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# “It's Not Only About Them:“ Female Family Members' Understanding of Indeterminate Negative BRCA1/2 Test Results

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# "It's Not Only About Them:" Female Family Members' Understanding of Indeterminate Negative *BRCA1/2* Test Results

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ABSTRACT

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Genetic test results have important implications for close family members. *Indeterminate negative* results are the most common outcome of *BRCA1/2* mutation testing. Little is known about family members' understanding of indeterminate negative *BRCA1/2* test results. The purpose of this qualitative descriptive study was to investigate how daughters and sisters received and understood genetic test results as shared by their mothers or sisters. Participants included 81 women aged 40-74 with mothers or sisters previously diagnosed with breast cancer and who received indeterminate negative *BRCA1/2* test results. Participants had never been diagnosed with breast cancer nor received their own genetic testing or counseling. This IRB approved study utilized semi-structured interviews administered via telephone. The research team developed descriptive codes, and NVIVO software was used during qualitative analysis. Participants reported low amounts of information shared with them. Most women described test results as negative and incorrectly interpreted the test to mean there was no genetic component to the pattern of cancer in their families. Only 7 of 81 women accurately described test results consistent with the meaning of an indeterminate negative result—meaning a genetic cause for cancer in their family could still exist. Our findings demonstrate that indeterminate negative genetic test results are not well understood by family members. Lack of understanding may lead to an inability to effectively communicate results to primary care providers and missed opportunities for prevention, screening and further genetic testing. We recommend providing family members letters they can share with their own primary care providers whenever genetic testing is performed.

Keywords: family communication, BRCA1/2 genetic testing, genetic counseling, genetic risk communication, precision medicine, indeterminate negative test results

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## Female Family Members' Perception of Indeterminate Negative BRCA1/2 Test Results

Breast cancer is the second leading cause of cancer death in women (American Cancer Society, 2016). While as many as 10% of all breast cancers are hereditary, an additional 15-20% of breast cancers occur in family clusters due to shared genetics and environmental factors (BreastCancer.org, 2018). Women who have elevated breast cancer risk require different detection and prevention options from the general population. Appropriate detection and prevention options can be provided through precision medicine where providers individualize care based on specific genetic risk. Specific genetic risk may be identified through genetic tests and family history. Genetic counselors are specifically trained to help people understand individual genetic test results, and provide counsel according to those results.

Genetic counselors provide results of genetic tests to women with breast cancer who have obtained genetic testing (counselees). Counselees then are primarily responsible for sharing test results and risk information with family members. Informed family members may follow up with their own primary care providers to discuss personal risk potential and receive precision care.

Unfortunately, identified genetic information is not always shared with family members from counselees, or, when shared with family members, is often incomplete or incorrect. Additionally, genetic information may be misunderstood by counselees, family members, or both (Vos et al., 2011). Indeed, Vos et al. (2011) referred to family communication following genetic counseling as a "whisper game," with errors accumulating each time information is shared, recalled, or interpreted. Counselees often believe family members understand shared results, when in fact misunderstandings of information are common (Vos et al., 2011).

Indeterminate negative results appear to be particularly difficult to communicate and understand (Cypowyj et al., 2009). An indeterminate negative result is the most common outcome of *BRCA1/2* mutation testing and, in the absence of a previously identified mutation, means an unidentified genetic cause may still underlie patterns of cancer in the family. The term indeterminate negative has been used interchangeably in the literature with words such as "inconclusive" and "uninformative" to indicate that no specific genetic mutation was found. It is important to note that a test result of "no mutation identified" is not synonymous with "no mutation exists." When no mutations have been identified, future risk assessments must be based on family and personal history factors (Himes, Root, Gammon, & Luthy, 2016). Counselees often have difficulty understanding the implications of indeterminate test results for themselves and their relatives (Cypowyj et al., 2009) and may view these as true negative results. Knowing that misunderstanding and miscommunication of genetic information is common within families, a method used by genetic counselors to help counselees share accurate information is a summary letter of test results and implications (Roggenbuck et al., 2015).

Summary letters recount discussions from genetic counseling sessions and include any genetic test results. Genetic counselors typically write a section in the letter pertaining to counselees' family members, including: impact of genetic conditions, implications of test results, how to acquire individual genetic testing and counseling if needed, and appropriate screening/prevention measures. Summary letters are intended to be used by counselees as an aid to share genetic test results with family, and assist with communicating indeterminate negative results, which can be difficult to understand, remember, and explain. Additionally, summary letters may be used to alert counselees that genetic science evolves over time and can, thus, explain that additional testing may become available in the future.

