are more likely to communicate genetic information than men, and information is more often communicated to female relatives and younger generations (Cheung et al., 2010; Elrick et al., 2017; Etchegary et al., 2013; Finlay et al., 2008; Kardashian et al., 2012; MacDonald et al., 2007; Patenaude et al., 2006; Vadaparampil, Malo, de la Cruz, & Christie, 2012).

Previous research about family sharing related to hereditary breast cancer has focused almost exclusively on disclosure of *BRCA1* and *BRCA2* results (D' Audiffret Van Haecke & de Montgolfier, 2016; Ricker et al., 2018). Ricker et al. (2018) is the only published study to explore family sharing among those with a gene mutation in other hereditary breast cancer genes (though not exclusively hereditary breast cancer genes); however, they did not assess for barriers and facilitators related to family sharing. Rather, a survey containing a combination of “yes/no” and open-ended questions was utilized to measure rates of communication of genetic test results

and family follow-up and a single Likert scale question was used to measure attitude about the benefit of family sharing. *PALB2* carriers were included in the Ricker et al. (2018) study, though the number of *PALB2* participants was not specified.

Given the limited data on family sharing and hereditary breast cancer beyond *BRCA*, the present study sought to further our understanding of family sharing among women with pathogenic *PALB2* variants compared to women with pathogenic *BRCA* variants. It is unclear whether different gene carriers experience unique barriers and facilitators with family sharing and require different approaches to improve rates and quality of family sharing. Eliciting and comparing disclosure behaviors in these two groups is therefore a critical first step in identifying potentially modifiable factors that may serve as effective targets for interventions for *PALB2* carriers, as well as assessing the applicability of *BRCA*-related interventions (Cheung et al., 2010; Elrick et al., 2017). Addressing the most salient barriers and facilitators will be necessary to increase rates of family sharing, enable more at-risk individuals to be proactive in cancer risk management, and ensure that all have the opportunity to benefit from genetic testing. The current study utilized qualitative methods to capture more in-depth and comprehensive data underlying the motivators and barriers to family sharing, as this was not captured as part of the survey conducted by Ricker et al. (2018) (Bradley, Curry, & Devers, 2007).

## Methods

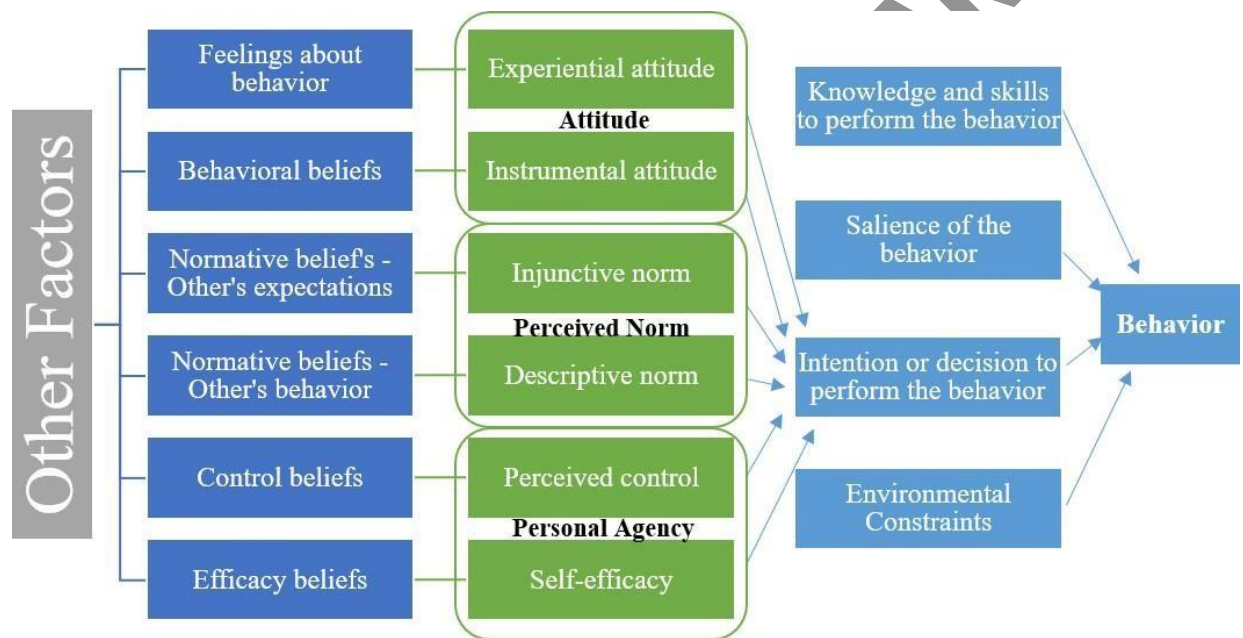
### *Participants*

Participants were recruited by a research team at Vanderbilt University from a group consented to the GeneCARE study. GeneCARE participants were English-speaking females, 18 years or older, and living in the United States with a documented pathogenic/likely pathogenic variant or variant of uncertain significance (VUS) in a gene associated with hereditary cancer. The current study was limited to women enrolled in GeneCARE with a *BRCA1*, *BRCA2*, or *PALB2* pathogenic/likely pathogenic variant who indicated willingness to take part in an in-depth telephone interview. All *PALB2* carriers and a subset of *BRCA* carriers who met this criteria were purposively selected for interviews in order to maximize diversity in family sharing and medical management practices. We aimed to conduct 10 interviews for each carrier group according to recommendations for achieving theoretical saturation, or the point at which no new themes are emerging. Prior studies have found that small sample sizes ranging from 10 to 12 participants can be sufficient for collecting most of the salient ideas and reaching saturation (Guest, Bunce, & Johnson, 2006; Saunders et al., 2018; Weller et al., 2018). The study was approved by Institutional Review Boards at Vanderbilt University and the University of South Florida.

### *Instrumentation*

The study team developed a semi-structured interview guide (Appendix A) based on the Integrated Behavioral Model (IBM) (Figure 1), which proposes that five main constructs directly influence behavior and intention is the most important determinant (Montano & Kasprzyk, 1992). The IBM framework is an extension of the Theory of Planned Behavior (TPB), which has

been utilized in previous studies to understand family communication of genetic risk (Montgomery et al., 2013; Wiens, Wilson, Honeywell, & Etchegary, 2013). Both theories state that behavioral intention is the product of attitudinal, normative, and control beliefs; however, the IBM incorporates knowledge and skills, salience, and environmental constraints as behavioral modifiers. The IBM was chosen as the framework for this study as these additional constructs may play an important role in the family sharing process.



**Figure 1.** The Integrated Behavioral Model.

An essential step in applying the IBM is conducting interviews with the population of interest to elicit information about their beliefs (Montano & Kasprzyk, 1992). The interview guide was designed to elicit seven IBM-related domains we thought would reveal underlying differences and similarities in the family sharing behaviors of *BRCA* carriers and *PALB2* carriers (Table 1). These domains included: 1) attitudes, 2) normative influences, 3) perceived control



and environmental constraints, 4) self-efficacy, 5) knowledge and skills, 6) salience, and 7) intention or decision.

