The Surgical and Management Decision-Making Process of BRCA1 and BRCA2 Mutation Carriers

THESIS

Presented in Partial Fulfillment of the Requirements for the Degree Master of Science in the Graduate School of The Ohio State University

By

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The Ohio State University
2016

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Abstract

Women with a *BRCA1* or *BRCA2* mutation have a 40-74% breast cancer risk and 11-46% ovarian cancer risk by age 70. Due to this elevated risk, it is recommended that mutation carriers have increased breast screening and a risk-reducing salpingo-oophorectomy. They are also given the option to have a risk-reducing mastectomy. The process of deciding if and when to undergo prophylactic surgery has been found to be a complex and difficult process for many carriers. To evaluate the decision-making process, recorded interviews were conducted with 20 *BRCA1* and *BRCA2* mutation carriers. Information about the cancer risk management decision-making process was analyzed using grounded theory. Factors involved in decision-making, ease of decision-making, and individuals involved in the decision-making process emerged as major themes. Mutation carriers who have not had breast cancer (previvors) were found to have a more difficult time coming to a cancer risk management decision than women with a breast cancer history. Physicians were often discussed as being an integral part of the decision-making process by providing support and management recommendations. Family members and other mutation carriers filled a similar role during the decision-making process by providing decisional and emotional support for carriers. Genetic counselors were short-term providers of risk information and management recommendations for this study population. If a carrier was not receiving the information or support she needed
from one of these groups, she often turned to another party, most commonly a healthcare provider. Thus, data from this study suggests that previvors or mutation carriers struggling with the risk management decision-making process may need additional support and information to assist them during this process. It is important that healthcare providers are educated about risk-management strategies for HBOC and work together to best help mutation carriers through the decision-making process. In summary, this study revealed that women with BRCA1 and BRCA2 mutations utilize physicians, genetic counselors, family members, and other mutation carriers in the cancer risk management decision-making process. The support and assistance from these individuals can impact the decision-making process.
Dedication

This document is dedicated to my parents, Alethea and Jason, and my husband, Richie.
Acknowledgments

I would first like to thank my thesis advisor, Dr. Amanda Toland, for all of her assistance and input throughout the process. Dr. Shelly Hovick has also been immensely helpful in teaching me how to do qualitative research and coding, for which I am grateful. Thank you also to Leigha Senter-Jamieson in her assistance with writing and editing. I would also like to thank Jessica Bachman, who helped me code all of the transcripts. Without her help, this project would not have been possible. I am also thankful for the support from my family and husband throughout this process.
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Fields of Study

Major Field: Genetic Counseling
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Chapter 1: Background

Hereditary Breast and Ovarian Cancer

According to the 2012 Surveillance, Epidemiology, and End Results (SEER) Program through the National Cancer Institute, breast is the most common type of cancer in females, representing 14% of new cancer cases in the United States. In the general population, women have approximately a 12.3% lifetime risk of developing breast cancer. The survival rate of breast cancer is fairly high, with an average of 89.4% of women surviving 5 years or more after diagnosis. In contrast, ovarian cancer is rare with the average woman having a 1.3% lifetime risk of developing the disease and a lower 5-year survival rate at 45.6% (Howlader et al., 2014). This is likely because ovarian cancer is difficult to detect at an early stage and has a poor prognosis at an advanced stage.

Mutations in the \textit{BRCA1} and \textit{BRCA2} genes cause hereditary breast and ovarian cancer (HBOC) syndrome, with an estimated 40-74% risk of breast cancer and 11-46% risk of ovarian cancer by age 70 (A. Antoniou et al., 2003; Chen & Parmigiani, 2007; King, Marks, Mandell, & New York Breast Cancer Study Group, 2003). Hereditary breast cancer accounts for approximately 10% of cases of breast cancer, with over 90% of these hereditary cases due to a mutation in \textit{BRCA1} or \textit{BRCA2} (Ford et al., 1998; Foulkes, 2008). Epidemiological studies have estimated that the frequency of \textit{BRCA} mutations is
approximately 0.06 to 0.26% among Caucasians (A. C. Antoniou et al., 2002; Ford, Easton, & Peto, 1995; Whittemore, Gong, & Itnyre, 1997; Whittemore et al., 2004), and 1.1 to 2.74% among individuals of Ashkenazi Jewish descent (K. A. Metcalfe et al., 2010; Roa, Boyd, Volcik, & Richards, 1996; Struwing et al., 1997).

There are many common features associated with HBOC. Early-onset breast cancer diagnosed under age 45-50 or bilateral breast cancer can be an indication of HBOC (De Leeneer et al., 2012; Graeser et al., 2009). \textit{BRCA1} mutations are associated with triple negative breast cancer, which is a relatively rare type of breast cancer that does not express estrogen receptors, progesterone receptors, or human epidermal growth factors receptor 2 (HER2) (Atchley et al., 2008). \textit{BRCA2} mutations are associated with an increased risk for male breast cancer (Evans et al., 2010). HBOC is also associated with epithelial ovarian, fallopian tube, or primary peritoneal cancer, which are together referred to as ovarian cancer (A. Antoniou et al., 2003; Liu et al., 2012).

In addition to breast and ovarian cancer, other types of cancer, such as prostate, melanoma, and pancreatic cancer, may be also associated with HBOC. For males with a \textit{BRCA1} or \textit{BRCA2} mutation, the relative risk (RR) for prostate cancer is 1.82-7.3, depending on the age of the male, mutated gene, and location of the mutation, with \textit{BRCA2} carriers having a higher risk (Breast Cancer Linkage Consortium, 1999; Levy-Lahad & Friedman, 2007; D. Thompson, Easton, & Breast Cancer Linkage Consortium, 2002; D. Thompson & Easton, 2001; van Asperen et al., 2005). \textit{BRCA2} mutation carriers
have a RR of approximately 2.6-3.2 for melanoma (Breast Cancer Linkage Consortium, 1999; Levy-Lahad & Friedman, 2007; Liede, Karlan, & Narod, 2004), and the RR for pancreatic cancer is approximately 2 for BRCA1 mutation and 3.5 for BRCA2 mutations (Breast Cancer Linkage Consortium, 1999; Levy-Lahad & Friedman, 2007; D. Thompson et al., 2002).

BRCA1 and BRCA2 are tumor suppressor genes located on chromosomes 17q21.31 and 13q13.1, respectively. The proteins encoded by these genes are important for transcriptional regulation and genomic integrity maintenance—they are thought to be involved with repairing double-stranded breaks and homologous recombination. Cancer pathogenesis of mutation carriers is thought to follow the two-hit hypothesis, with somatic mutations (the “second hit”) occurring from genetic instability due to regions of repetitive elements within BRCA1 and BRCA2. Cells without functioning BRCA1 or BRCA2 protein accumulate chromosomal abnormalities and can acquire uncontrolled growth, leading to cancer (Welch & King, 2001). This two-hit hypothesis mechanism accounts for 60-80% of cancer pathogenesis among BRCA1 and BRCA2 mutation carriers (Dworkin, Spearman, Tseng, Sweet, & Toland, 2009).

When there is suspicion of HBOC due to personal or family history of cancer, individuals should be referred for genetic counseling and a comprehensive risk assessment (National Comprehensive Cancer Network, 2016). According to the National Society of Genetic Counselors practice guideline on risk assessment and genetic counseling for HBOC,
genetic counseling should involve a risk-assessment that takes into account published guidelines, family history, and personal medical history, such as age of onset and type of cancer. During this process, other genes associated with breast and ovarian cancer besides $BRCA1$ and $BRCA2$, such as CHEK2, ATM, and PALB2, should be considered. If genetic testing is determined to be appropriate based on guidelines because it will assist in clarifying the client’s cancer risk, aspects of testing and potential outcomes, including the psychosocial impact of testing, should be discussed to help the client make a decision about testing. Management options using published guidelines and clinical judgement should then be discussed based on genetic test results, risk assessment, and family history (Berliner, Fay, Cummings, Burnett, & Tillmanns, 2013; Riley et al., 2012).

**HBOC Management**

The current National Comprehensive Cancer Network (NCCN) management guidelines for women with a $BRCA1$ or $BRCA2$ mutation include annual breast MRI or mammogram beginning at age 25, annual mammogram and breast MRI beginning at age 30, risk-reducing salpingo-oophorectomy (RRSO) between the ages of 35-40 after completion of childbearing, and a discussion about the option for a risk-reducing mastectomy (RRM). Women with $BRCA2$ mutations may elect to defer RRSO until age 40-45 if they have maximized breast cancer prevention with a RRM (National Comprehensive Cancer Network, 2016). Transvaginal ultrasound and CA-125 tumor marker analysis can be done at the discretion of the physician for ovarian cancer screening. However, current guidelines do not endorse these screening methods because
studies have not shown that they are a reasonable substitute for RRSO among BRCA1 and BRCA2 carriers (National Comprehensive Cancer Network, 2016). Some physicians and women may also choose to use chemoprevention with medications such as tamoxifen and raloxifene to reduce breast cancer risk (Stan et al., 2013).