Because indeterminate negative results are so difficult to discuss and understand, it is important to investigate information transfer within families. Presently the current state of information transfer related to uninformative negative BRCA test results has not been explored from family members' perspectives. Improved understanding may lead to enhanced ways to facilitate communication about genetic test results and risk within families. Therefore, the purpose of this qualitative descriptive study is to investigate if and how daughters and sisters (participants) received and understood information from the mother's and sister's (counselees) who received indeterminate genetic test results following *BRCA1/2* testing for breast cancer.

Specifically, this study aims to answer the following questions: 1) How much information did participants perceive was shared by counselees? 2) What are participants' understanding of indeterminate negative genetic test results? 3) What method of communication was used to share genetic testing information with participants? 4) Did participants report that summary letters were shared with them by counselees?

## Methods

This paper presents qualitative analysis of semi-structured interviews that were collected as part of an Institutional Review Board (IRB) approved study. Interviews were conducted from October 2013 to February 2014. A quantitative analysis of other study aims have been published elsewhere (Himes et al., 2016).

### Sampling and Recruitment

As part of a larger study (Kinney et al., 2014; Kinney et al., 2016), breast cancer survivors were identified through the Utah Population Database and recruited through the Utah Cancer Registry. All survivors met the National Comprehensive Cancer Network (NCCN) criteria (National Comprehensive Cancer Network, 2013) and received testing for *BRCA1/2*

mutations. Genetic counseling was provided via standardized in-person or telephone genetic testing and counseling. Post-test counseling was provided along with standardized summary letters alerting to the possibility that close relatives may be at increased risk for breast cancer. Additionally, all received an educational brochure with information about *BRCA1/2*-related cancer risks, genetic testing, hereditary and familial risk, and recommended medical management (e.g., screening guidelines). All survivor genetic testing results were indeterminate negative.

Each survivor (counselee) referred a sister and/or daughter (participant) who had not previously been diagnosed with breast cancer. Daughters or sisters who agreed to participate met the following inclusion criteria: women 40-74 years of age, fluent in English, having a mother or sister with a personal history of breast cancer who received *BRCA 1/2* genetic counseling and testing between 2010 and 2013, and who received an indeterminate negative *BRCA1/2* test result. Participants were excluded if they had a personal history of any type of cancer besides non-melanoma skin cancer, had ever received genetic counseling or *BRCA1/2* testing themselves, had a prophylactic mastectomy or oophorectomy, lived outside the United States, and/or were incarcerated. Women of Ashkenazi Jewish descent were not included because of their elevated risk due to the prevalence of founder mutations in *BRCA 1/2* (Heramb et al., 2018).

**Data Collection**

A mailed questionnaire and a telephone interview were used to obtain data from consenting participants. (See previously published manuscript for full details on study protocol [Himes et al., 2016]). Data obtained during the telephone interview is the focus of the present manuscript. Further details on measures and the results of data obtained through the questionnaire are reported elsewhere (Himes et al., 2016).

During the telephone interview a semi-structured interview guide was used. Interviews began with the broad question, "Tell me about the experience of having a [sister/mother] go through genetic counseling." Probing questions included, "What did she share?", and "How did she share the information?" Participants were asked specifically about their understanding of the counselees' genetic test results and if they were aware of a summary letter generated through the counseling session. Interviews were audio taped and transcribed.

**Data Analysis**

Transcripts were read multiple times to immerse researchers in data and to identify key concepts. The research team used descriptive coding as defined by Saldana (2009) to categorize interview content. Descriptive codes were developed and defined by the research team. Initially, each team member coded five interviews using NVIVO version 10 software. The team then met to ensure that descriptive codes reflected the entire dataset. In addition, codes were discussed and refined to develop themes and definitions. Once mutually-agreed-upon definitions were developed, each interview was coded by two researchers using the refined descriptive codes and themes. Interrater reliability of major descriptive themes was measured by Cohen's Kappa, and agreement was 90% or above.

## Results

Participants included 81 women from 63 families, with the range of relative participants per family being 1-4. Ages of participants were 40 to 74, of various races/ethnicities who lived in the United States (see Table 1 demographics). Lifetime risk calculations were obtained using the Claus model (Claus, Risch, & Thompson, 1994; Himes, Clayton, et al., 2016) and 5-year risk calculations were obtained using the Breast Cancer Risk Assessment Tool.

**1 - How Much Information?**

Participants were asked to rate on a scale of 0-5 how much information was shared with them by their sister or mother about her genetic testing and counseling. Most participants rated the amount as very low, with 42% reporting a 0 or 1 on a 0-5 scale (see Figure 1).

