Genetic Counseling for BRCA1/2 Mutations: Women’s Experiences, Preferences, and Psychosocial Outcomes of Counseling

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Genetic Counseling for BRCA1/2 Mutations: Women’s Experiences, Preferences, and Psychosocial Outcomes of Counseling

Kate Elizabeth Dibble, Ph.D.

University of Connecticut, 2020

One in 8 women will be diagnosed with breast cancer in her lifetime, but only 5-10% of women who are diagnosed have a BRCA1 and BRCA2 (BReast CAncer) genetic mutation. These mutations naturally occur in biological family units, and women with these mutations live with an increased rate of breast/ovarian cancers. The standard of genetic counseling care is individualized counseling, in which one at-risk family member is tested at a time. A new and potentially more relevant genetic counseling approach, family-based genetic counseling, tests all at-risk family members at one time as one cohesive patient group.

The current study explored lived experiences and preferences (i.e., individualized, family-based) of women who have tested positive for these mutations and psychosocial outcomes of genetic counseling.

A sample of 60 BRCA1/2-positive women was recruited through multiple online support groups. Participants completed an online questionnaire outlining demographic characteristics, genetic counseling information, and HRQoL outcomes. A subset of this sample (n=34) were interviewed to gain experiential insight into their genetic counseling experience and preferences for genetic counseling. Six themes emerged: sources affecting perceived risk, preventive concerns and decisions, experiences in healthcare, emotional reactions to genetic counseling, future recommendations, and family support and communication. Three interesting
subthemes were also identified, including 1) “pre-vivor”, how women of this demographic describe themselves to others, 2) “testing intuition”, the idea of knowing one’s genetic test results were positive before receiving them, and 3) the “hard truth” that prophylactic surgeries are the only true option, whereas ongoing surveillance just buy time. Preferentially, women would have chosen family-based genetic counseling instead of the standard individualized counseling if given the choice. Anxiety ($p<.01$) and stress ($p<.01$) were found to be significantly worse in the current sample compared to the general female population. Other HRQoL domains differed, with physical role limitations ($p<.05$), emotional role limitations ($p<.05$), energy ($p<.01$), emotional wellbeing ($p<.01$), social functioning ($p<.01$), and MCS scores ($p<.01$) being worse in the current sample. These findings can inform future research and practice focused on improving women’s experiences and psychosocial health in those being tested for $BRCA1/2$ genetic mutations.

*Keywords: BRCA1, BRCA2, hereditary mutations, breast cancer, ovarian cancer, family-based genetic counseling*
Genetic Counseling for BRCA1/2 Mutations: Women’s Experiences, Preferences, and Psychosocial Outcomes of Counseling

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B.A., University of Connecticut, 2013
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Submitted in Partial Fulfillment of the Requirements for the Degree of Doctor of Philosophy at the University of Connecticut

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2020
Genetic Counseling for BRCA1/2 Mutations: Women’s Experiences, Preferences, and Outcomes of Counseling

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INTRODUCTION

Genetic Counseling and Women’s Experiences

Testing for BRCA1 and BRCA2 (Breast Cancer) mutations, through formal genetic counseling and direct-to-consumer, at-home tests, have been steadily increasing after 2010 (Walcott & Dunn, 2015). Although increasing attention has been paid to providing women with important information to aid in preventive treatment (e.g., prophylactic surgeries, chemoprevention, surveillance), very little attention has been paid to the genetic counseling experience and its associated mental health outcomes. One in eight women (i.e., about 12%) will be diagnosed with breast cancer in her lifetime (ACS, 2019). However, not all women who are diagnosed with breast and/or ovarian cancer(s) have a genetic mutation, called BRCA1/2, that predisposes them to having such cancers. The function of BRCA1 and BRCA2 genes produce tumor suppressor proteins that protect individuals from numerous cancer growths. If these genes are mutated, cancer cells can grow out of control, leading to a significantly higher risk of the cancers discussed above (NHGRI, 2018).