The management guidelines for BRCA1 and BRCA2 carriers aim to either prevent cancer with prophylactic surgery or detect cancer at an early and treatable stage with screening, thus reducing cancer mortality. RRSO reduces mortality and ovarian cancer risk by up to 85% (S. M. Domchek et al., 2010; Kauff et al., 2008; Rebbeck, Kauff, & Domchek, 2009), and RRM reduces breast cancer risk by 90% (Hartmann et al., 1999; Hartmann et al., 2001). Many studies have shown that RRSO also reduces breast cancer risk by about 50% depending on age at RRSO (S. M. Domchek et al., 2010; S. M. Domchek et al., 2006; Eisen et al., 2005; Kauff et al., 2008). However, a study has recently been published suggesting that RRSO may not actually lower breast cancer risk, given that previous studies may have had biases that led to the conclusion that RRSO reduces breast cancer risk (Heemskerk-Gerritsen et al., 2015). Although no studies have been done to show that mammogram reduces mortality from breast cancer in women at high risk, trials have shown that mammography reduces breast cancer mortality by 15-40% for women at general population risk (Coldman et al., 2014; Nelson et al., 2009; Njor, Schwartz, Blichert-Toft, & Lynge, 2015). Annual breast MRI is associated with a 4.7% reduction of advanced-stage breast cancer among BRCA1 and BRCA2 carriers (Warner et al., 2011).
Despite the reduction of mortality and late-stage diagnosis associated with the management options available to mutation carrier, it is difficult to know on an individual basis when or if to pursue surgical intervention. To determine the optimal age for preventative surgery, one study used a Monte Carlo model and found that RRM at age 25 and RRSO at age 40 increased survival probability to age 70 by 26% for BRCA1 carriers and 12% for BRCA2 carriers. However, postponing RRM until age 40 reduced survival probability by only 1-2%, and doing breast screening instead of RRM reduced survival probability by 2-3% (Kurian, Sigal, & Plevritis, 2010). For some women, this reduction in survival probability from doing breast screening instead of RRM may be acceptable and they may decide not to have a RRM.

Studies have shown a wide range in uptake of prophylactic surgeries. A large international prospective longitudinal study found that 46% of mutation carriers had an RRM by age 70 and 71-86% had a RRSO by age 50 (Chai et al., 2014). A different large international prospective study found that 18% of mutation carriers had a RRM within 1.5-10.3 years after genetic testing, while 57% had a RRSO during this time (K. A. Metcalfe et al., 2008). In the United States, 0-44% of mutation carriers have been found elect to have a RRM (Botkin et al., 2003; Garcia et al., 2014; Scheuer et al., 2002), with the study finding that 0% underwent a RRM had a follow-up time of only one year and included members of one family who is predominately Mormon (Botkin et al., 2003). Studies have found an uptake of 46-74% for RRSO in the United States (Botkin et al., 2003; Garcia et al., 2014; Scheuer et al., 2002).
As for participation in cancer surveillance, a large prospective international study found that 45.8% of women with a *BRCA1* or *BRCA2* mutation elected to do breast screening only with no preventive surgery; of these women, 75% had mammogram and 19.5% had breast MRIs (K. A. Metcalfe et al., 2008). Rates of adherence to surveillance recommendations vary between studies with 26-100% of carriers having ovarian cancer screening, and 35-95% having breast cancer screening within the first year after testing (Botkin et al., 2003; Claes et al., 2005; Garcia et al., 2014). However, among women who rely on surveillance for cancer risk management, they may not follow recommendations on how often to have screening (Wainberg & Husted, 2004). Five years after testing, adherence to breast and ovarian screening recommendations has been shown to be only 2-7% by a large retrospective chart review. However, this may be an underestimate of adherence rates because in order to be considered adherent in this study, participants had to have a transvaginal ultrasound or CA-125 tumor marker analysis and an MRI or mammogram at least once every 12 months. Some women may not have been having ovarian cancer screening since it is not an official recommendation, which could account for the low ovarian screening adherence rate (Garcia et al., 2014). The evolution of published recommendations can also cause confusion among carriers and physicians about what screening is recommended and may lead to low screening adherence rates. The most common reason cited in a cross-sectional, descriptive study of 107 women at high risk for having a *BRCA1* or *BRCA2* mutation for not participating in surveillance is because it was not recommended by their physician. Other women in this study noted that
they didn’t have screening because they are unfamiliar with procedures (Loescher et al., 2009).

Effects of Management Options on Health and Quality of Life

Despite the benefits of preventative surgeries, there are also potential adverse outcomes and side effects associated with the procedures. RRSO induces surgical menopause, which can be associated with symptoms such as hot flashes, changes in mood and sexuality, and sleep disturbances due to the rapid decrease of circulating estrogen and androgens. Long-term effects of RRSO include increased risk for cardiovascular disease and osteoporosis. Some studies have also shown an increased risk for Parkinsonism, anxiety, depression, and cognitive impairment or dementia, especially if not treated with estrogen replacement. There is a higher all-cause mortality for women who had a RRSO before age 45 and were not treated with estrogen replacement, as well (Parker, Jacoby, Shoupe, & Rocca, 2009). RRSO has also been associated with metabolic syndrome, which includes a combination of the following factors: abdominal obesity, elevated blood pressure, raised fasting plasma glucose, raised triglycerides, and/or lowered HDL-cholesterol (Michelsen, Pripp, Tonstad, Trope, & Dorum, 2009).

The psychosocial effects of the cancer management decision varies between studies, but most studies have shown prophylactic surgery to increase psychological well-being. \textit{BRCA1} and \textit{BRCA2} carriers are often very satisfied with their ovarian cancer management decision once it is made, whether it be surgery or screening, but a large
A retrospective study has shown that those who choose RRSO are statistically more satisfied with their decision than those who elect screening (Westin et al., 2011). In a nationwide cross-sectional study of 846 high-risk women, those who underwent a RRSO reported less anxiety, cancer worry, and perceived cancer risk than those who chose screening (Madalinska et al., 2005). However, those who have a RRSO report more menopausal symptoms, worse sexual functioning with more discomfort, and less sexual satisfaction and pleasure (Madalinska et al., 2005; Robson et al., 2003). Although women who have a RRSO may have less sexual satisfaction, several studies have shown that there is no statistically significant difference in general quality of life between those who undergo RRSO or have screening and the general population (Campfield Bonadies, Moyer, & Matloff, 2011; Madalinska et al., 2005; Robson et al., 2003; Westin et al., 2011). However, according to a small retrospective study, menopause-specific quality of life scores are reduced for parameters such as sexual quality of life, psychosocial support, physical status, and vasomotor symptoms among women who underwent a RRSO due to a family history of ovarian cancer (Elit, Esplen, Butler, & Narod, 2001). Despite these negative effects of menopause on quality of life, 86% women who had a RRSO would choose to have the procedure again and 63% would recommend RRSO to a friend at increased risk for ovarian cancer (Madalinska et al., 2005).

RRM can also have effects on quality of life by affecting comfort, body image, and sexual enjoyment. RRM can cause breast pain and discomfort, which was shown to disrupt sleep or daily activities for 22-36% of women in retrospective study (Gahm,
Two separate studies (a Chinese qualitative study that interviewed 12 women and a Swedish retrospective survey of 59 women) found that RRM can also have a negative impact on body image and sexual enjoyment (Gahm et al., 2010; Kwong & Chu, 2012). However, a large prospective study found that there was no significant difference in sexual pleasure or discomfort between women at high risk for breast cancer who underwent RRM compared to those who did not have a RRM (Hatcher, Fallowfield, & A'Hern, 2001).

Despite the potential negative impact of RRM on body image and sexual enjoyment, many studies have shown that RRM can also have a positive impact on life. A large prospective study showed that psychological distress and anxiety decreases over time among carriers who elect to have a RRM, while women who choose not have a RRM have higher anxiety levels and their psychological distress remains the same over time (Hatcher et al., 2001). In addition to RRM helping psychological well-being, women are often happy with the decision they made as well. A prospective study of 246 women found that 75-95% were satisfied with their decision to have a RRM because it gave them a sense of reassurance and relief (Haroun et al., 2011; Kwong & Chu, 2012). A smaller retrospective study by Gahm, Wickman, and Brandberg (2010) assessed quality of life and decisional regret of fifty-nine woman at an increased risk for breast cancer post-RRM using validated measures. Quality of life for carriers who had a RRM was shown to be very similar to that of control groups, and although some expressed regret of their decision, over 80% strongly agree it was the right decision and they would do it again if
they had to do it over. However, about 10% of participants of this study feel the decision to have a RRM was not wise and did them a lot of harm (Gahm et al., 2010). So although women are often satisfied with their RRM decision and it doesn’t negatively affect quality of life, outlook on RRM is not necessarily completely positive.

Management Decision-making Process

If and/or when to have a risk-reducing surgery is often a very complex and difficult decision for many women with a BRCA1 and BRCA2 mutation (Hamilton, Williams, Bowers, & Calzone, 2009; Leonarczyk & Mawn, 2015; Ray, Loescher, & Brewer, 2005). For unaffected women found to have a BRCA1 or BRCA2, the decision-making process is often a journey that requires weighing pros and cons over time and is based on each individual woman’s situation, characteristics, and preferences (Leonarczyk & Mawn, 2015). Some describe the decision-making process as empowering, especially if they have support from friends and family (Hesse-Biber, 2014). A Swedish qualitative study found that many unaffected BRCA1 and BRCA2 carriers (previvors) feel as though they have no other option besides following published management guidelines because they feel it is a rational and moral responsibility, while others in this study questioned the management options and had more difficulty deciding what management path to follow, in part because of the uncertainty of when/if cancer will develop (Caiata-Zufferey et al., 2015). A qualitative study found that uncertainty of cancer development, as well as the irreversibility of surgery, make the decision-making process complex for women with a BRCA1 or BRCA2 mutation (Howard, Balneaves, Bottorff, & Rodney, 2011). Some
carriers may have difficulties deciding to pursue prophylactic surgery because of fear or lack of knowledge about the surgery, and uncertainties about menopause and hormone replacement therapy use (Ray et al., 2005).