**Table 1.** Targeted IBM-Related Constructs

Construct	Definition	Example Questions to Elicit Construct
Attitudes	Emotional response to the idea of sharing, beliefs about the anticipated or actual outcomes of sharing	<ul style="list-style-type: none"> <li>• How did you feel about the idea of sharing your genetic test result with family members?</li> <li>• Were there any benefits of sharing?</li> <li>• Were there any negative effects of sharing?</li> </ul>
Normative Influences	Social pressures to share or not share results with family	<ul style="list-style-type: none"> <li>• Did a healthcare provider encourage you to share your result with family?</li> <li>• Who would support you sharing your result?</li> <li>• Did some relatives not want to hear about your result?</li> </ul>
Perceived Control & Environmental Constraints	How easy or difficult it is to share test results with family and environmental conditions that might prevent sharing	<ul style="list-style-type: none"> <li>• What made it easy for you to share your genetic test result?</li> <li>• What made it hard for you to share your genetic test result?</li> </ul>
Self-Efficacy	Confidence and effectiveness in sharing	<ul style="list-style-type: none"> <li>• On a scale of 1-10, how confident were you sharing your result with family?</li> <li>• Which family members did you feel most confident sharing with?</li> <li>• What types of resources do you think could be helpful?</li> </ul>
Knowledge & Skills	Possessing the knowledge and skills to communicate results to family and convey the value of genetic testing	<ul style="list-style-type: none"> <li>• Describe for me how you shared you result with family?</li> <li>• What information did you tell your family about your result?</li> </ul>

**Table 1** (continued)

<b>Construct</b>	<b>Definition</b>	<b>Example Questions to Elicit Construct</b>
Salience	Beliefs about how important it is to share results	<ul style="list-style-type: none"><li>• When someone tests positive for a cancer gene mutation, on a scale of 1-10 how important is sharing that result with family?</li></ul>
Intention or Decision	Indication of readiness or decision to share or not share results with family	<ul style="list-style-type: none"><li>• Which family members did you talk to about your genetic test result?</li><li>• Do you intend to share your result with this family member in the future?</li><li>• Did you tell any non-relatives about your result?</li></ul>

### *Procedures*

Baseline demographic and clinical data were available on all participants through completion of the GeneCARE survey. Participants provided informed consent to the interview at the time of enrollment in the survey portion of GeneCARE and consent was confirmed verbally prior to audio-recording each respective interview. The semi-structured interview guide was used to assess their initial reaction to their genetic test result, information about medical management decisions (results of which are not within the scope of the current study), and what they perceived to be facilitators and barriers to sharing their result with various family members. The discussion focused on the at-risk side of the family, if that could be determined based on the family history. Otherwise, both sides were considered at-risk and data for both sides of the family were obtained. Each interview lasted approximately 30-60 minutes.

Interviews were conducted by 2 investigators trained in human subjects' protection. Audio recordings were transcribed verbatim and memos were created by the interviewer after each interview to document important themes, memorable quotes, or striking observations.

Memos and transcripts of the first several interviews were analyzed to assess the need for additional questions, revisions to the guide, and additional codes.

### *Data Analysis*

A codebook was developed by three of the researchers based on the interview guide and a single coder analyzed each transcript using RQDA qualitative data analysis software. Data analysis utilized a thematic approach, with steps related to data immersion, generating codes, and identifying, reviewing, and defining themes (Nowell, Norris, White, & Moules, 2017). Prior to coding, transcripts from the first several interviews were reviewed in-depth in order to become familiar with the data (Bradley et al., 2007). Transcripts were then coded line-by-line using theory driven a priori codes and inductive, data-driven thematic codes developed through an iterative process (Tracy, 2012; Tracy & Hinrichs, 2017).

The following codes were added during initial analysis of the interview transcripts: *ATT\_fam positive*; *ATT\_fam negative*, *ATT\_fam other*; *KNOW\_information*; and *DEC\_not shared non-family*. Furthermore, the following codes were anticipated but subsequently deleted from the codebook after initial analysis: *ATT\_ignore*; *KNOW\_risks to family*; and *KNOW\_risks and benefits*. All transcripts were then re-analyzed to ensure that a priori and data-driven codes were utilized appropriately. Interviews were classified using a total of 31 codes within seven theoretical constructs. The final codebook can be found in Appendix B.

Through coding, sorting, and review of the data, the most salient themes regarding family sharing were identified and interpreted in the context of the IBM framework. Particular attention was paid to items mentioned in one carrier group, but not the other. Illustrative quotes were selected to accompany each theme related to factors that facilitate or inhibit sharing of genetic

test results with family members. Demographic and clinical characteristics of the study participants were summarized using descriptive statistics.

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## Results

### *Participant Characteristics*

A total of 168 *BRCA* carriers and 22 *PALB2* carriers who completed the GeneCARE survey expressed interest in participating in the in-depth interviews. Eighteen *BRCA* carriers and thirteen *PALB2* carriers were ultimately interviewed for this study. Additional participants were recruited beyond the original target sample size to ensure that at least ten interviews for each group were completed. These additional participants were recruited using the same methods discussed previously.

Participant demographic and clinical characteristics are shown in Table 2. *BRCA* carriers and *PALB2* carriers had a mean age of 53.67 and 55.62 years, respectively. The majority of carriers in both groups self-identified as Non-Hispanic White. Most of the *BRCA* participants and all of the *PALB2* participants reported themselves as college graduates. Approximately 72% of the *BRCA* carriers and 85% of the *PALB2* carriers reported having private insurance. 13 *BRCA* carriers and nearly all *PALB2* carriers had a personal history of cancer. While almost all *PALB2* participants were the first member of their family to be genetically tested, the majority of *BRCA* carriers were uncertain if other relatives had tested first.

### *Themes Related to Family Sharing*

In-depth interviews with *BRCA* carriers and *PALB2* carriers revealed twelve major themes related to family sharing. Themes were organized into seven IBM-related theoretical constructs and are described according to construct in detail below. The following themes emerged within the 'attitudes' construct: health protection, anticipated negative emotions from

**Table 2.** Participant Demographics

	<i>BRCA1/2</i> n = 18	<i>PALB2</i> n = 13
Age, years		
Mean (Range)	53.67 (30 – 71)	55.62 (39 – 69)
Race/Ethnicity, n (%)		
NHW	8 (44.4)	12 (92.3)
Black	4 (22.2)	1 (7.7)
Hispanic	6 (33.3)	0
Highest Completed Education, n (%)		
≤ 12 <sup>th</sup> grade/GED	2 (11.1)	0
Vocational School/Some College	3 (16.7)	0
College Graduate	13 (72.2)	13 (100)
Other	0	0
Insurance, n (%)		
Private	13 (72.2)	11 (84.6)
Military/Veteran	2 (11.1)	1 (7.7)
Medicare	2 (11.1)	1 (7.7)
Medicaid	1 (5.6)	0
Personal History of Cancer, n (%)		
None	5 (27.8)	1 (7.7)
Breast	12 (66.7)	12 (92.3)
Breast and Ovarian	1 (5.6)	0
First Family Member Tested, n (%)		
No	1 (5.6)	1 (7.7)
Yes	4 (22.2)	12 (92.3)
Unknown	13 (72.2)	0

family members, and family reactions range from supportive to not supportive. The ‘perceived norms’ construct revealed normative influence from providers and family. Themes within the ‘perceived control and environmental constraints’ construct included strong family relationships, lack of contact and communication barriers, and impact of public knowledge and awareness of *BRCA*. Themes related to the ‘self-efficacy’ construct were confidence in sharing basics and informational resources boost self-efficacy. In terms of knowledge and skills, participants knew what to share and how to share. The ‘salience’ construct revealed the theme that sharing is important when risks are high, actionable and the relative is prepared. Finally, the ‘intention and decision’ construct showed high rates of sharing and intention to share.

## Attitudes

Participant attitudes towards family sharing were divided into positive and negative attitudes. Both *BRCA* and *PALB2* carriers felt strongly that sharing their positive genetic test result would protect the health of their family members via follow-up genetic testing and/or increased cancer surveillance. Both groups frequently quoted the saying “knowledge is power” to succinctly describe why they felt positively about sharing. It was apparent that participants viewed sharing as a way to protect not only their living relatives, but also future generations.

**Table 3.** Theme: Health Protection

Brief description	Illustrative quotes
Participants felt that sharing would allow relatives to be proactive in their own cancer risk management	“I think information is power, because once you have the information you can make better decisions. So I wanted them to have the necessary information for them to make decisions for themselves and their families.” ( <i>PALB2</i> carrier, age 63)
	“I wanted to make sure if anybody out there has the gene, they needed to know about it so they could take whatever precautions were necessary so that they didn’t get breast cancer. I felt sort of empowered to get this information to them and make sure that they protected their health.” ( <i>PALB2</i> carrier, age 60)
	“I feel like that if you know your chances are better to get cancer, then you can do something about it before it happens.” ( <i>BRCA</i> carrier, age 69)
	“I wanted them to know so that they have the option of testing. That they would know it is available and make the decision... if you just don’t know that you have it, things can happen in the future, and if you do know then you can do stuff to prevent it.” ( <i>BRCA</i> carrier, age 71)

Women in both the *BRCA* and *PALB2* groups were concerned about how their family members might respond when learning about their positive genetic test result. The two groups

acknowledged that sharing may cause family members to experience a variety of negative emotions, including fear, worry, distress, and guilt for having passed down the mutation. A few participants were uncertain if their family members would have access to knowledgeable providers or recommended follow-up care after learning about their risk. Interestingly, only a single participant in each carrier group was hesitant to share their result with family due to concerns for privacy.