**2 – Participants Understanding of Test Results**

Although every participant had a mother or sister who received an indeterminate negative *BRCA1/2* test result, participants were categorized into those who understood the results to be negative, those who were unaware of test results, those whose understanding was consistent with indeterminate negative results, and those who believed the test results were positive.

***Participants who understand test results to be negative.*** The majority of participants (52/81) reported hearing their sister or mother describe their test results as only "negative." However, participants used different words when describing the meaning of "negative" and did not capture the inference of indeterminate negative.

Eighteen of the 52 family members in this group described negative test results in terms of the specific genes tested. For example, "*she probably just said . . . 'I don't have the gene'*," and, "*all she told me is that she tested negative for BRCA 1 and 2.*"

Indeed, 34 of the 52 family members who reported being told the test result was "negative" specified that to them "negative" meant there was no genetic component to the cancer. For example, one participant with a high lifetime risk of 22.7% reported, "*they told her whatever kind she has, is not the genetic, it's not the inherited [type]*".

***Unaware of test results.*** Many family members (22/81) were unaware of any aspect of the testing and/or test results or forgot if they were told. Some noted they may have been told but forgot (n=5), while others were certain they had never been told (n=7). Interestingly, some

family members (n=9) only found out about test results because of involvement in the present study. Women who found out about the test results because of this study were categorized as being unaware of test results, because they would not have become aware had they not been included in this study (see participant 12829 in Table 2).

***Perception consistent with indeterminate negative.*** While no family members described test results using terms "indeterminate", or the synonyms "inconclusive" or "uninformative", seven participants (7/81) interpreted the meaning of the test result consistent with an indeterminate negative finding. We categorized women's responses as consistent with indeterminate negative if the descriptions of test results allowed for the possibility that a genetic cause could still underlie the pattern of cancer in the family. One participant in this category attended genetic counseling with her family member and was able to accurately describe the meaning of an indeterminate result. Six of the seven women mentioned reported hearing the result was "negative", but they described a personal interpretation of the test result in direct contrast to what they were told (see participant 12937 in Table 2). For example, one participant referred to the summary letter during the interview process and recognized the initial impression of a negative result was not accurate. Another participant with a lifetime risk for breast cancer of 26.3% reported that when her mother told her about the genetic test results she simply stated, "*it came back negative*." However, when asked to describe the meaning of the test result she stated,

> *I don't know a whole lot about the B-R-C-A . . . it surprised me, you know, I thought that [BRCA] was* ***the*** *breast cancer gene. Obviously it's not, since. . . both my mom and my sister had breast cancer and if my mom is negative [but still got breast cancer], there's obviously lots of different types, so I don't know. (HR D 12937)*

Another woman described what she thought after her sister said the genetic test results were negative:

> *"Well, it just means that . . .other factors that contributed to her breast cancer, I need to be more careful with. . . Because when she got her results, I mean there's a reason why she got breast cancer, and if that reason is for her, then it could be for me because we are blood relatives because, I don't know." (S 12840)*

***Participants who understood test results to be positive.*** One participant described genetic test results as being "positive". She stated that many genes were tested and her sister was "positive for one" (see participant 12749 in table 2). It is possible that this sister had other genetic testing outside of this particular study.

**3 – Method of Communication**

***Indirectly shared through counselee.*** Participants were asked how genetic testing information was shared with them. A variety of methods referenced for communicating information shared in genetic counseling were reported, including: face to face conversations, telephone, text, email, social media, and family group discussions (see Table 2 fourth column). At times it was difficult to pinpoint exact methods of communication. Some reported receiving information multiple times and in multiple ways. For example, a counselee may have given initial information via text message followed by face to face discussions.

***Directly shared from genetic counselor.*** Two participants received direct information from the genetic counselor by attending genetic counseling with their sister or mother, therefore removing secondhand genetic test result information. Of the two, one described test results consistent with a definition of indeterminate negative; *"Since you're related, [and with a history] there's always an increased risk"* (S 12947). The second described the meaning of the

test results as only negative, saying, "*I'm not . . . going to be a person to get cancer because of my family genetics*" (D 12877).

**4 – Summary Letter**

Each participant was asked specifically if they had knowledge or awareness of a summary letter. Twelve of 81 participants (15%) were aware that a letter existed, either because they received a copy or because they were told that there was a letter. Conversely, 69/81 family members (85%) were not aware that counselees had been provided a summary letter containing information applicable to both the counselee and the extended family. No reference was made inferring the use of a family share letter within clustered families.