These feelings of uncertainty when it comes to making management decisions can affect decision satisfaction later. A large retrospective study found that women who feel like RRSO is a difficult decision to make and feel a lot of uncertainty about the surgery are less satisfied with their RRSO decision than women who do not feel that it was a difficult decision (Westin et al., 2011). However, individuals vary in how difficult the decision is and how much time they need to come to a surgical decision. A Dutch study that involved counseling eligible women about RRSO without giving them the option of ovarian cancer screening found that over 55% decided to undergo RRSO after only 1 counseling consultation, while 26% needed 4 or more consultations to come to a decision. Individuals with a first degree relative with breast cancer were more likely to need fewer consultations before coming to a decision, while those with a first degree relative with ovarian cancer needed more consultations (van der Aa, Jessica E. et al., 2015). Although some women may need more time to make a decision, once a decision is made, they are unlikely to change their mind. A descriptive prospective study of 62 women found that about half of women who had a RRM or RRSO had the intent to do so before genetic testing, while those who had decided not to have surgery had maintained that stance over time (Ray et al., 2005). Those who have made up their mind before they even have genetic test results are likely to maintain that decision over time. However, this
doesn’t mean it will always translate into action, as some women do not end up following through with their decision to have surgery (van Dijk, van Roosmalen, Otten, & Stalmeier, 2008).

A qualitative study by Howard, Balneaves, Bottorff, and Rodney (2011) found that the goal of *BRCA1* and *BRCA2* mutation carriers for making management decisions is to preserve four dimensions of self: emotional well-being, physical health, relationships with others, and self-identity as a woman. Women with a *BRCA1* or *BRCA2* mutation imagine how screening, prophylactic surgery, and cancer can affect these dimensions of self. If it is difficult to accept the changes to self, decisions are made to maintain their current self-identities. However, if these changes to self are acceptable, decisions are made to prepare for and accommodate these changes. This study identified five decision-making styles (if-then, deferred, deliberative, intuitive, snap), that are characterized by a combination of engaging with others, looking inward, relying on intuition, paying attention to emotions, making sense of the numbers, taking time, and weighing the pros and cons. The decision-making style and approach used by carriers changes over time, varies with proposed management strategies and what dimension of self needs to be preserved, and are influenced by the context in which the decisions are made (Howard et al., 2011).

For many, but not all, *BRCA1* and *BRCA2* mutation carriers, the management decision-making process is complex and involves many different decision-making styles and
approaches. A study evaluating surgical decision-making among women at an increased risk for breast and ovarian cancer found that common questions these women ask during the management decision-making process include whether to undergo surgery, what surgery to undergo and when to do it, and what the side effects and complications may be from surgery (Klitzman & Chung, 2010). In answering these questions and trying to decide what to do for cancer management, there are many different factors that contribute to the final decision of BRCA1 and BRCA2 carriers.

Factors Involved in Risk Management Decision-making

The life stage of the woman is often a major factor in decision-making; this includes factors such as age, relationship status, and desire to have children and/or breastfeed (Hesse-Biber, 2014; Hoskins & Werner-Lin, 2013; Leonarczyk & Mawn, 2015; Ray et al., 2005). Women between the ages of 35-45 may have a particularly difficult time making surgical decisions because the seemingly “demanding” guidelines, such as a RRSAO, conflict with life issues such as maternity and breast feeding (Caiata-Zufferey et al., 2015). These demanding recommendations may make young previvors feel pressure to create a timeline for life events such as dating, marriage, having children, and breast-feeding, in order to allow for preventive surgery in the future (Hoskins & Werner-Lin, 2013; Leonarczyk & Mawn, 2015), but they may be unsure about how to manage their timeline (Hamilton, 2012). For this reason, young previvors are more likely to rely on surveillance and delay surgery until they meet some life milestones, such as getting married and having children (Hesse-Biber, 2014).
Factors that can affect risk perception, such as family history, history of a previous cancer, and the feeling that cancer is inevitable, and can play an important role in management decision-making (Hamilton et al., 2009; Haroun et al., 2011; Hesse-Biber, 2014; Hoskins & Werner-Lin, 2013; Kim et al., 2013; Leonarczyk & Mawn, 2015; Litton et al., 2009). Among women at an increased risk for breast cancer, those who decide to pursue RRM are more likely to believe that breast cancer is inevitable (Hatcher et al., 2001), with many women describing their breasts as “ticking time bombs” (Hoskins & Werner-Lin, 2013; Kwong & Chu, 2012). Women who perceive their lifetime risk of developing cancer to be greater than 50% are more likely to elect RRM than women who perceive their risk to be lower because prophylactic surgery reduces/prevents cancer (Botkin et al., 2003; Haroun et al., 2011). For some, this high perception of risk may be attributed to their experience with cancer. BRCA1 and BRCA2 mutation carriers who have had dramatic experiences with cancer in family members, such as loss of a close family member or strong involvement of caring for someone with cancer, show a strong desire to be proactive against the cancer risk (Caiata-Zufferey et al., 2015; Howard et al., 2011). For example, women who lost their mother to cancer are likely to have a preventive surgery in order to avoid the fate of their mothers, although this decision may not have been easy to make. Women with some cancer in their family but who did not lose their mother often lean towards surgery as well because they have difficulty trusting surveillance protocols (Hamilton et al., 2009; Haroun et al., 2011; Hesse-Biber, 2014). Women who elect continued surveillance often have a lower risk perception and do so
because they feel as though they may not ever get cancer and they want to wait and see if surgery will ever be needed (Hesse-Biber, 2014). On the other hand, a personal history of cancer can affect the type of treatment management chosen. Carriers who had genetic testing because they were diagnosed with breast cancer often choose the most aggressive treatment, including contralateral mastectomy, in order to decrease their risk for a future cancer diagnosis. These women often wish they had more time to make the decision, though (Hamilton et al., 2009; Hesse-Biber, 2014).

Cancer worry and anxiety about cancer screening procedures that could have abnormal findings can influence the decision-making process, as well (Hamilton et al., 2009; Haroun et al., 2011; Hesse-Biber, 2014; Hoskins & Werner-Lin, 2013; Litton et al., 2009). Cancer distress can be positively or negatively correlated with health-related behavior. For example, it is thought cancer worry can decrease the likelihood of disease detection actions, such as surveillance, because these actions can lead to cancer detection, which can be threatening. However, cancer distress can make it more likely to follow through with risk reducing actions, such as prophylactic surgery, because these actions can lead to anxiety reduction (Decruyenaere, Evers-Kiebooms, Welkenhuysen, Denayer, & Claes, 2000). This was shown in several studies that have found extreme anxiety and fear of getting cancer increased a sense of urgency to have risk-reducing surgery, making it more likely for women to elect to have a RRM, whether they have had cancer (Kwong & Chu, 2012) or not (Haroun et al., 2011; Hesse-Biber, 2014). For these women, having a RRM makes them feel empowered after surgery by allowing them to take control and
eliminate their cancer threat (Hesse-Biber, 2014). For some women, surveillance does not seem worth it because it would create a lot of anxiety and the feeling that something is wrong or will be found every time they go in for screening (Botkin et al., 2003; Hoskins & Werner-Lin, 2013; Kwong & Chu, 2012). This can push them towards having prophylactic surgery. However, a large prospective study found that experiences with screening did not have an effect on the decision to pursue RRM, as there was no difference in surgical decision based on dissatisfaction with screening (Haroun et al., 2011). The effect of screening anxiety on a woman’s surgical decision seems to be limited and be partly determined by the woman’s surgical preference before having genetic testing (Hoogerbrugge et al., 2008).

For some carriers, the changes to their body image that would come from surgery, attachment to their body parts, and the irreversibility of surgery are critical factors in their decision-making process (Botkin et al., 2003; Hoskins & Werner-Lin, 2013; Leonarczyk & Mawn, 2015; Ray et al., 2005). A qualitative study that interviewed twenty-two BRCA1 and BRCA2 carriers found that sometimes the carriers have feelings that they will no longer be a woman, won’t be attractive, or that their spouse won’t desire them anymore after a preventative surgery. These feelings are a common reason for mutation carriers to not have a RRM. Women who have these concerns about changes to body image often realize that their ovaries and breasts hold very personal meanings for them after some time of introspection, but may feel vain for having these concerns. Other women in the study felt as though their breasts and ovaries are not an important part of
their womanhood, so it is not a concern during their decision-making process (Howard et al., 2011). In addition to visual changes to the body from prophylactic surgery, other changes to the body, such as side effects of surgical menopause and hormonal changes, can be a factor in deciding against RRSO as well (Botkin et al., 2003).

For some women with \textit{BRCA1} or \textit{BRCA2} mutations, a sense of familial obligation and responsibility to be available for their children by avoiding cancer is a very important component of their decision and can make them feel pressure to adhere to management guidelines (Caiata-Zufferey et al., 2015; Haroun et al., 2011; Hesse-Biber, 2014; Howard et al., 2011; Leonarczyk & Mawn, 2015). A qualitative study found that this goal of staying alive for their children can push \textit{BRCA1} and \textit{BRCA2} carriers into having surgery sooner rather than later (Hamilton, 2012). Other women may feel as though they have a moral obligation to use the genetic information that their ancestors did not have (Caiata-Zufferey et al., 2015).