**Table 4.** Theme: Anticipated Negative Emotions from Family Members

Brief description	Illustrative quotes
Participants felt that sharing their result could cause family members to feel scared, worried, and overwhelmed	<p>“I was concerned because, like I said, I know it’s going to be stressful for them. They’re going to have to make their decision as to what they want to do with this information.” (<i>PALB2</i> carrier, age 57)</p> <p>“I was sad, and then obviously people I told are gonna be sad...it’s a scary thing to learn, to know you could carry this gene.” (<i>PALB2</i> carrier, age 49)</p> <p>“Fearful of putting an element of fear about that person’s health in their head. You know, possibly making them fearful of dying from ovarian cancer more so than breast cancer.” (<i>BRCA</i> carrier, age 52)</p> <p>“If my mother were still alive I may have been more hesitant for her to know because I would think, knowing her personality, she would feel guilty for having passed this along to us.” (<i>BRCA</i> carrier, age 59)</p>

Despite participants’ concerns about negative emotional reactions, participants found that many family members reacted positively when learning about the positive genetic testresult. Relatives were reported as being supportive, grateful, receptive, and not surprised by the information. Nonetheless, some family members did not show interest in learning about the test result and, as anticipated by participants, certain relatives became worried or scared. Several



family members reportedly ignored the information. Multiple *BRCA* carriers stated their family members were in denial, confused, or did not fully understand the result.

**Table 5.** Theme: Family Reactions Range from Supportive to Not Supportive

Brief description	Illustrative quotes
The reactions of family members ranged from positive/supportive to negative/not supportive	<p data-bbox="623 527 1427 632">“I had one uncle who said, “You are very brave.” I didn’t get any negative. I only got positive, “Yeah thanks for letting us know” kind of thing.” (<i>PALB2</i> carrier, age 63).</p> <p data-bbox="623 667 1427 814">“I don’t think they were that surprised, because like I said, the breast cancer has been running in the family, and my sister had the ovarian cancer, so it wasn’t like totally out of the blue.” (<i>PALB2</i> carrier, age 57)</p> <p data-bbox="623 850 1427 955">“One of my sisters told me to mind my own business. Her health is her prerogative, and I should mind my own business. That was very unexpected.” (<i>PALB2</i> carrier, age 59)</p> <p data-bbox="623 991 1427 1064">“Two out of three [siblings] were glad. One wasn’t...she was more like, “I wish I didn’t know.” (<i>PALB2</i> carrier, age 60)</p> <p data-bbox="623 1100 1427 1247">“I think my sisters were more supportive, because they have daughters too, and the breast cancer runs in women more so my sisters were probably the most supportive and the most interested in it.” (<i>BRCA</i> carrier, age 64)</p> <p data-bbox="623 1283 1427 1430">“A lot of them were happy that they were given the information, but it was one of those things, “thanks for giving me the information” but they didn’t really follow up on it.” (<i>BRCA</i> carrier, age 49)</p> <p data-bbox="623 1465 1427 1581">“Well, with my sister she just said...”I’m not doing this, I’m not dealing with this, if I’m meant to die from breast cancer or ovarian cancer then I will.” (<i>BRCA</i> carrier, age 52)</p> <p data-bbox="623 1617 1427 1791">“His attitude is kinda like, it doesn’t affect me right now, I don’t really care. But he’s also, he’s very much one to be in denial and that’s his personality. He’d rather not know the “I don’t have to think about it” kind of thing.” (<i>BRCA</i> carrier, age 59)</p>

## Normative Influence

Healthcare providers had the most significant normative influence for participants with regards to family sharing. The majority of these providers were genetic counselors, but other genetics professionals (e.g. geneticist, genetics nurse), oncologists, and surgeons were also mentioned as encouraging family sharing as part of recommended follow-up. All but a few participants recalled a specific conversation when their provider encouraged them to share their result with close and extended relatives on the at-risk side of the family, if that could be determined. In terms of familial influence, most participants expected their family members would want to know about their positive genetic test result. However, participants in both groups expressed concerns that certain family members would not be receptive to this information or they would not understand the significance. Only one *PALB2* participant (age 65) mentioned her religious upbringing and “Catholic guilt” as a source of pressure to share.

**Table 6.** Theme: Normative Influence from Providers and Family

Brief description	Illustrative quotes
Most participants were encouraged by a healthcare provider to share their positive genetic test result with at-risk relatives.	[Were there any recommendations the genetic counselor gave you that you hadn't done at this point?] “Yeah, there was nothing else for me to do outside of just talk with your family, just to let them know, to share what my diagnosis [test result] was.” ( <i>PALB2</i> carrier, age 49)  [What were some of the things that your breast surgeon told you to do because you have this <i>BRCA</i> result?] “First thing was to get the other breast removed...second thing was to get my ovaries removed...and then tell like my family, so that they can also get tested.” ( <i>BRCA</i> carrier, age 60)
Many participants felt their families would be supportive of sharing, though some expected their families would not be supportive.	“I pretty much knew my cousins that I shared it with, that they would appreciate it, so I didn't feel like I was telling them anything they would not appreciate knowing.” ( <i>PALB2</i> carrier, age 69)

**Table 6** (continued)

Brief description	Illustrative quotes
Many participants felt their families would be supportive of sharing, though some expected their families would not be supportive.	“I knew they were gonna be supportive. I knew nobody was gonna question my decision. I didn’t feel anybody was gonna not believe me. They’re a very rational, reasonable, supportive bunch of people. So I just knew I could share it with them.” ( <i>PALB2</i> carrier, age 60)
	“I’m afraid though, because I feel like they might be the type to say, “What is that? Mind your own business.”” ( <i>PALB2</i> carrier, age 59)
	“[My sister-in-law] had asked me to do genetic testing. They were waiting for results to see if my brother should get tested.” ( <i>BRCA</i> carrier, age 64)
	“I would like to sit down with her [niece] and talk to her about it but other family members say, “No, don’t.” There’s a divide in the family...because she’s getting married next year and it might make her feel less of...that she might feel like damage goods.” ( <i>BRCA</i> carrier, 59)
	“I didn’t expect much a reaction from them. They didn’t have much of a reaction...when it comes to cancer we all kind of share anyway.” ( <i>BRCA</i> carrier, age 38)

#### *Perceived Control and Environmental Constraints*

When asked specifically about what made it easier to share genetic results with family, participants in both the *BRCA* and *PALB2* groups endorsed strong familial relationships. Open family communication styles and frequent contact made the task of sharing less daunting and more convenient. Additionally, it was helpful if relatives had prior knowledge of a participant’s personal cancer diagnosis and/or the family history of cancer. Help from other family members by communicating to other relatives, sharing contact information, or even initiating the sharing process made it easier for many participants.

**Table 7.** Theme: Strong Family Relationships

Brief description	Illustrative quotes
Participants with strong family ties found it easier to share their result with at-risk relatives	“I was very open with my family about everything, my treatments and everything, so they were already aware of what I was going through and what I was having done. So, I guess that made it easier because it wasn’t like I was calling them out of the blue and telling them that I have this. They already knew.” ( <i>PALB2</i> carrier, age 51)
	“We’re a pretty close bunch, so I had full access to them... there’s definitely open lines of communication.” ( <i>BRCA</i> carrier, age 49)
	“My dad has some nieces that we don’t talk to directly but we know how to get ahold of them indirectly through mutual family members and friends.” ( <i>BRCA</i> carrier, age 52)

Conversely, participants in both groups felt that distant relationships with relatives and difficulty contacting family members made it harder to share. A *BRCA* carrier mentioned it was difficult for her to share with her family members in a different country due to lack of resources in their native language. Only a single participant in the *PALB2* group cited relatives’ education level as a source of hardship in sharing.