Two individuals shared the following:

*She didn't talk directly about [the summary letter]. Somehow she got her results. I don't know if they called, or they showed her the letter, I just didn't see it?* (S 12838)

*So now . . .I want to . . .contact her and ask her for that information. Or if they could reprint [the summary letter]. And if she could . . .copy it to me.* (D 12885)

While many had no knowledge about a letter, others (n=12) were aware of the summary letter. Indeed, one participant, who received a copy of the letter, referred to it during the telephone interview and discovered she had not fully understood the test results on her first reading.

*"She did send me a copy of it, and also a copy of the . . .pedigree. I see that here as well. And she did send me the results of that. It does say no mutation detected. So . . . I think when I saw that, I just kind of put it in the drawer and didn't think much more about it. I think it was very good for her to give us this report . . . It gives us some good information, and there is somewhat of a relief to know that there is no mutation detected. I think it's*

*good that it tells you that that doesn't mean that you're free and clear and don't have to worry about anything. Because, with the history there, I think it's good that they do describe that… you still need to watch things, and do your due diligence for your own health. So, I think that was a good communication to have"* (S 12936).

## Discussion

This is among the first studies to evaluate *family members'* understanding of indeterminate negative genetic test results. Other studies have evaluated *counselees* understanding of indeterminate negative test results (Baars, Ausems, van Riel, Kars, & Bleiker, 2016; Cypowyj et al., 2009; Dorval et al., 2005; van Dijk, 2005). Findings related to counselees understanding are mixed. Studies by van Dijk (2005) and Dorval et al. (2005) reported only a small minority of counselees took the indeterminate negative status as an indication of a negative test result. In contrast, Cypowyj et al. (2009) found that of 30 counselees with indeterminate *BRCA1/2* tests, 14 (47%) were uncertain about the meaning of the test, 9 (30%) believed the results were negative, and 7 (23%) believed the results were positive. The lack of clarity about the meaning of genetic test results and genomic literacy levels for counselees may be barriers to sharing accurate genetic test information, either because the information is perceived to be of little or no use to family members, or is not well enough understood to convey clearly (Cypowyj et al., 2009). Indeed, indeterminate negative test result interpretation can be difficult to understand, even for counselees who received the information first hand.

In the present study, many participants were unaware their sister or mother had attended genetic counseling at all, reporting that no or very little information was shared with them about genetic counseling. This finding was surprising because counselees provided contact information

for their family members, knowing their family members would be contacted for a study related to family communication about genetic counseling and test results.

Many who were aware their sisters and mothers attended counseling were completely unaware of test results. Our finding of limited family communication about indeterminate negative *BRCA* test results is similar to findings in studies of *BRCA* positive families. Indeed, even when genetic counselors undertake interventions to help counselees notify family members, a large portion of potentially *BRCA* positive family members remain uninformed (Mendes, Paneque, Sousa, Clarke, & Sequeiros, 2016; Sermijn et al., 2016; Suthers, Armstrong, McCormack, & Trott, 2006)

Prior research offers possible explanations for lack of family disclosure. Genetic test results may not be shared because the cancer experience is at the forefront of family focus and diminishes capacity to focus on anything else (Peters et al., 2011). Alternatively, family members may prefer to share only good news (Peters et al., 2011). Generous and Keeley (2017) suggested another reason for avoiding topics of family conversation is emotional protection. Emotional protection involves evading topics that may cause worry, or result in negative consequences. Another possible explanation for lack of sharing indeterminate negative results is results can be difficult to understand and explain; therefore the information is truncated to "negative" (Cypowyj et al., 2009). In the present study, limited information sharing within families appears to have impacted understanding of genetic test results.

We were unable to report whether including family members in genetic counseling enhanced their understanding of test results. Only two participants attended genetic counseling with counselees. One participant understood the indeterminate negative test result and was able to describe that result clearly. The other participant who attended counseling incorrectly

described genetic test results as not having any genetic connection. With such a small number of participants attending genetic counseling, drawing a conclusion about the effectiveness of first-hand information is not possible.

Participants were asked what mode of communication was used to convey genetic test results. While participants received information through many methods, our analysis did not identify connections between mode of communication and those who had an accurate understanding of genetic test results. Over time, written forms of communication such as email, blog posts or summary letters may be a source of reference to look back on for clarity when questions arise or when family members are ready to accept and assimilate the information. Indeed, several participants mentioned looking back at an email or a letter while gathering family history information for this study.