**Involvement of Others in Decision-making**

Some women make their decision in isolation, while others get support or feel pressure to make a certain decision from people in their social networks (Hoskins & Werner-Lin, 2013). Family, friends, online relationships, support group members, and healthcare providers can all play a role in the decision-making process (Hesse-Biber, 2014; Hoffman et al., 2014; Hoskins & Werner-Lin, 2013; Howard et al., 2011; Leonarczyk & Mawn, 2015). However, some people may avoid sharing and receiving input from friends
because individuals without a *BRCA1* or *BRCA2* mutation often don’t understand what carriers are going through and do not behave in a supportive manner (Hoskins & Werner-Lin, 2013). Family can provide information and advice for decision-making, but may also put too much pressure to make a certain decision on these women, especially as these women reach the age when their family members were diagnosed with cancer (Hoskins & Werner-Lin, 2013; Howard et al., 2011). In China, one study found that only 25% of *BRCA* mutation carriers with breast cancer involved their family in the decision to undergo contralateral RRM because they feel it is a very personal decision (Kwong & Chu, 2012). A qualitative study of 32 women at increased risk for breast and ovarian cancer from the United States found that other people, such as family members and friends, are involved in the decision-making process to help the carrier answer questions such as whether or not to have surgery and when to undergo prophylactic surgery. Although these decisions can be very personal and subjective, family and friends are often involved in the decision-making process to provide more input aside from the clinical expertise from healthcare providers (Klitzman & Chung, 2010). However, a different qualitative study that interviewed 22 *BRCA1* and *BRCA2* mutation carriers found that family and friends may be excluded from the decision-making process completely if they are unsupportive and/or have unpleasant reactions (Howard et al., 2011). Spouses may also be involved in surgical decision-making for many reasons, such as the implications it has on family planning, and because the surgical process and outcomes may be difficult for them as well (Howard et al., 2011; Matloff, Barnett, & Bober, 2009).
Involvement of physicians, such as surgeons, oncologists, gynecologists, and primary care physicians, in surgical decision-making was perceived to not be as useful as hoped by many BRCA gene mutation carriers. Several qualitative studies that interviewed women who were making RRM or RRSO surgical decisions because of a known BRCA1 or BRCA2 mutation or a strong breast and/or ovarian cancer history found that some women wished their physician provided more guidance and was more directive because this is the type of interaction they are accustomed to in the medical setting (Howard et al., 2011; Klitzman & Chung, 2010). However, the physicians may not have provided the desired straight-forward advice because of the subjectivity and complexity of the decision at hand (Klitzman & Chung, 2010), and as an effort to be non-directive. However, during these interviews, other women had the opposite feeling and expressed that they thought their physicians were too directive and felt pressure from them to make a certain decision. Many participants also felt like the physicians were forceful and insensitive in the manner in which they provided guidance and information (Klitzman & Chung, 2010). This pressure can have a positive impact at times, though. A Swiss qualitative study that involved interviews with 32 unaffected BRCA1 and BRCA2 mutation carrier women found that this pressure from physicians can encourage women to follow through with the published recommendations; if they don’t follow the recommendations, they feel as though their physicians think they are irresponsible (Caiata-Zufferey et al., 2015).
Studies have found that risk management recommendations sometimes vary between healthcare professionals, and contradictions of recommendations between physicians can contribute to a feeling of disorientation in the management decision-making process (Caiata-Zufferey et al., 2015). Some carriers, particularly those who seek care in rural areas, feel concerned that there is a scarcity of healthcare providers that are capable of providing guidance for preventive care for BRCA carriers. Some women have to educate their healthcare providers about prophylactic options for BRCA1 and BRCA2 carriers, which makes them feel as though they are not being well taken care of and have to ‘guide the ship’ themselves (Leonarczyk & Mawn, 2015). This lack of information and decisional support from healthcare providers can impede the decision-making process, making it more difficult. Although not as prevalent in the literature, there are also examples of BRCA mutation carriers feeling as though their physician provided appropriate information, support, and decisional help (Howard et al., 2011; Josephson, Wickman, & Sandelin, 2000; Leonarczyk & Mawn, 2015).

In addition to physicians, genetic counselors can also be involved in the decision-making process. A prospective, descriptive, cross-sectional study of 62 women at an increased risk for breast or ovarian cancer showed that the information provided by genetic counselors, such as information about genetic testing and cancer risk, is helpful in surgical decision-making by providing clients with risk information that can be incorporated into their risk perception (Ray et al., 2005). Genetic counseling has also been shown to promote surveillance, preventive surgeries, and early cancer detection in a
large prospective study (Scheuer et al., 2002). However, a qualitative study of 15 women at an increased risk for breast and/or ovarian cancer who had genetic counseling and underwent a RRM showed that 7 out of the 15 participants were satisfied with the factual information they received from genetic counseling, but only 4 were satisfied with the psychological support they received (Josephson et al., 2000).

Although there has been a lot of qualitative research done on the cancer risk management decision-making process among BRCA1 and BRCA2 mutation carriers, there are still gaps in our understanding of this process, especially about the roles of other individuals. It is not clear how important the involvement of others is in the decisional process. Many studies have found ways in which healthcare providers and family members may impede risk management decisions and make it more difficult, but there is a lack of in-depth information about how individuals can assist the decision-making process and what roles they may play. The goal of this study is to gain better insight into the management decision-making process of BRCA1 and BRCA2 mutation carriers and roles individuals play during this process. One-on-one interviews with BRCA1 and BRCA2 mutation carriers were used to determine the factors that influence decision-making and gain to insight about who is involved in assisting these women in making a decision and in what manner. By attaining more information about this complex process, we can better understand the role played by healthcare providers in helping these women come to a decision. Understanding how different individuals are involved in the cancer risk management decision-making process and what their unique roles are can help direct
support and assistance to mutation carriers struggling with making a risk management decision. This could inform development of decision-making aids to ultimately empower women to be confident in their choices, and critical factors involved in the decision-making process in order to develop interventions to aid in the decision-making process and improve decision satisfaction.
Chapter 2: Methods

Study Participants

Potential participants were identified through a clinical database search of individuals previously seen for cancer genetic counseling at The Ohio State University. Female \textit{BRCA1} and \textit{BRCA2} mutation carriers that had been seen for genetic counseling within the last five years were contacted via telephone. To be eligible to participate in the interview, women had to have the ability to read and speak English and have no previous or current diagnosis of ovarian cancer. Prior to participation, participants provided written informed consent. Demographic information was collected from the database that was used for identifying participants and is summarized in Table 1. This study was approved by the Cancer Institutional Review Board at The Ohio State University.

Procedures

Twenty individual interviews were conducted in person or over the phone by three members of the research team (SH, PD, and AP) using a semi-structured interview guide. Interviews were audiotaped and lasted approximately 30-60 minutes. Participants received a $20 gift card for compensation.
The interview guide for this study (Appendix A) was originally designed to obtain information about the best way to communicate personalized risk information; however, participants naturally brought up and discussed risk management decision-making throughout the interviews. Participants were asked to discuss their risk perception, adjustment to cancer risk over time, things they have done to reduce their cancer risk, interest in getting a personalized risk estimate, and how best to communicate personalized risk information. During this discussion many participants talked about the management decisions they have made or are struggling with, the factors that have gone into making these decisions, who has been involved in the decision-making process, and what kind of information carriers feel is helpful for making management decisions.

**Data Analysis**

Interviews were transcribed verbatim by an outside transcription company (Landmark Associates, Inc.) with the removal of all personal identifiers to maintain confidentiality of the participants. Each participant was assigned a pseudonym for identification purposes during analysis. Grounded theory was used to analyze the data in order to allow recurrent themes to emerge from the data itself (Charmaz & Smith, 2003). Codes were developed from topics frequently discussed and organized into major themes to create a codebook. Three authors (AP, JB, SH) independently coded a subset of transcripts using the initial codes that were identified and then met to discuss coding consistency and rework the codebook. Once a final codebook was established, AP and JB coded all transcripts together, discussing discrepancies until 100% agreement was reached for each coding
Table 1: Demographics of the Participants

<table>
<thead>
<tr>
<th>Carriers</th>
<th>n=20</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Gender</strong></td>
<td></td>
<td></td>
</tr>
<tr>
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</tr>
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</tr>
<tr>
<td>30-39</td>
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</tr>
<tr>
<td>40-49</td>
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</tr>
<tr>
<td>50+</td>
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<td>10</td>
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<tr>
<td><strong>Gene Mutated</strong></td>
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<td></td>
</tr>
<tr>
<td>BRCA1</td>
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</tr>
<tr>
<td>BRCA2</td>
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</tr>
<tr>
<td><strong>Cancer History</strong></td>
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<td></td>
</tr>
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<td>40</td>
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<tr>
<td>Other*</td>
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<td>10</td>
</tr>
<tr>
<td><strong>Time Since Genetic Testing</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;1 year</td>
<td>2</td>
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</tr>
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<td>15</td>
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<tr>
<td>Unknown (not mentioned during interview)</td>
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<td>25</td>
</tr>
<tr>
<td><strong>Prophylactic Surgeries Completed</strong></td>
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<tr>
<td>Mastectomy</td>
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</tr>
<tr>
<td>Oophorectomy</td>
<td>11</td>
<td>55</td>
</tr>
</tbody>
</table>

* Other cancers include thyroid and skin
instance. Major themes and subthemes were analyzed for recurrent topics or interesting comments. Analysis and comparisons were done using NVivo (QSR International, Version 10). Pseudonyms are used for participants throughout the paper in place of their real names.
Chapter 3: Results

Participant Demographics

The mean age of participants at the time of the interview was 39 years and it had been an average of 3 years and 4 months since they had genetic testing. A majority of the women (60%) did not have a personal history of breast cancer. Two women were undergoing cancer treatment at the time of the interview, and six had a previous breast cancer diagnosis. At the time of the interview, 8 women had undergone a RRM and 11 had undergone a RRSO. At least 60% of the participants had children, although not all women discussed this during the interview, so it is unknown if more than these 12 women have children (Table 1).