**Table 8.** Theme: Lack of Contact and Communication Barriers

Brief description	Illustrative quotes
Participants had to overcome estranged relationships and communication barriers to share with certain at-risk relatives.	“Everything was difficult. I had to find them... because you lose touch with people.” ( <i>PALB2</i> carrier, age 65)
	“I don’t communicate with them very often, so it’s not like it was purposely done. It just didn’t happen.” ( <i>PALB2</i> carrier, age 48)
	“I’m just not that close with them... we just don’t see each other that often and we don’t really share information that personal.” ( <i>BRCA</i> carrier, age 30)
	“The problem is that a lot of the information is English and not all of them speak English, so that made it a little bit harder.” ( <i>BRCA</i> carrier, age 46)

Of note, publicity and awareness of the *BRCA* genes was mentioned by participants in each group as impacting family sharing, but for different reasons. One *BRCA* carrier felt the publicity from celebrity disclosures, specifically Angelina Jolie, made sharing with family easier. Similarly, a *PALB2* participant (age 48) used the *BRCA* gene as an example when describing *PALB2* to family members. She stated, “I told them the type of genetic mutation it was and what I understood, [and] how it related to *BRCA*.” A different *PALB2* carrier (age 59) felt that awareness of *BRCA* actually made non-relatives that she shared with less sympathetic: “They’re like, “It’s not *BRCA*. ” I mean they believe me, but it’s just not known. They probably just think, “Oh, you don’t have the real gene, cause you don’t have *BRCA*.” Fortunately, this participant said that her family members felt differently about *PALB2*, and “figured whatever it was, it was bad and important.”

**Table 9.** Theme: Impact of Public Knowledge and Awareness of *BRCA*

Brief description	Illustrative quotes
Participants felt that awareness of <i>BRCA</i> among the public impacted the sharing process.	<p>“Having it be <i>PALB2</i> makes me feel like lesser than, like it’s not a real risk...Even in my own mind, it’s not <i>BRCA</i>, it’s a smaller risk, even though I have evidence that it’s very active in my family. Then how I feel others just discount me so much, because it’s not <i>BRCA</i>. Even people who know, but not my family, but people who know. Even doctors, I feel like just really discount the risk, cause it’s not <i>BRCA</i>. ” (<i>PALB2</i> carrier, age 59)</p> <p>“Because that [Angelina Jolie <i>BRCA</i> disclosure] was in the news, they understood, there was no explaining or anything. They got it.” (<i>BRCA</i> carrier, age 60)</p>

#### *Self-Efficacy*

Overall, participants in the *BRCA* group and the *PALB2* group felt confident in their ability to effectively share with family members. Participants felt most confident communicating

with their close female relatives, such as sisters, mothers, and cousins. Some participants felt less confident because they “didn’t know everything” or were unable to answer all of their family members’ questions. On the other hand, one *BRCA* carrier (age 37) with a background in genetics felt especially confident talking to her family about her result, saying, “I think just having a background in genetics made it easier for me, both in that I had a better understanding of it and also because people trusted me more.”

**Table 10.** Theme: Confident in Sharing Basics

Brief description	Illustrative quotes
Participants felt very confident sharing their test result with at-risk relatives, but less confident answering subsequent questions.	<p>“I wasn’t worried about it, I wasn’t insecure about it, I wasn’t not confident that I understood it. I felt I knew enough to share it intelligently.” (<i>PALB2</i> carrier, age 54)</p> <p>“There’s a lot of questions that I didn’t know the answer to and even my doctor didn’t. Because again, five years ago they really didn’t have a lot of information about the <i>PALB2</i> gene.” (<i>PALB2</i> carrier, age 49)</p> <p>“I felt confident about it because I had it done after my cancer diagnosis and my sister had already had it done. I kind of had an inkling that I would be positive with that, so we talked about it among ourselves, my sisters and I.” (<i>BRCA</i> carrier, age 60)</p> <p>“Well I don’t have all the answers, you know. There’s a lot of questions... there’s a lot of things I didn’t know.” (<i>BRCA</i> carrier, age 64)</p>

Confidence was bolstered by written information about their gene mutation and the associated risks from a provider or even the genetic testing laboratory. While the majority of women in both groups were offered resources to aid disclosures, roughly 1/3 of participants did not report receiving materials. Several *PALB2* carriers found that the family letter from their provider (most often a genetic counselor) made sharing accurate information much easier. One

*BRCA* participant watched videos prior to genetic counseling, and thought that alternatives to printed handouts could be beneficial for sharing, too. When asked what other resources may be helpful with family sharing, both the *BRCA* and *PALB2* groups suggested a handout containing information on why sharing is important and a short script of what to say and additional online resources for gathering more information. When asked if having a healthcare provider disclose results would help participants feel more efficacious about sharing, there were mixed feelings in both groups for fear of bombarding relatives without notice or the disclosure being too impersonal. Though, one *BRCA* carrier (age 52) mentioned her sister would “take it more seriously” coming from a healthcare provider.

**Table 11.** Theme: Informational Resources Boost Self-Efficacy

Brief description	Illustrative quotes
Participants feel more confident when given resources about their gene mutation and the associated risks to use for sharing.	<p>“I would say perhaps when you receive the information from the geneticist, maybe a little script from the geneticist on, “Here’s why it’s important to share with your family members. Here’s some talking points. Here’s some nice ways to deliver it.” Maybe that would’ve been a nice thing to have in your back pocket when you’re going out to share this information.” (<i>PALB2</i> carrier, age 60)</p> <p>“Well, I think having that letter, and even recently my younger brother said, “I need to get that stuff done.” So I scanned my letter and resent it to him. I think having that written information is very, very helpful.” (<i>PALB2</i> carrier, age 63)</p> <p>“With the letter, more confident, because here’s what I got, here’s the results, here’s a copy. It helped versus just telling someone because I think people believe, whether it’s right or wrong, if they have something in print and shows research and shows the lab and shows whatever, I think they’re more likely to believe it.” (<i>PALB2</i> carrier, age 60)</p> <p>“Maybe some more links of like websites to visit, that would have given me more information to look on my own. You know, like reputable ones.” (<i>PALB2</i> carrier, age 39)</p>

**Table 11** (continued)

Brief description	Illustrative quotes
Participants feel more confident when given resources about their gene mutation and the associated risks to use for sharing.	“Maybe if there’s some type of short, little animated something, not so serious, comic or something, they could see on the social media because especially young people, that’s where everyone is.” ( <i>BRCA</i> carrier, age 38)
	“I was especially [confident] with the information that I got straight from the testing company because that made it so much easier. I mean it was detailed and it was really good information, so if I had to do it myself it would be hard to explain it to them...it also probably showed them how serious it was because it wasn’t just coming from me.” ( <i>BRCA</i> carrier, age 60)
	“Maybe a brochure that gives you helpful hints on how to share.” ( <i>BRCA</i> carrier, age 52)
	“So when I had my genetic testing they sent me a video...I thought that was really interesting to me even though I felt like I already had a good understanding it still was interesting to watch, but I feel like for people who didn’t have as good of an understanding I thought that was really helpful.” ( <i>BRCA</i> carrier, age 37)

Participants were asked to offer advice or suggestions to other patients considering sharing a test result with family. Both groups recommended sharing simple information with at-risk relatives. Collectively, they would encourage others to research and prepare before sharing and send resources to family members afterwards. Both groups stressed focusing on facts rather than emotions, and if conflicted about sharing, considering which family members need to know and why.

#### *Knowledge and Skills*

Participants were well-informed about which family members were at-risk and utilized a variety of methods to disclose their positive result. Many participants shared in-person, either in an individual or group setting, or via phone, text, email, or social media. Both groups frequently



enlisted the help of other family members to ensure that all at-risk relatives were contacted.

Many participants provided or at least offered their family members resources to supplement the initial conversation. Interestingly, more *PALB2* carriers compared to *BRCA* carriers utilized a family sharing letter from their provider to disseminate the information. The information communicated to relatives was fairly consistent between the two groups, focusing on the cancer gene involved, associated risks, heritability, and availability of genetic testing and follow-up care options. *PALB2* carriers consistently mentioned breast and pancreatic cancer risks when recalling their conversations with family members, however there was variability in reporting ovarian cancer risk.