Despite the fact that summary letters were provided to all counselees to assist with communication, very few participants reported knowledge of a letter. Having genetic test results and follow up recommendations in writing, whether a summary letter, an email, or text, provides a stable source of information for family members to go back and review when needed. Indeed, several participants verbalized a wish for a copy of the summary letter, expressing a desire to read the information available.

> *[If my sister had been told to send us] a copy of the letter …that might have been very helpful to have in my records [rather than to] just say, "Oh, you guys are good. You don't have the gene." … I don't know how much counseling goes on at that point, because … when they do the genetic testing, obviously it's about them, but it's not just about them."* (S 12874)

## Study Limitations and Strengths

This study is limited because only participants' perceptions of test results were evaluated. It is possible that counselees had a clearer understanding of the meaning of "indeterminate negative" than their family members. Because interviews were conducted with relatives of counselees, it is unclear whether misinterpretation was due to misunderstanding by women being counseled, or because of the way the information was received by participants. Additionally, only including women age 40 and above is a limitation. Involving women as young as age 30 would have been more impactful clinically because screening guidelines differ based on risk level beginning at age 30. Finally, there are several risk-calculating models that take significant family history into account including Claus, BRCAPRO, BODACIA and Tyrer Cuzick. The team relied on the Claus model to calculate lifetime risks for participants in this study because that model was used most commonly at the counseling center where the research took place at the time. Risk-calculating models provide different results and the finding that 10% of the sample had greater than 20% lifetime risk for breast cancer may have been slightly different if another model had been used.

This study's strengths include being among the first to evaluate *family members'* understanding of indeterminate negative test results and awareness of summary letters. Additionally, because all counseling and testing was conducted as part of a study protocol, one can be certain that (1) counselees did receive indeterminate negative test results and (2) all received a summary letter with instructions to share information with family members.

## Practice and Research Implications

Screening recommendations for breast cancer vary based on risk level. In a separate analysis published elsewhere, Himes, et al. (2016) found 10% of participants in this study had

risk levels qualifying them for annual breast MRI screenings in addition to mammography. However, none of the participants at elevated risk had been offered, or received, screening MRI by their primary care providers. These findings demonstrate the importance of communicating genetic information to family members.

It is important to emphasize that counselees received only *BRCA1/2* mutation testing, not multigene panel testing. Multigene panel testing became available in 2013. It is estimated that 2.9 - 11.4% of women who receive multigene panel testing following indeterminate negative *BRCA1/2* test results are found to have genetic mutations associated with either familial or hereditary risk (Chadwell et al., 2018). The overwhelming belief by our participants, that the genetic test results indicated a lack of any genetic component, is of concern to the research team, because this belief might deter participants or other family members from receiving multigene panel testing. Thus, mutations may go undiagnosed due to lack of information.

Identifying and informing at-risk family members will require collaboration among genetic specialists and primary care providers. Future research should evaluate the most effective ways to communicate risk, both to family members and their care providers. This study adds to a body of evidence demonstrating that filtered information is rarely effective. Therefore, clear verbal and written information is needed for family members. Previous research has demonstrated it is more effective to provide information directly to family members through mailing information directly (Suthers et al., 2006; Trottier et al., 2015) than attempting to facilitate communication through counselees (Hodgson et al., 2016). However, even direct communication with family members does not result in all receiving appropriate testing or screening.

## Recommendations

To improve the information sharing process, we suggests improving terminology to use lay definitions and increase learning. Reporting results as, "*BRCA 1/2:* no mutation identified, other genetic contributions undetermined," could improve the overall understanding of an indeterminate negative genetic test result. We further recommend writing specific letters for family members of counselees that can be supplied to family members either by the counselee or directly from genetic counselors, if counselees' consent. In addition, with counselees' consent, a letter similar to a consultation note should be provided to each family member with instructions to deliver it to their primary care provider. Colleague to colleague letters could provide information about counselees' test results, a note about potential risk to family members, as well as information about risk-appropriate screening and prevention measures. Instructions to family members to deliver the letter and discuss the level of risk and screening with their primary care providers will add another opportunity for accurate information sharing and may improve risk-appropriate prevention and screening practices. Letters and other types of printed materials provide a stable, reliable source of information that can be reviewed at a later date.

## Conclusion

Indeterminate negative test results are often difficult to explain and challenging to understand. This study demonstrated that family members of breast cancer survivors often do not receive much information about what was discussed in genetic counseling and often do not understand indeterminate negative results. Genetic counselors as well as oncology and primary care providers alike must work together to identify ways to better inform family about genetic test results and help them understand implications for their own risk.