Participants with a current or previous breast cancer diagnosis were more likely to have already made their cancer risk management decision to have prophylactic surgery. Of the 8 women with a breast cancer diagnosis, 50% (n=4) had a contralateral prophylactic mastectomy during their breast cancer treatment process and 75% (n=6) had a prophylactic oophorectomy. Comparatively, 33% (n=4) of women without a breast cancer diagnosis had a RRM and 42% (n=5) had a RRSO. Of participants known to have children, 42% (n=5) had undergone a RRM and 58% (n=7) had a RRSO, compared to 33% (n=1) of participants known to not have children underwent a RRM and 33% (n=1)
had a RRSO. Among women who have not had breast cancer, 67% (n=8) discussed doing breast and/or ovarian screening during the interview. A quarter of woman without a breast cancer diagnosis (n=3) have the intention to have RRM and 25% (n=3) intend to have a RRSO in the future.

Main Study Themes

Three main themes arose from the data regarding decision-making including (a) factors impacting decision-making, (b) perceived ease of decision-making, and (c) individuals involved in the decision-making process.

Decision-making factors.

Many different factors were involved in the cancer risk management decision-making process. Some factors discussed by participants made them lean towards the decision to have a prophylactic surgery, while others were deterrents to prophylactic surgery. These factors were viewed as possible reasons a participant may decide to make a decision, with certain factors possibly having more importance than others, nudging the participant toward making a certain decision. Therefore, even if a participant mentioned a reason to have surgery, that does not necessarily mean she went through with that surgery, just that she viewed that as a factor in her decision-making process.

Common reasons to have prophylactic surgery discussed by participants included the increased cancer risk, a desire to prevent or avoid cancer, the desire to reduce cancer
worry, and the feeling that there was no choice to make because of a cancer diagnosis (Table 2). Increased risk was the factor mentioned most often by participants as a reason to have a prophylactic surgery. This was an especially important decision-making factor for women who have not had breast cancer. Women with a history of breast cancer often felt more at risk because of their cancer history rather than their BRCA mutation. Maggie (52, breast cancer) said, “I already know that my body knows how to make breast cancer, so I figured that’s probably there in my future.” Women who have had breast cancer were more likely to say that a healthcare provider’s recommendation played a role in their prophylactic surgery decision.

Reasons not to have surgery that the participants discussed during the interviews included the feeling that prophylactic surgery is drastic, timing of the surgery does not work, changes to the body and sexuality, and not being recommended to have surgery due to age or cancer history (Table 2). The most common reason not to have a prophylactic surgery cited by participants was that surgery is viewed as a drastic measure. Of the five women who talked about this decisional factor, four viewed RRM as being a drastic surgery, while only one participant (Christina, 33, no cancer) felt that RRSO was drastic. She felt that RRSO was drastic because of the associated side effects that can affect quality of life; for this reason she was considering removing her fallopian tubes only. Women who felt RRM was drastic had no intentions to have a mastectomy in the future, but all of them had already had a RRSO. One participant originally had plans for a RRM,
but has not followed through with the surgery, and does not have intentions to do so in the future:

I was fine because I was like, “I have a plan. I’m just gonna have a mastectomy; I’m gonna have my ovaries out. Everything’s great.” It’s over three years later and I still haven’t had a mastectomy because it freaks me out. (Cindy, 45, thyroid cancer)

Timing was another common reason given by participants to not have prophylactic surgery, but 75% (n=3) of those who discussed this factor also talked about planning to have surgery in the future. Timing was an issue for these women because they wanted to wait to have prophylactic surgery until they are older (either reach a specific age or have children), are currently undergoing breast cancer treatment and do not have time for RRSO, or because they have young children that would be difficult to care for after surgery. Changes to body image and sexuality, as well as the side effects of RRSO, were important factors that made participants hesitant to have prophylactic surgery. Several participants felt as though having prophylactic surgery didn’t feel right and they were not emotionally or mentally prepared for it. One participant (Cindy, 45, thyroid cancer) talked about how she did not feel as though prophylactic surgery was necessary, and she felt no rush to do it because there may be better surgical options available in the future: “Honestly, I’m kinda waiting to see if it gets better cuz I’ve got some time to wait and think.”
<table>
<thead>
<tr>
<th>Reasons to have surgery (n=17)</th>
<th>Example</th>
<th>Reasons not to have surgery (n=13)</th>
<th>Example</th>
</tr>
</thead>
<tbody>
<tr>
<td>Risk (12)</td>
<td>&quot;I think there's a high likelihood [of getting cancer]. I understand that if I don't start chopping off body parts, the risk is high.&quot; (Emily, 30, no cancer)</td>
<td>Drastic (5)</td>
<td>&quot;It’s really hard for me to then take this next step of having a prophylactic mastectomy, which I just—it’s such a major surgery and it’s such a life-changing thing.&quot; (Cindy, 45, thyroid cancer)</td>
</tr>
<tr>
<td>To prevent/avoid cancer (6)</td>
<td>&quot;I don’t want to deal with this [cancer] again in three or four years.&quot; (Debbie, 49, breast cancer)</td>
<td>Timing (4)</td>
<td>&quot;I have made the choices like having my ovaries removed and then I’ll be 39 in February, so I’m gonna try to have my breasts removed by the time I’m 40, if the time allows.&quot; (Tiffany, 38, no cancer)</td>
</tr>
<tr>
<td>To reduce cancer worry or because cancer is deadly (6)</td>
<td>&quot;You can't run away from it. It's just not something that I really wanna experience. It's not how I want to pass away.&quot; (Sarah, 32, no cancer)</td>
<td>Side effects, body changes, sexuality changes (4)</td>
<td>&quot;I went through the grieving process cuz I knew that I would be losing a part of my body.&quot; (Allison, 36, no cancer)</td>
</tr>
<tr>
<td>It feels right or gives comfort (3)</td>
<td>&quot;I went ahead and had a double mastectomy. That gives me some comfort in knowing that it’d be really rare for me to get cancer in a breast again, because there are just no breasts.&quot; (Abby, 48, breast cancer)</td>
<td>Practicality (not recommended or there is still a cancer risk after surgery) (3)</td>
<td>&quot;I'm not really eligible [for surgery] because its [the cancer] spread to my liver and nodes, and all that.&quot; (Shannon, 36, breast cancer)</td>
</tr>
<tr>
<td>It was recommended (3)</td>
<td>&quot;I had my ovaries out because my oncologist said, 'That's a no-brainer. Sign up right now,' which I did.&quot; (Alice, 59, breast cancer)</td>
<td>It doesn't feel right, not emotionally or mentally prepared for surgery (3)</td>
<td>&quot;I mean, I'm not emotionally ready&quot; (Nicole, 40, no cancer)</td>
</tr>
</tbody>
</table>
Participants also talked about reasons for or against doing cancer screening, although this was discussed less frequently than prophylactic surgical decision-making factors.

Screening was often considered by participants as a way to achieve early cancer detection. This reasoning that screening will lead to early detection was most important for women who have not had cancer, while women with a cancer history primarily discussed doing screening because it was recommended for them by their physician or genetic counselor. Participants also discussed increased cancer risk was a factor in deciding to do cancer screening instead of or until surgery. Less commonly discussed reasons to do cancer screening included feeling content with the screening process and as though they are in good hands, and as a management option to use until they decide to have prophylactic surgery. Although not discussed often during the interviews, anxiety and worry created from going to screening appointments was discussed as a reason to not do screening by a few participants. For example, Allison (36, no cancer) said, “I just didn’t really wanna go [to screening appointments], and then getting that risk of—oh my gosh, are they gonna find it this time or not? I just wanted to be proactive.” Only one study participant, Jessica (24, no cancer), mentioned not screening for ovarian cancer because of not knowing what surveillance practices need to be done or what type of physician needs to be seen.

Many participants talked about how they felt a need to be behaviorally proactive by taking action against their cancer risk after genetic testing. These women have a strong
desire to do whatever they can to prevent cancer or achieve early detection. Family history was a particularly important in the need to be proactive for Sarah (32, no cancer), who was a caretaker for two different family members as they went through cancer treatment. After watching her grandmother go through ovarian cancer and her mother go through breast cancer, she felt a need to be proactive so as to not end up in their shoes someday:

I'm definitely a person that faces things head on, and if there's anything I can do to take that risk down and to lower my chances of getting it. I guess it's just the experience of watching a person from when they first got diagnosed with the cancer to when it takes their life, that time period, it really makes you think, “Okay, I need to do everything I need to do so that doesn't happen to me.”

**Ease of decision-making.**

Forty percent (n=8) of women commented on how they had a difficult time making a risk management decision, 30% (n=6) felt it was an easy decision to make about prophylactic surgeries and/or surveillance, and 30% (n=6) did not comment on their ease of decision-making. Those who expressed that the process was easy often made their management decision very quickly. Many times these women said the decision felt like a “no-brainer” or they felt as though they would be crazy not to have a prophylactic surgery. Kristy (36, breast cancer) said, “It was truly, it wasn't even like I had to make a decision. It was done, it was what I had to do.” However, the ease of decision-making was not always the same for breast and ovarian cancer risk management. More women found the decision to
have a RRSO to be easier than the RRM decision, saying that the RRSO decision was simple, but RRM is difficult to mentally prepare for.

Participants who not had breast cancer often had a harder time coming to a cancer risk management decision, with 50% (n=6) saying the decision was difficult. A majority of the women who had a difficult time with this process noted how it was a really “tough” decision, such as Christina (33, no cancer), who said “[My decision] was very—it’s been very calculated. It’s been tough, honestly.” Several participants talked in detail about their decision being very complex with many different factors and components to consider during the process. A few women felt it was an emotionally hard decision to make.

Seeking out information about prophylactic surgical options, surgical outcomes, and risk information was common among women who described a difficult decision-making process. Of the eight women who described a challenging decision-making process, seven of them sought out surgical and risk information on their own. Only one of the six women who said that decision-making was easy (Emily, 30, no cancer) talked about actively seeking out information to help with decision-making. However, she was also the only individual to talk about the decision to do screening as being simple, yet she did not mention if prophylactic surgery decision-making was difficult or easy. The most common type of information sought out by participants was information about cancer
risks, information on surgical procedures and outcomes, and information from other people about their personal experiences with cancer or management options.