**Table 12.** Theme: Knowing What to Share and How to Share

Brief description	Illustrative quotes
Participants demonstrated strong knowledge of methods for sharing and relevant information to provide when sharing their test result.	<p>“So what I decided to do on my own was make a list of paternal cousins and send out letter letting them know what had happened with our family, and then sent them a little information on <i>PALB2</i> – not a lot to overwhelm.” (<i>PALB2</i> carrier, age 65)</p> <p>“I doubt I told any in person, initially...I probably sent out a group text or something.” (<i>PALB2</i> carrier, age 39)</p> <p>“I said, “I had the genetic testing. I do carry the <i>PALB2</i> gene. After mom was tested, it confirmed which side of the family the <i>PALB2</i> gene comes from, what the result is, it is higher probability of breast and pancreatic cancer and just you should be aware of that. You should get tested if you’re interested.” And that’s how I put it.” (<i>PALB2</i> carrier, age 60)</p> <p>“I mean because some people have said they sent letters to their family members and stuff like that. I mean I would have never dreamed of doing that. You know, I called everybody.” (<i>BRCA</i> carrier, age 64)</p>

**Table 12** (continued)

Brief description	Illustrative quotes
Participants demonstrated strong knowledge of methods for sharing and relevant information to provide when sharing their test result.	<p>“We had a big, not a big, but my dad, his 80<sup>th</sup> birthday was a few weeks ago, and yeah, everybody now in my family, which most of them I hadn’t seen for several years, but they all know now.” (<i>BRCA</i> carrier, age 52)</p> <p>“I talked to them about it and told them basically passed on all of the information and the chances that they might have it... I told them about their increased chances of the certain types of cancer. I told them I had the documentation if they’d like to see it.” (<i>BRCA</i> carrier, age 59)</p>

### *Salience*

Both the *BRCA* and *PALB2* groups felt that family sharing was highly important because it enabled family members to take appropriate actions. Many admitted they would want this information from another family member who tested positive. Multiple participants in each group cautioned that there are circumstances in which sharing may be less important, for example if the recipient is not prepared to learn of the risk or is expected to react poorly based on their personality or past behaviors. Nonetheless, participants who anticipated or experienced a negative reaction from a family member stated they would still share despite the perceived or actual negative outcome. One *PALB2* participant (age 69) shared, “I felt like if I had angered her or made her upset, then she wasn’t thinking in her best interest, and that wasn’t going to stop me [from sharing].”

Surprisingly, participants in each group explained that the importance of sharing with family was somewhat dependent on our understanding of the gene’s penetrance and associated risks. Specifically for *PALB2*, one participant explained that as the gene became more understood and the management recommendations changed, sharing became more important. Participants in both groups admitted having perceptions that female relatives were at greater risk

given the associated cancer risks compared to male relatives, but acknowledged that both women and men could be carriers of the familial variant.

**Table 13.** Theme: Important When Risks Are High, Actionable and Relative is Prepared

Brief description	Illustrative quotes
Participants felt that sharing a positive genetic test result is important in most situations.	<p data-bbox="646 527 1421 737">“It’s important. People need to know what they’re up against. If you don’t know what your history is, your medical history is, how can you counteract it? How can you start making changes as early as you possibly can to thwart off any possible disease inflicting you.” (<i>PALB2</i> carrier, age 49)</p> <p data-bbox="646 779 1421 842">“I felt like it was information that I didn’t have before that was very important to share.” (<i>PALB2</i> carrier, age 69)</p> <p data-bbox="646 884 1421 1062">“I think it’s very important because then I at least have the knowledge and then I can do with it what I want... I might have been really upset if a couple of my cousins hadn’t shared this and then all of a sudden I got breast cancer.” (<i>BRCA</i> carrier, age 64)</p> <p data-bbox="646 1104 1421 1325">“I think it’s probably most important if it affects their health perspective, but if it’s just mostly about you sharing information about your own health, I don’t think it’s important. I mean, if we’re talking about you find out that you have a mutation that they might have too, then I guess I’d say it’s more [important].” (<i>BRCA</i> carrier, age 30)</p>
Participants felt that sharing a positive genetic test result is less important if family members cannot handle knowing.	<p data-bbox="646 1367 1421 1514">“You have to know the people that you’re going to be telling and come up with an idea of, “Should I do this or not?”... You have to weigh the pluses and the minuses of telling them or not telling them.” (<i>PALB2</i> carrier, age 57)</p> <p data-bbox="646 1556 1421 1761">“It’s important to share it...it’s important to share it with the ones you want to share it with, if there was a reason why. Let’s say it was not going to be a good idea to tell someone because of their particular state of mind or health or something, then of course it’s not going to be necessary.” (<i>PALB2</i> carrier, age 48)</p>

**Table 13** (continued)

Brief description	Illustrative quotes
Participants felt that sharing a positive genetic test result is less important if family members cannot handle knowing.	“It depends on how close you are with them and just how much information they really need to know. Like I said, some people can’t handle it for the fact that they may not understand everything that you are talking about.” ( <i>BRCA</i> carrier, age 60)
Participants felt that sharing a positive genetic test result is more important when there are high risks and relatives can take action, especially females.	<p>[On a scale of 1 to 10, how important is sharing your result with family?] “When I first got diagnosed and they said, “Oh we don’t know much about <i>PALB2</i>,” I would have said maybe like a 3 or 4. Now that I think the standard of care is if you have the <i>PALB2</i> you do get a mastectomy right away and you’re put on high alert, that’s a lot different than what we talked about in 2015.” (<i>PALB2</i> carrier, age 54)</p> <p>“I think maybe if they would have been female, I don’t know why, but I would have been more praying to maybe tell them, because of the breast and ovarian aspect.” (<i>PALB2</i> carrier, age 57)</p> <p>“I think it’s mostly just how it affects their health, and I guess how severe your mutation is, whether it’s something that carries a higher risk of having a disease or if it’s like, “No, you’re definitely gonna get it.” (<i>BRCA</i> carrier, age 30)</p> <p>“In my mind, she’s the only one [at risk], it’s not true that it only affects her but it affects her more because she’s female.” (<i>BRCA</i> carrier, age 55)</p>

### *Intention and Decision*

Participants reported sharing their result among various first-, second-, and third-degree relatives on the at-risk side of their family (if known), otherwise both sides of their family.

Immediate family members, including children, siblings, and parents, were consistently informed, whereas nieces, nephews, aunts, uncles, and cousins were not always contacted. For both the *BRCA* and *PALB2* groups, family members that were not directly contacted by the participant were often informed by a different family member. Young children were usually not

informed of the positive genetic test result, though participants expressed intention to share with them in the future. Participants did not want to burden their children with this information and felt it would be better to wait until the information could be fully understood and used for medical decision-making.

Overall, participants were very satisfied with their decision to share their result with family members. Several participants expressed frustration, though, due to lack of follow-up among their family members. While reflecting on her decision to share, one *BRCA* carrier (age 52) felt “completely satisfied with [my decision], completely unsatisfied with their reactions. All of them.” Participants in both groups said that financial issues, competing life demands, perceived lack of relevance, and preference towards not knowing their carrier status were frequent barriers that family members faced related to genetic testing. Some participants expressed that sharing may need to be an on-going conversation to ensure appropriate family follow-up.

Results were frequently shared with non-family members, such as friends, support groups, co-workers, and spouses. Similar to their attitudes with family members, participants reported sharing with non-relatives to increase awareness of genetic testing and the importance of screenings, like mammograms, provide life updates, and receive support. Many participants were prompted to share their result with non-relatives when the topic came up in conversation. A *PALB2* carrier (age 51) said, “I didn’t bring it up unless it was something that somebody asked me about or whatever. But I’m not the kind of person to hide things, so if somebody asked me something I’d tell them.” When asked about disclosing to co-workers, a few participants preferred to keep their private and professional lives separate, thus chose not to share in work settings.