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Figure 1

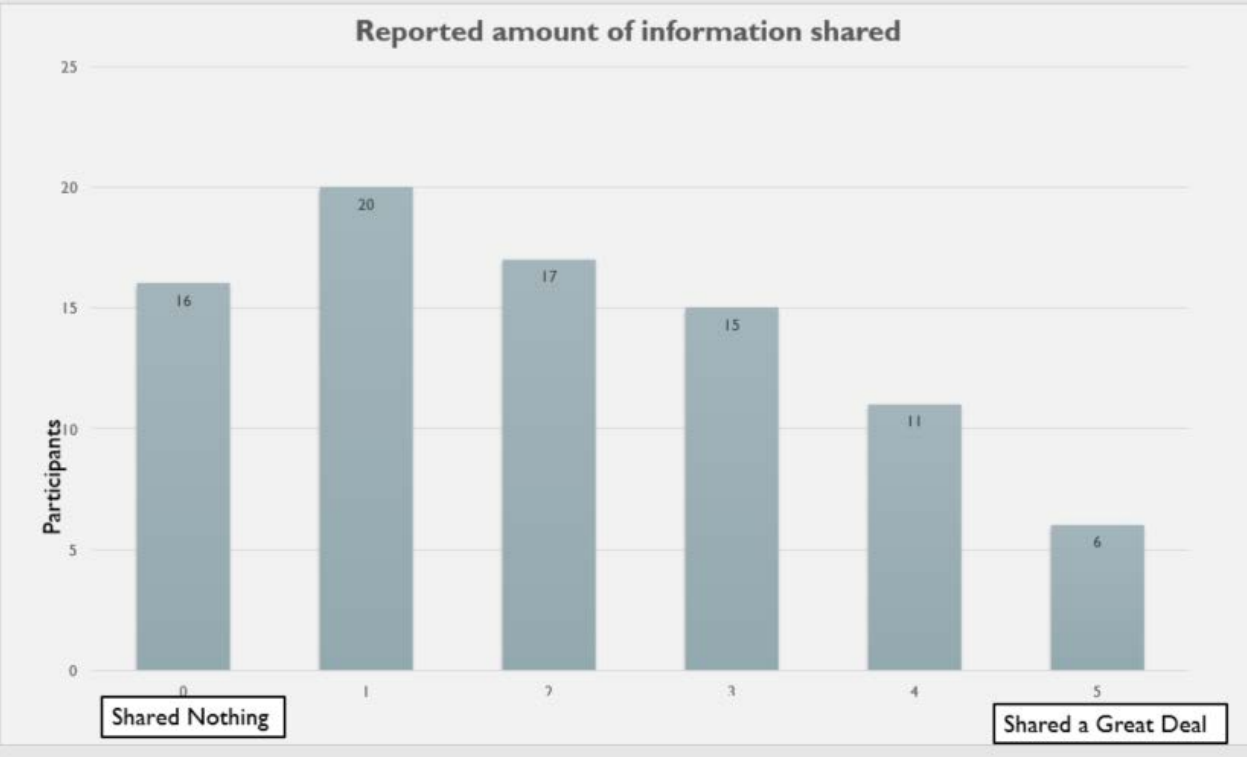

Table 1

Demographics

| | | Participants | | | |
|---|---|---|---|---|---|
| Category | | *n* | (%) | *M* | *(SD)* |
| Age | | | | 52 | (9.0) |
| Race/ ethnicity | | | | | |
| | Non-Hispanic White | 80 | (98.8) | | |
| | Asian | 1 | (1.2) | | |
| Education | | | | | |
| | High school/ GED | 11 | (13.6) | | |
| | Some college/ technical school | 30 | (37.0) | | |
| | College graduate and beyond | 40 | (49.4) | | |
| Marital status | | | | | |
| | Married or living as married | 65 | (80.2) | | |
| | Separated or divorced | 13 | (16.0) | | |
| | Widowed | 1 | (1.2) | | |
| | Never married | 2 | (2.4) | | |
| Total | | 81 | (100.0) | | |

Table 2:

S*elected Comments of Women with a Sister or Mother Who Attended Genetic Counseling*

| Participant | Participants' perceptions of test result as shared by family member | Participants' personal interpretation of test result | Participants' description of how information was shared by counselee |
|---|---|---|---|
| 51 y.o. sister of counselee (HR S 12923)<br><br>Lifetime Risk 25.2<br>5-year Risk 3.8<br><br>Fam Hx:<br>Sister breast 30's - died 30's<br>Sister breast 50's - died 60's<br>Paternal aunt breast 80's - died 90's<br>Maternal aunt breast 70's - died 80's<br>Nephew Non-Hodgkin's lymphoma Teens - died 20's<br>Nephew bone 30's - died 40's<br>Niece cancerous brain tumor 20's - died 40's<br><br>Reported Amount Information Shared 3/5<br><br>**No Summary Letter Shared** | **Negative not genetic**<br><br>Oh, do you know what and, sorry [crying?]. . .um, do you know what, it was very traumatic when she was first diagnosed, just because our previous sister had had cancer and, and had passed away. But . . . after the initial stuff, and she had her genetic tests, and then she was very relieved, we were all very relieved when it came back negative, that it wasn't genetic, and um, yeah, and do you know what? She's gone through the treatment and done beautifully and, is back to her normal self. | **Not Increased Risk**<br><br>Um, and, I, all I know is that it's, that there's not a, that the cancers were not genetically, it's not in our genetics. | **Family told together at lunch**<br><br>Do you know what? She just told us. We um, at that time we were getting together for lunch every week, just as sisters, and she just told us at lunch that she had gotten the results of her test and, do you know what? . . .She really didn't go into lots of details about what it means, but just that, it meant that our risk wasn't increased for that. |

| Participant | Participants' perceptions of test result as shared by family member | Participants' personal interpretation of test result | Participants' description of how information was shared by counselee |
|---|---|---|---|
| 54 y.o. sister of counselee (HR S 12899)<br><br>Lifetime Risk 22.7<br>5-year Risk 4.6<br><br>Fam Hx:<br>Sister Breast 30's/ Leukemia 30's - died 30's<br>Sister Breast 50's - died 50's<br>Maternal grandmother gastric 60's - died 60's<br><br>Reported Amount Information Shared 2/5<br><br>**No Summary Letter Shared** | **Negative – not genetic**<br><br>…my first sister was diagnosed and then my second sister was diagnosed and she's younger than me and then I got really worried, but she went right to genetic counseling and they told her whatever kind she has is not the genetic, it's not inherited, or I'm not sure what the . . . yeah. | **Not Increased Risk**<br><br>So it's not really making me at any more risk, I feel. | **Phone Call**<br><br>And so then she just called me as soon as she was through and said, "It's not. You don't need to worry about this," you know. So she put my mind at ease. |
| 45 y.o. sister of counselee (S 12809)<br><br>Lifetime Risk 11.1<br>5-year Risk 11.6<br><br>Fam Hx:<br>Sister breast 40's - died 60's<br>Mother ovarian 40's - died 80's<br>Paternal aunt lung 70's - died 70's<br>Maternal cousin breast 30's - died 30's<br>Maternal cousin breast 50's - died 60's<br>Maternal cousin breast 30's - died 30's<br><br>Reported Amount Information Shared 1/5<br><br>**No Summary Letter Shared** | **Negative**<br><br>And, um, and she said yes and that it came back negative | **Decreased Risk**<br><br>WHAT'S YOUR UNDERSTANDING OF WHAT THAT MEANS FOR YOU AND YOUR RISK?<br><br>Um, I, I guess, I would think that my risk is somewhat lower. | **Prompted to ask because of study – asked through Facebook**<br><br>YOU MENTIONED EARLIER THAT SHE DIDN'T SHARE A LOT WITH YOU UNTIL YOU ASKED HER ABOUT IT. CAN YOU TELL ME ABOUT THAT?<br><br>Um, I didn't even know that she'd had it, um, until basically this research study came and on the front, it said something to the effect that I'd been identified as someone who- how did it word it? – um, related to someone who'd had genetic counseling. And so when, uh, I was trying to remember my, my sister's youngest daughter's age, I just messaged her on Facebook and asked her, um, you know, her age and also asked her if she had had genetic counseling because I didn't know. She'd never mentioned it before. |