Half of carriers who noted difficulties with making a cancer risk management decision also had negative emotions related to decision-making, such as fear, anxiety, and frustration. Many women who expressed negative emotions about making a decision were not thrilled about, upset by, or overwhelmed with surgery or the management options available to them. Diane (48, breast cancer) was upset and overwhelmed by her physician’s change in surgical recommendations during her cancer treatment after seeing her genetic test results and felt forced into making a decision by her physician:

"Okay. You have to have now [have] a double mastectomy and your ovaries removed." I was really overwhelmed. It went from being, like, "Okay. It's a really small tumor. It's stage I. It's no big deal," to now she's not even giving me a choice and saying, "Okay. Now you've gotta get it all done."

Several participants were upset with the side effects of RRSO or RRM and were grieving the future loss of body parts before having gone through with RRM. Nicole (40, no cancer) had a very difficult time with surgical decision-making as she was having a tough time with the idea of having a mastectomy; “I miss them [my breasts] already.” A couple of women talked about being scared of going through a major surgery and the associated pain during the healing process.
Individuals involved in the decision-making process.

Of the 20 participants, 15 (75%) talked about the involvement of other people in their risk management decision-making process. For these women who discussed other individuals involved in their cancer risk management decision-making, each party typically played a different role in the process and were used for different types of support. Often physicians were an active participant in decision-making, providing recommendations and acting as an instigator to take action. Genetic counselors were a short-term provider of information and recommendations. Family members and other carriers/support group members filled a similar role of providing support and information about personal experiences (Figure 1). About half of the participants who discussed involvement of other people in decision-making talked about only one other party playing a role, and about half used two different parties in their decision-making process. When women talked about having more than one party involved in decision-making it was mostly physicians and family members/other carriers that played a role in the process, while several participants involved physicians and genetic counselors together (Table 3).

A majority of the participants had a positive involvement of other people in their decision-making process. One-third (n=5) of women who involved others in their decision had an individual that was not useful in their decision-making process by not being supportive. However, all 5 these women were able to fill this lack of support by involving a second party in their decision-making process. Most commonly they reached
out to a healthcare provider to help with decision-making when they had a lack of positive support from family members or a different healthcare provider (physician or

Figure 1: Involvement of Others in the Management Decision-Making Process

Data from this study suggests that physicians are most involved in the management decision-making process of BRCA1 and BRCA2 mutation carriers by recommendations and support. Family members and other mutation carriers are also involved in the decision-making process by providing support and information about their own experience with surgeries, screening, and/or cancer. Genetic counselors were found to be less involved in decision-making by being a short-term provider or information and recommendations.
genetic counselor). Women who received only positive support and involvement from other people were less likely to discuss utilizing two parties in decision-making.

**Physicians.**

Half of participants (n=10) discussed physicians playing an important role in the decision-making process (Table 3). Physicians often had an active role in helping these women come to a decision. Participants talked about having trust in their physicians and consulting with and involving them in the risk discussion:

I had doctors who were willing to really talk it through with me, not just the numbers but the experience, I felt like it could hand it over to them. I'm going to my appointments. I'm taking my medicines. I'm having my MRIs and my mammograms. I'm doing everything. They're watching me. I can live my life.

(Alice, 59, breast cancer)

During the decision-making process, participants often sought surgical information and reassurance from various physicians, such as oncologists, gynecologists, breast specialists, and plastic surgeons, to learn about the opinions and options available from each physician:

I spent a lot of time going to multiple doctors after this like OBGYN, the breast specialist, the plastic surgeon. I mean you kind of need reassuring that you're doing the right thing because you're not given a cancer diagnosis, but you're also being told, though, you have a very high risk of it. For me, I needed a lot of reassuring that what I was getting ready to do was the right choice. (Brooke, 36, no cancer)
Table 3: Individuals Involved in Decision-making

<table>
<thead>
<tr>
<th>Participant</th>
<th>Age (y)</th>
<th>Cancer History</th>
<th>Physician</th>
<th>Genetic Counselor</th>
<th>Family</th>
<th>Other Carriers</th>
<th>Ease of Decision-making</th>
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<td>59</td>
<td>Breast</td>
<td>X</td>
<td>X</td>
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<td></td>
<td>RRM hard</td>
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<td>Allison</td>
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<td>Easy</td>
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<td>Christina</td>
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<td>X</td>
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Support from physicians was important for the decision-making process for one-third of the women who discussed the utilization of physicians in making a management decision.

The most common way physicians were involved in decision-making was by providing management recommendations. Physicians were able to take the risk information and
published management guidelines to help the women interpret what it means in the context of their lives. Alice (59, breast cancer) discussed how her physician was important in assisting in decision-making because he was able to put the risk and surgical information into context of her history and experiences: “There's certain things you need. You need to have the statistics, but I can't imagine somebody not going back to their physician that knows them, not just somebody who's reporting a statistic.” Helping to interpret what the risk information means helped these women determine what they should do and come to a decision.

In providing recommendations, participants viewed some physicians as having a discussion with them about the risk management options and other physicians as being more directive about what risk management measures need to be taken. This advice was often received positively, even if women were not thrilled about the recommended action. For example, Emily’s (30, no cancer) physician helped her realize the benefits of having a prophylactic mastectomy:

I've had advice from a couple of doctors… and I know it [mastectomy] needs to happen. I'm not thrilled about it, but I guess, especially once you have kids, if you're thinking that you wanna be around for a while, it's like, “Maybe I don't need my boobs anymore.”

Instead of just providing opinions, some physicians pressured the participants into making a certain decision: “[my physician] really was like, ‘okay, so when do you wanna
do this?’ It wasn't like, ‘If you do this.’ It was ‘When?’” (Sarah, 32, no cancer). Except for in one situation, the participants were fine with this pressure; the one woman who was not happy with the recommendations from her physician (Diane, 48, breast cancer) switched to a different physician who supported her decision. Only one woman (Jessica, 24, no cancer) mentioned that her physician did not know what to recommend for ovarian management, so she was not doing ovarian screening and did not mention if she was considering a RRSO. All other participants talked about their physicians being helpful in the decision-making process.

Physicians were more involved in the decision-making process if women were struggling with a surgical decision. Of the women who had a difficult time coming to a decision, 75% (n=6) discussed utilizing their physician in the decision-making process. However, only 33% (n=1) of women talked about involving their physician in a management decision with which they did not struggle.

**Genetic counselors.**

While physicians were involved throughout the decision-making process and often played an integral role, genetic counselors were generally viewed as a short-term provider of information. Five participants discussed the involvement of their genetic counselor in decision-making (Table 3). For all of these women, the genetic counselor provided information and recommendations, which involved cancer risk numbers as well as the management options available to them. A few participants received information
about online risk calculators from the genetic counselor, which was found to be very useful in gaining a better understanding of cancer risk.

Although the participants did not talk about the genetic counselor being involved throughout the decision-making process, a majority of women had a positive experience with their genetic counselor. The information and support provided by the genetic counselors was found to be helpful for the participants:

[The genetic counselor] couched the conversation in a way that said, ‘Here’s what your future looks like, and here’s what you can do to plan for it, and how you can prevent some of these things.’ I felt like it was a positive conversation (Maggie, 52, breast cancer).

**Family and other BRCA1 and BRCA2 carriers.**

Family members and other mutation carriers were used mainly for support and as providers of information about their personal experiences with risk management options and cancer during the decision-making process. A total of eight participants talked about the reliance on family and/or other carriers in coming to a decision (Table 3). Discussion with family members and/or other carriers was especially important for previvors, as only one breast cancer survivor talked about the involvement of family in her decision making. Women who had a difficult time with the decision-making process were also more likely to involve family or other carriers in the process; 50% (n=4) of women who struggled with decision-making received input and/or support from family members and/or other
carriers with regards to the surgical decision they struggled with, while only 33% (n=1) involved family or other carriers in management decision with which they did not have difficulty.

The most common way family members and other carriers were involved in decision-making was by providing support. Both family and unrelated carriers provide social and emotional support during the decision-making process, with carriers having a more important role in this realm. Other mutation carriers provide comradery and reassurance that everything will be alright:

A virtual person on the computer can't hold my hand, and say, “It's gonna be all right. We'll get through this, and you have options.” Just to help you deal with the emotions of it. I mean, people need the human touch. I really feel that, especially in this situation. I mean, yeah these women I'm in support groups with, it's hard. We help each other, but still how many of them, I can tell you, feel so lonely (Lauren, 43, no cancer).

The type of support most often provided by family members was support and understanding of the decision the woman has already made. Allison (36, no cancer) said, “My family overall was very supportive, and completely understood my thought process and my rationale for wanting to do it; very, very supportive.”

Only two participants discussed a negative involvement of family members in their decision-making process. Cindy (45, thyroid cancer) was pressured by her sister to have a
RRM, which makes her feel worried about her cancer risk and the decision she has made to not have a prophylactic mastectomy. Sarah (32, no cancer) has decided to have a RRM and RRSO, but her father is not supportive of this decision:

My dad asks me all the time cuz he's not happy that I've made a decision to just get rid of everything. He always says to me, “But what will make you a woman?”
I say, “The way I think will make me a woman. The fact that I am a woman. Breasts and ovaries don't make you that.” He's not happy about it.