**Table 14.** Theme: High Rates of Sharing and Intention to Share

Brief description	Illustrative quotes
Participants reported sharing (directly or indirectly) or intending to share with the majority of their at-risk relatives.	<p>“I knew that my mom had talked to her brother about it and then he talked to his sons. So that took care of that family. My mom had another sister that she wasn’t really in touch with, but I think her brother ended up talking to the sister. I didn’t really feel the need to share the news with anyone.” (<i>BRCA</i> carrier, age 30)</p> <p>“I think that when he’s a little bit older I’m going to suggest that, you remember how I had cancer, and that you might wanna ask your doctor about getting tested for this just to see if you have it.” (<i>BRCA</i> carrier, age 52)</p> <p>“I would say 18, but I would probably push it out. So maybe out of college or something when he’s starting to actually get a life and get things settled for himself.” (<i>PALB2</i> carrier, age 39)</p>
Participants reported sharing with non-family members, such as friends, coworkers, and support groups.	<p>“I’ve told most of my friends about it. and because I think it’s very important for people to know even if they’re not facing it, that maybe they know somebody else who should be tested or whatever and every time I hear about somebody whose parent had pancreatic cancer or whose had ovarian cancer, my first question is have they ever been tested? Have you ever been genetically tested?” (<i>BRCA</i> carrier, age 59)</p> <p>“They’re friends, people who I care about and who care about me and wanted to know what was happening with my diagnosis and all the things that went along with it like this...people wanted to be informed and involved and so I informed them and I involved them” (<i>PALB2</i> carrier, age 48)</p>

### *Barriers and Facilitators to Family Sharing*

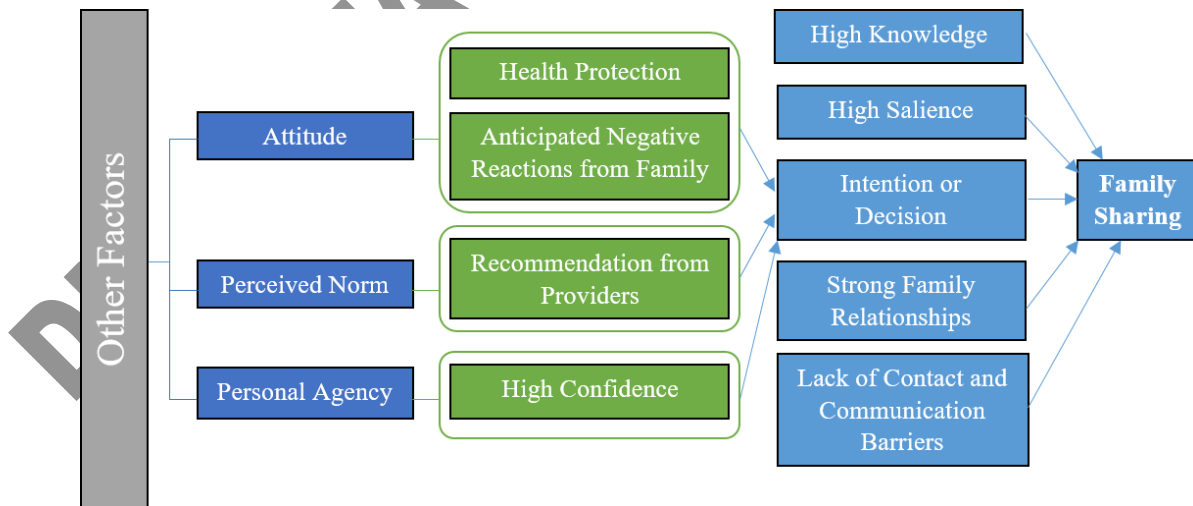
The major themes identified through in-depth interviewing with *BRCA* and *PALB2* carriers served as barriers and facilitators to family sharing. Facilitators, or factors that promoted family sharing, included the following themes: health protection; normative influence from providers; strong family relationships; and high confidence, knowledge, and salience. Barriers, or factors that inhibited family sharing, included the themes anticipated negative emotions from

family members and lack of contact and communication barriers. The remaining themes were endorsed as both promoting and inhibiting sharing, thus could not be discretely assigned as a facilitator or barrier to sharing.

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## Discussion

Our findings suggest that women with a pathogenic *BRCA* variant or *PALB2* variant experience similar barriers and facilitators when disclosing a positive genetic test result to at-risk relatives and may benefit from similar interventions to improve rates and quality of family sharing. To our knowledge, this is among the first studies to qualitatively examine barriers and facilitators outside the realm of *BRCA*-related test results, thus adding to the literature on family sharing in other hereditary breast cancer genes. According to the Integrated Behavioral Model (IBM), participants' attitudes, normative influences, and personal control beliefs served as barriers and facilitators that influenced their motivation to share and, in combination with other factors, their ultimate decision to share. These findings applied to the IBM framework are shown in Figure 2. It seems that facilitating factors outweighed barriers to sharing, which enabled the high rates of disclosures reported by this sample.



**Figure 2.** Family Sharing for *BRCA* and *PALB2* Carriers Using IBM Framework.



*Theme: Health Protection*

Participants in both the *BRCA* and *PALB2* carrier groups reported similar facilitators to family sharing, most importantly the attitude that disclosing to at-risk relatives enables health protective behaviors. This theme is consistent with previous studies that have found the most salient motivators for sharing were to make relatives aware of possible risks and enable appropriate follow-up care (Hughes et al., 2002; McGivern et al., 2004). A more recent study by Ricker et al. (2018) similarly found that both high- and moderate-penetrance gene carriers agreed that family sharing is important for facilitating early detection and prevention strategies among at-risk relatives. Although previously reported in the literature, sharing for the purpose of receiving emotional support and advice was not a primary motivator among this sample (Hamilton et al., 2005; Hughes et al., 2002). Many participants already felt supported and informed, so they were not motivated to share for these reasons.

*Theme: Anticipated Negative Emotions from Family Members*

Women with *BRCA* variants or *PALB2* variants had similar concerns about family sharing, particularly related to how family members would respond. Although this did not keep participants in this study from sharing, the majority of women in both groups felt that disclosing their positive genetic result might cause certain family members to feel fear, worry, distress, and even guilt. Other studies have similarly found that individuals are less likely to share if they anticipate family members reacting poorly (Derbez, 2018; Forrest K et al., 2003; Hamilton et al., 2005; Lafrenière et al., 2013). It appears that this barrier can be overcome, as observed in this sample, when individuals are sufficiently motivated by other factors, such perceived benefits, importance, confidence, and ease of sharing.

*Theme: Family Reactions Range from Supportive to Not Supportive*

Family reactions have been reported as ranging from interest to disinterest (Gaff, Collins, Symes, & Halliday, 2005). For the most part, family members in this study were reported as responding positively during family sharing (e.g. supportive, grateful, receptive, not surprised) or indifferent to the news. Individuals in the position to disclose a positive genetic test result to family may find relief in knowing that family members are often receptive during family sharing, even if they do not act upon the information provided; however, individuals should be prepared for any relatives that may react negatively.

*Theme: Normative Influence from Providers and Family*

Encouragement from healthcare providers to share genetic test results was the most consistent source of normative influence among both *BRCA* and *PALB2* carriers, as expected support from family members was variable. As discussed in Black et al. (2013), healthcare professionals have an important role in initiating the family sharing process and identifying all at-risk relatives. Providers are especially important for helping patients understand the significance of sharing with their more extended relatives. The pre-test counseling session has been viewed as an advantageous opportunity to introduce the idea of family sharing, though ongoing support after the pre-test meeting is important for patient follow-through (D' Audiffret Van Haecke & de Montgolfier, 2016). Although a few participants could not recall a specific conversation, they all reported that their provider(s) presumably encouraged them to notify family.

*Theme: Strong Family Relationships*

Women in both carrier groups felt that strong relationships with their families made it easier to share their positive genetic test result. Participants who communicated with relatives

frequently and openly found it easier to disclose this information, even more so if the family members had known about the participant's cancer diagnosis and/or the family history. Family communication styles, norms, and awareness have been shown to influence willingness to share (Dean & Rauscher, 2018; Etchegary et al., 2013; Kardashian et al., 2012; Katapodi et al., 2013; Koehly et al., 2009; Lafrenière et al., 2013; Peters et al., 2011). Strong family relationships were also exhibited via relatives' willingness to help with the disclosure process. The significance of involving other family members in the sharing process has been described (Koehly et al., 2009).