| Participant | Participants' perceptions of test result as shared by family member | Participants' personal interpretation of test result | Participants' description of how information was shared by counselee |
|---|---|---|---|
| 42 y.o. daughter of counselee<br>(D 12890)<br><br>Lifetime Risk 11.6<br>5-year Risk 1.9<br><br>Fam Hx:<br>Mother Breast 40's, 60's - died 60's<br>Maternal grandma breast 50's - died 90's<br>Maternal grandfather prostate 70's - died 90's<br>Paternal grandma Breast 40's - died 90's<br>Father liver 60's - died 60's<br>Maternal cousin thyroid 40's - died 40's<br><br>Reported Amount Information Shared 4/5<br><br>**No Summary Letter Shared** | **Negative - for specific gene**<br><br>I know, yeah, I know very little about it. All she told me is that she tested negative for BRCA 1 and 2. | **Feels literal interpretation is no increased risk, but emotional interpretation is an increased risk**<br><br>Um, while there's a lot I don't understand, I suppose it would mean that . . .I mean on one hand I take it as I don't have a higher risk than any other average person. . .<br><br>But I just have a hard time believe that with both my grandmothers and my mom having had breast cancer, so. In my mind, I feel like I'm very high risk, even without that test. . .<br><br>Even though my mother is negative, there still seems to be a family trait of it. . .<br><br>So I don't feel like her testing negative, um, that does, that just doesn't, that makes me feel a little safer, but not a lot safer. (laughs). . . | **Verbal sharing**<br><br>. . .she told me |
| 43 y.o. daughter of counselee<br>(HR D 12829)<br><br>Lifetime Risk 28.7<br>5-year Risk 1.4<br><br>Fam Hx:<br>Mother breast 40's -died 60's<br>Maternal aunt breast 40's - died 60's<br>Maternal aunt breast 40's & 50's - died 60's<br><br>Reported Amount Information Shared 0/5<br><br>**No Summary Letter Shared** | **Results Not Known – No recollection of test results shared**<br><br>She did- she really didn't share anything with me. . .<br><br>Yeah, I think I just had forgotten and I, uh, I didn't, you know what, that's amazing. I, I've gotta ask my mother what, what she learned in that. I, she may have shared it with me and I may have just forgotten. . .Or she may not have shared it, I just can't, I can't believe I can't remember that. I should, I should remember that but I just don't. | **Did not recall test result** | **Informed name was added to potential participant list for study.**<br><br>OK? SO HOW'D YOU EVEN BECOME AWARE THAT SHE HAD HAD, UM, GENETIC COUNSELING?<br><br>She told me she had and then she said that she, um, had written my name down as someone who would be interested in participating in a test and I said yes, absolutely, I would do that. . .<br><br>So that's, and, but that is all my mom told me |

| Participant | Participants' perceptions of test result as shared by family member | Participants' personal interpretation of test result | Participants' description of how information was shared by counselee |
|---|---|---|---|
| 64 y.o. sister of counselee (S 12936)<br><br>Lifetime Risk 9.3<br>5-year Risk 7<br><br>Fam Hx:<br>Sister breast 30's - died 50's<br>Mother breast 70's - died 80's<br>Maternal aunt ovarian 30's - died 30's<br>Maternal aunt cervical 40's - died 40's<br>Paternal grandma cervical 40's, gastric 40's - died 40's<br><br>Reported Amount Information Shared 5/5<br><br>**Summary Letter Shared** | **Negative**<br><br>She did not have the mutation. And that's what I'm finding out again as I look at this [the summary letter]. | **Interpretation is consistent with definition indeterminate test result.**<br><br>. . .And I, I knew when I got it [the summary letter] from her, that, you know, I read it, and I wasn't that concerned after seeing it, although I know that this is not the only thing that shows whether you kind of have a risk for breast cancer.<br><br>. . .Well, I think, I think it was a small relief, but in reading the materials that went with it. . .It did also say that that's only one part. That there's still, um, a, somewhat of a heredity factor. . . or. . .risk<br><br>M-HM<br><br>Because family members do have cancer, and there's just that susceptibility there. . .The way I understood it. | **Family Gathering**<br><br>We, we do sort of have a family reunion maybe once a year?. . .But I can't remember this particular subject coming up that often. Except I think she did pass these, uh, things [summary letters] out at one of those, uh, times when we were all together. . . But, but discussing it, probably didn't happen for more than 10 or 15 minutes. . .<br><br>And, um, and since it did come back that, uh, it, there was no mutation. . .<br><br>I think, probably, there wasn't, you know, that kind of, in the discussion that there wasn't that much to talk about. |
| 44 y.o. sister of counselee (HR S 12749)<br><br>Lifetime Risk 33.6<br>5-year Risk 2.7<br><br>Fam Hx:<br>Sister breast 30's - died 40's<br>Mother breast 40's - died 60's<br><br>Reported Amount Information Shared 4/5<br><br>**No Summary Letter Shared** | **Positive**<br><br>Oh, there were multiple genes I thought they were testing for and it seems like we were part, she was positive for one. | **Increases Risk for Family**<br><br>I just know it puts us in a higher risk factor and definitely her daughter… | **Family Discussion**<br><br>Oh, we just get together as sisters every once in a while<br><br>M-HM?<br><br>Just talk, and so that's how she just educated us, told us, followed up on it, and told us |

* Statements in all caps were spoken by the interviewers.