Another important role of unrelated carriers and family members discussed by participants was the information about personal experiences they can provide. The experience of watching family members go through a cancer diagnosis or prophylactic surgery provides valuable information. Seeing or helping a family member battle cancer provided the impetus to be proactive and want to make a decision to prevent cancer for several participants. Gaining information about the surgical process, healing time, and side effects from people who have already gone through it is extremely important as well. Observing the effects of prophylactic surgery on family members or seeking out information and guidance from other carriers helped participants realize what the surgical process may be like for themselves. Sarah (32, no cancer) talked about how watching her mother undergo prophylactic surgery provided her with information about the difficulties of recovery: “I know exactly what's in store for me for the recovery. I don't obviously know how painful it's going to be because my mom couldn't articulate that, but I know how tough it's going to be.” Christina (33, no cancer) spoke with other carriers at support
groups about RRSO side effects: “I’ve actually talked with other women in some of these support groups, and they’re like, ‘Oh my God. It was a nightmare. I was clinically depressed for six months,’ or ‘My sex life has never been the same.’” This feedback from other carriers has played a role in her considering to have just her fallopian tubes removed rather than a complete RRSO.
Chapter 4: Discussion

The data from this study elucidates the complexity of the management decision-making process for \textit{BRCA1} and \textit{BRCA2} mutation carriers. Some of these carriers have a much easier time making their management decision than others. There are many different factors that can be involved in this process, which may nudge a woman towards or away from having a prophylactic surgery or doing continued cancer surveillance. Many different people can be involved in helping with this decision-making process. Physicians often provide support and recommendations while genetic counselors serve as a short-term provider of risk information and management recommendations. Family members and other mutation carriers provide information about their experience with management options and decisional support. It is important that \textit{BRCA1} or \textit{BRCA2} mutation carriers have positive involvement from one of these parties, especially if they are having difficulty coming to a risk management decision.

The risk of getting cancer was frequently talked about as a reason to have prophylactic surgery among participants. This concern may have emerged as a prevalent theme given the nature of the interviews. The interview guide consisted of several questions about cancer risk perception, which could have brought cancer risk to the front of the participants’ minds. However, other studies have shown that feeling at high risk for
cancer can have an important impact on the decision to have a RRM or RRSO (Haroun et al., 2011; Hatcher et al., 2001).

Our data showed that the underlying risk motivation for prophylactic surgery was different for those who have had cancer and those who have not. Women who have had breast cancer do not necessarily feel at increased risk because of the *BRCA* mutation, but rather because of their history of cancer. These women were more likely than previvors to cite physicians’ recommendations/suggestions as a reason for their screening or prophylactic surgery decision. This could be because women with a cancer diagnosis are faced with many different decisions to make in a short amount of time after their diagnosis. This may be overwhelming for some and lead to following whatever their physician suggests. Previvors are not necessarily inundated with time-sensitive management decisions at the time of genetic testing, so they may be more likely to consider multiple factors besides their physicians’ recommendations when making a cancer risk management decision. Women who had genetic testing after their breast cancer diagnosis and treatment also discussed physician recommendations as being important in their decision making process. This could be because they developed a relationship with their physician during their cancer treatment and have trust in their recommendations.

Another qualitative study found differences in reasons to have prophylactic surgery between previvors and *BRCA* carriers who had cancer, like we did. Many of those with a
cancer diagnosis were driven to prophylactic surgery as part of their diagnosis and treatment (Hesse-Biber, 2014). Genetic testing for women diagnosed with breast cancer as a method to help direct treatment has been found to be acceptable and useful to women with breast cancer in a qualitative study of 26 women. This study found that genetic testing often serves as an important tool to help guide decision-making, with the majority of these women likely electing contralateral RRM if they were found to have a mutation because of the risk of contralateral breast cancer (Zilliacus et al., 2012). Of the eight participants in our study with a history of breast cancer, six had their genetic testing during their cancer treatment, and four ended up having a bilateral mastectomy during the treatment process. Therefore, knowing about a *BRCA1* or *BRCA2* mutation often has an impact on how women with breast cancer elect to manage future cancer risks.

The data from our study found that women who had not had breast cancer were more likely to have had a difficult time with management decision-making. Another study also found that previvors felt less prepared to make a cancer risk management decision and had more decisional conflict that *BRCA* mutation carriers with cancer (Connors, Voian, Shi, Lally, & Edge, 2014). This is likely because previvors face more uncertainty because they are unsure whether they will actually even get cancer in their lifetime. Participants in our study and others have expressed that they feel as though they have many different surgical options, and they feel as though they have time to think about and weigh their options before making a decision, which can complicate the decision-making process as well (Hesse-Biber, 2014). Women who have difficulty making a decision about RRSO
have been found to be less satisfied with their decision than women who did not struggle with the decision (Westin et al., 2011). Therefore, unaffected *BRCA1* and *BRCA2* carriers may need more decisional assistance than women who have had cancer.

For many of the women who participated in our study, the decision to have a RRSO was often easier and the surgery was considered less drastic than RRM. This finding is expected due to the way these women were counseled. Since RRSO is a standard published cancer risk management guideline for *BRCA1* and *BRCA2* mutation carriers, the discussion of RRSO is often framed in a way to show the importance of this surgery due to the difficulty of detecting ovarian cancer at an early and treatable stage. This is in contrast to RRM, which is presented as an option for breast cancer risk management, not as an essential surgery. Because RRSO is presented and viewed as a non-optional surgery by healthcare providers, the participants in this study likely did not view RRSO as an optional surgery, either. However, RRM may have also had many ambiguities surrounding it for participants, such as whether or not to have a RRM, when to have it, and what type of prophylactic mastectomy and reconstruction to undergo. Research has shown a higher rate of uptake of RRSO compared to RRM, suggesting that the RRSO decision may have been easier to make for the women in these studies as well (Botkin et al., 2003; Claes et al., 2005; Ray et al., 2005; Singh et al., 2013), although this was not the case in all studies (Lynch, Snyder, Lynch, Riley, & Rubinstein, 2003). However, these studies did not directly discuss the ease of decision-making for the participants, just the decisional outcome.
We found that different groups of people tend to play different roles in assisting \textit{BRCA1} and \textit{BRCA2} mutation carriers in the decision-making process. Genetic counselors were found to mostly have a brief involvement in decision-making around the time of genetic testing by providing risk information and relaying published management recommendations. This is to be expected, given that participants were counseled in a setting that did not provide ongoing follow-up genetics appointments. However, the information received from genetic counselors is still helpful in decision-making. A retrospective study found that women who received a genetic consultation for HBOC felt prepared to make management decisions and had low decisional conflict after pre-test counseling and results-disclosure, suggesting that genetic counseling assisted the decision-making process (Connors et al., 2014). The participants in our study used the information from their genetic counselor to help make a decision. Some took the information from the genetic counselor and interpreted it into their own life context, and some women talked with their physicians about risk and management options, which is standardly recommended in follow-up counseling notes.

Physicians were very involved in the decision-making process and served as providers of information, recommendations, and support for the participants of this study. A majority of the participants in this study had a positive experience with the involvement of their physicians in the decision-making process. They felt as though their physicians provided support and reassurance with their decision. In the instances where the physicians were
applying pressure to make a certain decision, most of these women appreciated the pressure and understand why their physician was pushing them to make this decision. Only one participant (Diane, 48, breast cancer) was not happy with the recommendations and pressure from her physician and decided to switch to a different physician. Previous qualitative studies have found that many women were not satisfied with the amount of directive input from their physicians, with some wanting more input and others feeling as though their physicians applied too much pressure (Howard et al., 2011; Klitzman & Chung, 2010). Our data shows that the majority of these women did not express similar feelings with regards to input from physicians. This could be because they were content with the input from their physicians. However, it could also be because they were not asked directly about their thoughts on the directive nature of their physicians’ recommendations during the interview, and therefore these feelings were not expressed, which could be the case for 10 participants.

Unlike other studies that have found a lack of knowledge about HBOC management among some physicians, only one participant in our study talked about her gynecologist not knowing what to do for cancer risk management. Other studies have found that physicians sometimes provide contradicting information and recommendations or that physicians in rural areas may not know how to manage women with a BRCA1 or BRCA2 mutation, causing carriers to have to educate their physicians about preventive options (Caiata-Zufferey et al., 2015; Leonarczyk & Mawn, 2015). The difference in findings between our study and others may be because the participants in this study received
genetic counseling at a NCCN designated tertiary care cancer center with an academic focus and high awareness of HBOC.

Our data suggests that family members and other \textit{BRCA} mutation carriers serve a similar role in the decision-making process by providing decisional and emotional support and experiential information regarding surgery, screening, and cancer. Support can be very important in the decision-making process, both from family and support groups (Hesse-Biber, 2014; Leonarczyk & Mawn, 2015). Lack of confidence has been found to be associated with having difficulty adapting to the risk associated with HBOC (Heiniger, Price, Charles, Butow, & kConFab Psychosocial Group on behalf of the kConFab Investigators, 2015), but familial support has been found to decrease general distress, increase self-esteem, and make the decision-making process an empowering experience (den Heijer et al., 2012; Hesse-Biber, 2014). Therefore, having support from family members may increase confidence and ease the decision-making process. We found that unrelated mutation carriers can also fill a similar support role. A different qualitative study highlighted a woman who did not have support from her family members, especially her father, in her decision to have a prophylactic mastectomy, making her feel alone. This led her to participate in a support group with other mutation carriers in order to receive support from them, suggesting that other carriers can fill the supportive role of family members (Hoskins & Werner-Lin, 2013), as was found in our study. Our data and other studies have found that family members do not always provide mutation carriers with desired support, either by applying too much pressure to make a certain management
decision, or by not being supportive of the decision the carrier has already made (Hoskins & Werner-Lin, 2013). The results from our study and others (Klitzman & Chung, 2010) suggests that the use of support groups and other mutation carriers may be able to help fill the role of family members if they are not found to be useful in the decision-making process.