*Theme: Lack of Contact and Communication Barriers*

*BRCA* and *PALB2* participants both cited distant relationships and logistical struggles as factors that made sharing their positive test result with at-risk relatives more difficult. Minimal contact due to emotionally distant relationships with relatives has been reported as a barrier to family sharing frequently in the literature (Daly et al., 2016; Elrick et al., 2017; Etchegary et al., 2013; Hughes et al., 2002; Kardashian et al., 2012; MacDonald et al., 2007; McGivern et al., 2004). While many participants were able to overcome logistical hurdles, like trouble obtaining contact information or actually making contact, some *BRCA* and *PALB2* carriers did not and ended up not disclosing to all at-risk relatives. As reported in other studies, these communication barriers frequently inhibit family sharing, even if non-communication is unintentional (Nycum et al., 2009).

*Theme: Impact of Public Knowledge and Awareness of BRCA*

Participants in both carrier groups mentioned the publicity of the *BRCA* genes as impacting the family sharing process. On one end, family members' prior knowledge and awareness of *BRCA* made disclosing a *BRCA* or even a *PALB2* result somewhat easier since the concept was familiar. Celebrity *BRCA* disclosures, such as Angelina Jolie, and the influx of

direct-to-consumer genetic tests have been shown to increase awareness and even uptake of genetic testing (Roberts & Dusetzina, 2017). However, one *PALB2* carrier was frustrated that *PALB2* was viewed by others as a “less serious” hereditary cancer gene compared to *BRCA*. This participant’s experience is alarming, especially the misconceptions from healthcare providers, given that *PALB2*, like *BRCA*, is considered a highly penetrant cancer gene. On-going educational efforts are needed to raise awareness about hereditary cancer beyond *BRCA* among providers and the public so that lack of understanding does not inhibit family sharing and medical management (Dean & Rauscher, 2018).

*Theme: Confident in Sharing Basics*

*BRCA* and *PALB2* participants both reported high confidence in their ability to share their positive genetic test result with at-risk relatives, which is likely due in part to this sample’s high educational background. These women were most confident disclosing to their close, female relatives, which is consistent with previous reports that information is more often communicated to female relatives (Cheung et al., 2010; Elrick et al., 2017; Etchegary et al., 2013; Kardashian et al., 2012; MacDonald et al., 2007; Patenaude et al., 2006; Vadaparampil et al., 2012). Difficulty or uncertainty when responding to relatives’ questions diminished participants’ perceived ability to share effectively. This finding suggests that highly educated *BRCA* and *PALB2* carriers feel confident disclosing basic information about their result to relatives, but may benefit from having resources and contact information for genetics professionals on-hand when sharing. Individuals need to feel prepared in order for familial disclosures to occur and those that have more knowledge may feel more comfortable (Cheung et al., 2010; Dean & Rauscher, 2018).

*Theme: Informational Resources Boost Self-efficacy*

Most participants were at least provided some written information about their specific gene and the associated cancer risks from their provider (frequently a genetic counselor), but not all received resources related to or to assist with the family sharing process. Several women in the *PALB2* carrier group found that the family sharing letter from their provider was especially helpful in disseminating information to at-risk relatives. The use of family sharing letters as a patient resource has become standard practice when heritable genetic risks are identified (Dheensa, Lucassen, & Fenwick, 2018). Providers did not consistently offer participants in either group family sharing letters, so it is difficult to determine whether or not the utility of the letter was specific to the *PALB2* group. It is possible that *PALB2* carriers found the family sharing letter more helpful given that there is less information and awareness regarding the *PALB2* gene compared to *BRCA1* and *BRCA2*.

*BRCA* and *PALB2* carriers suggested that a handout explaining the significance of family sharing, tips for how to share, and even a short script of what to say would be helpful when disclosing to relatives. Kardashian et al. (2012) designed an educational sharing risk information tool (ShaRIT), consisting of genetic information along with family resources (including a letter to family members, FAQ sheet, contact information for providers, and support websites and brochures) that was well-received by participants. A similar web-based educational aid developed by Katapodi et al. (2018), called the Family Gene Toolkit, was well-received during focus groups. Based on participants' responses, interventions like these could be effective at increasing confidence with sharing and the likelihood of disclosures. None of these included a script of what to say to relatives, so that may be a valuable addition. An example family sharing script can be found in Appendix C.

*Theme: Knowing What to Share and How to Share*

The participants in both the *BRCA* and *PALB2* carrier groups demonstrated a clear understanding of which family members were ‘at-risk,’ what risk information was important to share, and how they might go about sharing, though this is not always the case (Blandy et al., 2003; Daly et al., 2016). This finding is likely related to higher education levels among our sample and consequently greater understanding of relevant information to share. Conversations with family members focused on the cancer gene involved, associated cancer risks, heritability, and availability of genetic testing and risk management. The associated cancer risks reported by the *PALB2* carriers varied slightly in terms of ovarian cancer, likely due to changes in the scientific community’s understanding of *PALB2*-associated risks over time (Metcalf, Akbari, Narod, & Lerner-Ellis, 2017). This highlights the importance of on-going communication with patients or finding ways for patients to receive updated information related to their gene mutation.

*Theme: Important When Risks Are High, Actionable and Relative is Prepared*

The belief that family sharing is highly important was a facilitator for both *BRCA* and *PALB2* carriers. Multiple participants in each group felt that family sharing was less important in certain situations, for instance if they expected that family members would react poorly to the news. This observation relates back to anticipated negative emotional reactions from relatives and concerns that certain family members may not be prepared to learn about possible risks. The Disclosure Decision-Making Model explores how individuals assess recipients when making disclosures, and in the context of *BRCA* has demonstrated an association between perceived readiness of relatives and likelihood of family sharing (Dean & Rauscher, 2018; Greene, 2009). Participants who experienced a negative reaction from a relative after sharing said they would

still have shared despite the outcome, which suggests that participants feel sharing is more important than the perceived duty to protect relatives from emotionally troublesome information.

The importance of sharing was also modified by the gene penetrance, associated risks, and availability of follow-up care. Participants expressed that sharing was more important when the cancer risks were high and more certain, and if relative's had the ability to consider risk-management strategies. Uncertainty regarding VUS test results has been shown to negatively impact family sharing due to the complexity of the result and lower perceived utility of this information for relatives (Hughes et al., 2002; Patenaude et al., 2006; Vadaparampil et al., 2012). Participants in this study were all gene positive, but may similarly see genetic test results as less relevant to family members when there is limited or evolving knowledge of gene penetrance, cancer risks, and recommended follow-up care. In some cases, this led participants to feel sharing was more important for at-risk female relatives rather than male relatives, given the breast and ovarian (when applicable) cancer risks. Participants admitted feeling that sharing was less important for older relatives, given the lower likelihood of pursuing genetic testing or follow-up care. These gender and age tendencies with sharing have been reported in the literature before (Dean & Rauscher, 2018; Finlay et al., 2008; Patenaude et al., 2006).

*Theme: High Rates of Family Sharing and Intention to Share*

Consistent with the literature on *BRCA* carriers, this sample of women with *BRCA* variants and even women with *PALB2* variants showed high rates of family sharing, frequently disclosing their result to immediate family members and variably notifying more extended relatives (Blandy et al., 2003; Daly et al., 2016; Elrick et al., 2017; Fehniger et al., 2013; Finlay et al., 2008; Hughes et al., 2002; Kardashian et al., 2012; Katapodi et al., 2013; MacDonald et al., 2007; McGivern et al., 2004; Ricker et al., 2018). Participants managed to inform the

majority of first-, second-, and third-degree relatives at least indirectly through other family members. Both groups were highly satisfied with their decision to share their positive genetic test result, but were less impressed with their family members' follow-up. It has been well-documented that rates of genetic testing among at-risk relatives are low and interventions are urgently needed to improve family follow-up (Blandy et al., 2003; Katapodi et al., 2017; Lieberman et al., 2018). Fortunately, participants were not deterred from sharing due to low rates of genetic testing among family members.

Participants in both groups usually did not discuss their result with young children because they anticipated a lack of understanding and utility of the information. This finding is appropriate given that genetic testing for adult-onset hereditary cancer syndromes is not recommended for minors and medical management would likely not change until around age 25-30 (Caga-anan, Smith, Sharp, & Lantos, 2012; Kesserwan, Friedman Ross, Bradbury, & Nichols, 2016; National Comprehensive Cancer Network, 2018). These women intended to wait until their child reached a certain age, point of maturity, or readiness, which is a common approach taken by other women in this type of situation (Hamilton et al., 2005; Patenaude et al., 2006).