The few women in our study who discussed having negative involvement from individuals sought out information and support from other people. Within our study population, women often discussed turning to a healthcare provider, most commonly a physician, for this additional decisional assistance in the form of medical information, support, and reassurance if they were not receiving positive assistance from another party. Family members and other mutation carriers mostly provide support, opinions, and personal experience information, but don’t necessarily have the background to provide medical information and recommendations. The versatility of physicians in decisional assistance can make them extremely useful in the decision-making process. A different qualitative study in which many participants were not satisfied with the involvement of their healthcare providers found that mutation carriers often turned to family members and support groups in place of physicians. However, the participants in that study seemed to have a less positive involvement of other people in the decision-making process, both from physicians and family members (Klitzman & Chung, 2010).
Our data showed that physicians, family members, and other mutation carriers seem to play an especially important role in the decision-making process with a surgical decision with which that woman is struggling. Those who have had difficulty coming to a decision have also put an effort into seeking out information. Our data suggests that previvors are more likely to have a hard time with risk management decision-making. Therefore, the results from this study suggest that previvors and women struggling with cancer risk management decisions may need more assistance than other women with the decision-making process by having more information made available to them, referring them to a physician who can assist with management decisions, and by making sure they have social support from family or other mutation carriers. This type of assistance is typically provided in a genetic counseling session, so referral to a genetic counselor may be especially helpful for these women struggling with the cancer risk management decisions.

By better understanding how people are involved in the cancer risk management decision-making process of women with a $BRCA1$ or $BRCA2$ mutation, and by recognizing which women may be struggling with making a decision, we as healthcare providers can best help them with this process. Physicians, genetic counselors, family members, and other mutation carriers can be very helpful in a myriad of ways to women with a $BRCA$ mutation when making cancer risk management decisions. When a mutation carrier is struggling with a risk management decision, healthcare providers can make referrals to specialists and/or advocacy groups to improve their decision-making experience. In this study population, physicians were most commonly described as being
helpful in coming to a risk management decision, however, genetic counselors, family members, and other mutation carriers also provided help in management decision-making. This means it is important that mutation carriers struggling with a cancer risk management decision have positive involvement from one of these parties to aid in the decision-making process.

**Study Limitations**

Since this is a qualitative study of participants from who received genetic counseling from one institution, the results from this study are not generalizable to the broader HBOC population, but rather help provide a better understanding of the decision-making process of *BRCA1* and *BRCA2* mutation carriers. This study was not originally designed to analyze decision-making among mutation carriers. Therefore, these results may not provide a comprehensive assessment of the decision-making process as questions were not specifically asked to probe about the process and experience. Because of this, the same information, such as ease of decision-making and physician involvement in decision-making, was not obtained for every participant. However, since these women were not directly asked about decision-making, yet most women commented on this process, our data suggest that decision making is of concern for participants. Finally, women who decided to participate in this study may represent highly motivated individuals or those who may have had a particularly striking genetic testing and decision-making experience that they wanted to talk about. Thus, they may not be representative of *BRCA1* and *BRCA2* mutation carriers in general.
Practice Implications

Data from this study suggest that physicians, genetic counselors, family members and other mutation carriers all play an important role in the cancer risk management decision-making process. Thus, it is important to ensure that these groups are educated such that they can be supportive or help set up support networks and provide up-to-date information. Each of these group can provide BRCA1 and BRCA2 mutation carriers with various forms of decisional support at different points during the risk management decision-making process. Due to the various roles different groups play in providing decisional assistance, it is important that we have efficient communication between healthcare providers and work together to best help mutation carriers with the cancer risk management decision-making process by providing them with the type of support and information they need.

Research Recommendations

Research needs to be done assessing the role of individuals in the management decision-making process for BRCA1 and BRCA2 mutation carriers on a wider scale to see if these results can be applied to other groups of women with HBOC. Studies can build on the findings presented here and could focus on gaining a better understanding of the relationship between ease of decision-making and the resources and individuals needed to aid in the decision-making process. Eventually, questionnaires can potentially be developed to help identify women struggling with the decision-making process and what
type of support and resources they need to make a decision in order to best allocate the type of assistance individual women need. Decision aids for women with \textit{BRCA1} and \textit{BRCA2} mutations have been developed to help guide women through management decision-making and have been found to be useful in the management decision-making process (Culver et al., 2011; K. A. Metcalfe et al., 2007). However, there is some conflict as to when it would be the best time to use the decision aid, with the thought being that for some people, it may be best to use immediately after genetic testing, but for others it may be more helpful to wait awhile after genetic testing (Culver et al., 2011). Research can be done looking at the decision-making process with healthcare professionals working as a team and providing decisional assistance in combination with decision aids, which may help determine the best timing for use of decision aids for each individual person.


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Appendix A: Interview Guide

Part 1: Perceptions of Risk

Thank you for your willingness to participate in this interview. As I mentioned, we’re talking with BRCA mutation carriers to determine how best to communicate personalized risk information.

To start things out, I would like to ask you a few questions about your experiences with cancer and genetic testing, and your thoughts about your risk for cancer. Please feel free to stop me and ask questions anytime and remember that you can skip any question.

1. How long ago were you tested for the BRCA 1/2 mutation?

2. Have you had cancer? Are you currently undergoing treatment?

3. Do you remember what the genetic counselor estimated your risk for cancer to be when you first got your genetic testing results? Do you remember your initial reactions to that estimate?

4. Do you feel like you have a good understanding of your cancer risk?

5. How susceptible to cancer/another cancer do you feel on a scale of 1-100? Why?

6. Do you feel like you are more or less at risk than women without a BRCA mutation? Why?

7. Do you feel like you are more or less at risk than other BRCA mutation carriers? Why?
8. How do you think have you “adjusted” to your cancer risk over time?

9. What kinds of things have you done to help reduce your cancer risk? Are you doing things now that you weren’t doing before you went through genetic testing?

10. Do you worry about cancer?

Part 2: Risk Communication Preferences

As I mentioned to you at the beginning, the goal of this research is determine how best to communicate refined cancer risk estimates to women with BRCA mutation. To better understand your preferences, I’d like to hear your responses to a few sample scenarios.

1. Let’s say a genetic counselor told you the BRCA mutation carriers have a 50-85% risk of developing breast cancer by age 80. If your genetic counselor could refine that percentage even further just for you, based on your age and variations in your DNA, would you be interested in that number? Why or Why not?

   a. Let’s say that the counselor tells you that you (personally) have an 85-95% risk of developing breast cancer by age 80, which is 10% higher than BRCA mutation carriers your age. Would that information be useful to you? How do you think that information might impact your decision-making?

   b. Now, imagine that the counselor told you that your personal risk of developing breast cancer was much LOWER than BRCA mutation carriers your age (say 30-40% risk versus 50-85%). How might you respond to that information? Do you think that information might that information impact your decision-making?

   c. But, what if that difference was only marginally LOWER (say your risk was 55-65% vs 50-85%)?

   d. Do you like being presented with risk estimates as a range (like 50-85%) or would you prefer a specific percentage (say 88%)? Why or Why not?
Would it matter the size of the range (i.e., a smaller versus a larger range)?

e. Would you prefer to be presented with an estimate of your **full** lifetime risk (from birth to age 80) OR would you prefer an estimate of your **remaining** lifetime risk (from now until age 80)? Would you prefer an estimate to age 80 or should we go higher?

[ask only if they are currently cancer free] What if that estimate was conditional on you being cancer-free by a certain age?

I would now like to hear your thoughts about the best way to present this risk information.

1. If I were going to give you an estimate of your breast cancer risk between now and age 80, would you prefer that I did so using words like “you have a high or low risk of breast cancer”? Or, would you prefer that I used numbers like “you have a 50-85% risk of breast cancer.” Why?

2. Next, I’m interested to know what you think would be the best way to communicate numerical information. Let’s pretend a genetic counselor was going to tell you your risk of breast cancer.

Taking a look at this list, would you prefer that the risk be presented as (1) a percentage, (2) a proportion or (3) in comparison to the general population?

   a) Why? Which of these formats do you think is the easiest to understand?

3. What if the genetic counselor added some *additional* information to the information you just saw. Take a minute to look at this figure and tell me what you think.

   a) What is different about this information from what you saw before?
   b) How might this additional information impact your perception of risk?
c) Would you prefer this format or the one that I showed you a minute ago?

4. Now, let’s say that a computer program could provide you with a more refined estimate of your breast cancer risk similar to what we talked about before. I want to show you two possible formats for how these estimates could be presented, to find out what you prefer.

Here is the first format

Here is the second format

a) What is different about these two formats?
b) Do you think they are presenting the same or different information?
c) Do you have a strong preference for one format versus the other?

Now here is a third way to present this information

What is your reaction to this format? Does it help you better understand your risk relative to what I just showed you?

d) What if the computer program could ONLY tell your risk relative to other people? In other words, what if we couldn’t provide you with the information in the first figure. Would you still be interested in the information?

5. Next, I am going to show you several figures to get your impressions of them.
   a) What is your understanding of this figure?
   b) What is your reaction to this figure?
   c) What do you like about this figure?
   d) What is confusing about this figure?
   e) What is your emotional response to the figure?

6. Of all the figures you just saw, which figure did you like best? Which figure was the easiest to understand? What changes would you suggest to that figure?
7. Now that you’ve seen all these different presentations of risk, let’s talk about the information that was presented. What impact do you think this personalized risk information might have on BRCA mutation carriers? Do you think it could impact their decision-making about behaviors to reduce their risk?

8. Would having additional risk information that was personalized to your age and mutation status make you feel more or less in control of your cancer risk?

9. Who would you like to deliver this risk information? Do you think we could deliver the risk information, following initial genetic testing, using a computer or web-based program? Why or why not?