#### *Study Limitations*

There are several strengths of this study to acknowledge. To start, this study is one of the first to compare family sharing behaviors among *BRCA* and *PALB2* carriers. Participants were required to provide documentation of a *BRCA1*, *BRCA2*, or *PALB2* pathogenic variant, so we did not need to rely on a self-reported carrier status. Additionally, the sample was diverse with regard to gene status, which enabled greater variety and comparison of interview responses.

Although participants were primarily white, highly educated, and privately insured, this sample was representative of the population traditionally accessing genetic services and, thus, most



likely to face the decision to share (Armstrong, Micco, Carney, Stopfer, & Putt, 2005; Cragun et al., 2017). Extra interviews for each carrier group were scheduled to ensure the target sample size would be reached, which ultimately allowed us to include a greater number of participants in this study. After coding the initial ten interviews for each carrier group and identifying themes, the additional eight *BRCA* and three *PALB2* interviews were completed and analyzed to ensure that major barriers and facilitators related to family sharing had been captured.

Despite these strengths, this study does have several limitations. Participants were selected from a highly motivated population of women who are part of a cancer registry and willing to participate in research, which may have introduced sampling bias despite attempts to purposively select those who did not share with all relatives and those from underserved ethnic/racial groups. In terms of generalizability of these findings, it should be noted that perceived barriers and facilitators may differ among those who indicated willingness to participate and those who did not, especially given the high rates of sharing reported in this sample. Furthermore, there may be barriers and facilitators unique to younger generations, as well as minority populations and lower socioeconomic status groups with historically lower rates of family sharing, that could not be captured with this sample (Cheung et al., 2010; Etchegary et al., 2013; Fehniger et al., 2013).

The current study relied solely on self-reported family sharing behaviors, so true rates of sharing may differ from the self-reported rates. Because the outcome or quality of participants' communication was beyond the scope of this study, we did not confirm family members' reactions or what information they were told. Nevertheless, we were able to infer some participant misconceptions about *PALB2*-associated cancer risks that may have been communicated from the interviews. The time elapsed between genetic testing and disclosure was

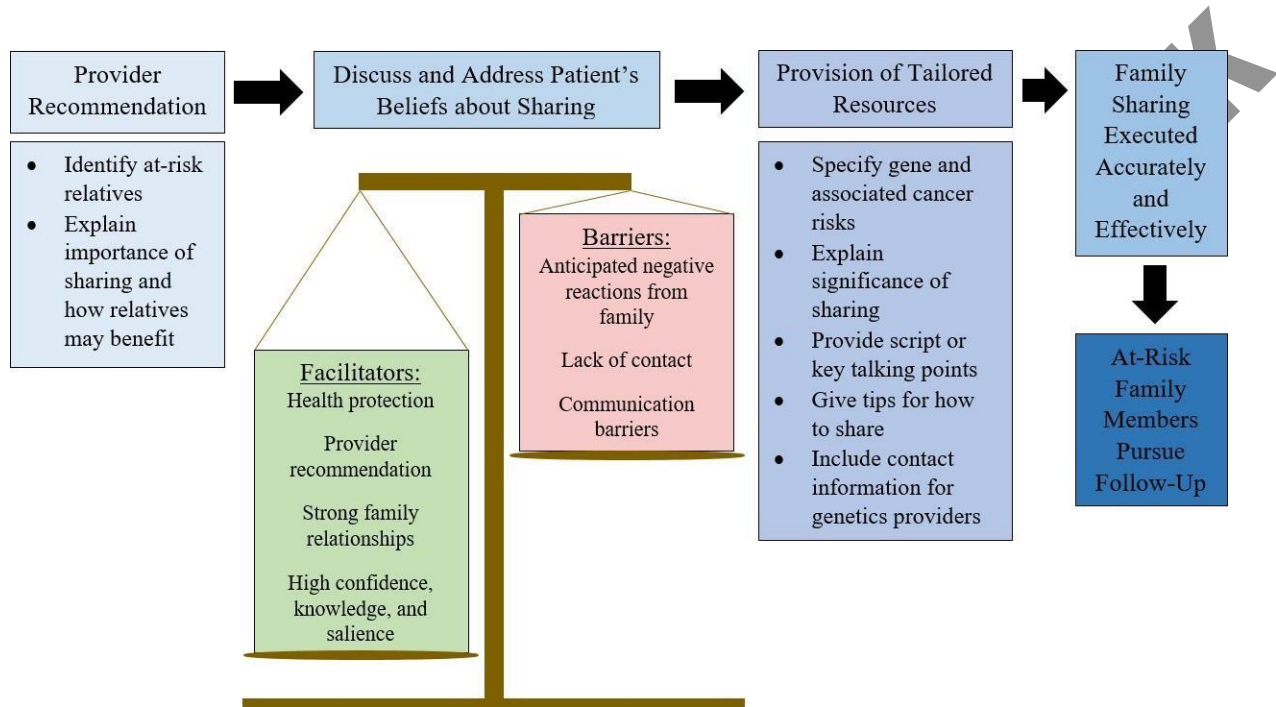
not assessed, though this may be an important factor to consider in future studies related to family sharing.

### *Practice Implications*

Current findings indicate that health care professionals play an important role in facilitating the family sharing process across both carrier groups (Black et al., 2013). Studies have found, though, that variability among providers and clinical sites makes it difficult to create a standardized protocol for addressing family sharing (D' Audiffret Van Haecke & de Montgolfier, 2016). It has been suggested that more time should be devoted to this topic during the post-test counseling visit and even afterwards via follow-up correspondence (D' Audiffret Van Haecke & de Montgolfier, 2016). During these conversations, it would be beneficial to clarify at-risk relatives and discuss patients' beliefs about family sharing in order to identify motivations and address any barriers (Gallo, Angst, & Knafl, 2009). Based on the current findings, we would expect similar, but tailored interventions to be successful at improving rates of family sharing for both *BRCA* and *PALB2* carriers.

Being well-supported and -informed throughout the family sharing process has been shown to positively impact the experience of disclosing genetic test results (Lafrenière et al., 2013). Providers should offer patients resources that explain why sharing is important and contain key talking points, tips for how to share, and contact information for genetics professionals to reduce uncertainty and build confidence (Mendes, Paneque, Sousa, Clarke, & Sequeiros, 2016). Family sharing letters and other types of educational aids are acceptable, effective reminders to share and even remind family members to follow-up (Mendes et al., 2016). Given *PALB2* carriers' high endorsement of family sharing letters, it may be particularly

helpful to offer these resources to carriers of less common gene mutations. Figure 3 depicts how these practice implications can be incorporated into the family sharing process.



**Figure 3.** Practice Implications for Providers Flowchart.

### *Research Recommendations*

Additional research is needed regarding family sharing outside the setting of high-risk hereditary cancer syndromes, such as Hereditary Breast and Ovarian Cancer syndrome and Lynch syndrome, to include other highly and moderately penetrant cancer genes. Research efforts should further examine disclosure behaviors in understudied, minority groups who may experience unique barriers and facilitators that require tailored interventions. Future studies should assess the role of providers, the utility of resources, and patients' efficacy in communicating genetic risk information to relatives in order to develop effective interventions to

improve rates and quality of family sharing, overcome age and gender discrepancies with sharing, and increase follow-up among at-risk relatives.

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## Conclusions

The Integrated Behavioral Model (IBM) provides a highly relevant framework for identifying issues related to family sharing of genetic risk information and developing appropriate and effective interventions. Current findings suggest that women with pathogenic *BRCA* variants and women with pathogenic *PALB2* variants experience similar attitudes, normative influences, and personal control beliefs when disclosing positive genetic test results to at-risk relatives, thus may benefit from similar, but tailored interventions to improve rates of sharing. Based on participant responses, future interventions should involve a discussion of patients' beliefs about sharing with healthcare providers to strengthen motivations and address other barriers and provision of informational resources to increase confidence and knowledge. It is crucial that these family sharing resources clearly specify which relatives should be informed, why sharing is important, and how at-risk relatives may benefit.

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