Only 5-10% of the United States population have familial links associated with breast cancer (due to non-genetic mutative factors), and only 1.8% actually have either (or both) BRCA1 and/or BRCA2 mutations (FORCE.org, 2019; Suryavanshi et al., 2017). Although this percentage seems low, the chance of having a mutation on the BRCA1/2 genes is considered rare in the general population. However, BRCA1/2 mutations naturally occur within biologically related family members and have dire consequences for those affected. Unfortunately, mutations such as these are inherited through a dominant gene, with offspring having a 50% chance of inheriting the mutation(s) if one parent is BRCA1/2-positive and an 100% chance if both parents are positive (FORCE.org, 2019; Suryavanshi et al., 2017). Women with BRCA1/2 live with a 69-
72% increased risk of having breast cancer and 11-39% of having ovarian cancer by the age of 70 (beBRCAware.org, 2020). Women living with these mutations also have an increased risk for prostate, pancreatic, and melanoma cancers, but the risk is lower among these compared to breast and ovarian cancers.

The structure of individualized genetic counseling for BRCA1/2 mutations, the current genetic counseling approach in the United States, does not reflect the hereditary nature of these mutations. Individualized genetic counseling involves testing each at-risk family member separately. This disconnect, combined with the overall stress of the genetic counseling experience (Wenzel et al., 2012), have been associated with adverse psychosocial issues relating to anxiety (Madlensky et al., 2017; Nordin et al., 2011; Schwartz et al., 2014) and health-related quality of life (HRQoL) (Harmsen et al., 2015; Tung & Garber, 2018). In addition to the adverse psychosocial issues associated with the genetic counseling experience, extraneous worries including privacy and confidentiality (Miller & Tucker, 2017), perceived utility of testing (Siegrist, 2002), and familial conflict (Gilbar et al., 2016) have also been reported. Furthermore, adverse psychosocial outcomes related to preventive measures following genetic counseling results (i.e., surgeries, chemoprevention, surveillance) have been noted in recent literature, but are not typically discussed in conjunction with the hereditary nature of testing overall. Thus, research is needed to understand the lived experience of women who have tested positive for BRCA1/2 mutations under the current genetic counseling approach to identify gaps in care to improve the experience for women.

The current study examines the lived experiences of women who tested positive for BRCA1/2 genetic mutations through the genetic counseling approach provided to them. Preferences for the approach of genetic counseling (i.e., individualized or family-based genetic
counseling) offered to these women will also be explored, including important psychosocial outcomes (i.e., stress, anxiety, perceived breast cancer risk, HRQoL domains) related to genetic counseling, test results, and preventive measures that occur after genetic test results are shared. Chapter 1 will provide an overview of BRCA1/2 genetic mutations, genetic counseling and associated approaches (individualized and family-based counseling), and associated cancer risk and treatment(s). Chapter 2 will highlight the current literature across the fields related to genetic BRCA1/2 counseling to provide an insight into psychosocial health and BRCA1/2 counseling-related risks. Additionally, the very limited research relating to individualized and family-based genetic counseling for BRCA1/2 will be presented. Chapter 3 will present the mixed-methods research design of the current study, and Chapter 4 will discuss findings of the current research, organized by the three overarching research questions based on an individualized genetic counseling approach, the standard in the United States. Lastly, Chapter 5 will discuss the study’s findings, in response to the existing literature and what these findings may mean for future research and best practices.
CHAPTER 1

OVERVIEW OF GENETIC BREAST/OVARIAN CANCERS

Genetic Breast/Ovarian Cancer(s)

In 2001, the Human Genome Project (HGP) mapped the complete set of human genes, identifying over 20,500 genes (NHGRI, 2018), with the goal of characterizing genetic predisposed disease. Although the entire human genome has been identified, more than 30% of their genetic functions remain unknown (Jung, Kim, & Yi, 2019). Depending on gene location and structure (e.g., mutated or intact), various health conditions may occur, including physical deformities, genetic diseases, and increased risk of certain chronic illnesses, including some cancers. Several types of mutations exist, but only germline (hereditary) mutations are passed from one or both parents to offspring (NIH, 2019). Acquisition of these mutations differ depending on condition and result from one of four causes: 1) a mutation in one gene (monogenic); 2) mutations in several genes (multifactorial inheritance); 3) a combination of genetic and environmental factors; or 4) by damage to the structure of deoxyribonucleic acid (DNA) (NHGRI, 2018).

In the United States, 24 million people are living with inherited genetic mutations. Worldwide, an estimated 280 million people have a genetic condition, whether or not they are aware of their condition (GDF, 2010). The next section specifically looks at BRCA1/2 inheritance pattern and associated breast and/or ovarian cancer risk.

BRCA Inheritance and Cancer Risk

Some breast and ovarian cancers are hereditary, comprised of 1.8% of the general United States population (Lu et al., 2019; Suryavanshi et al., 2017). The National Institutes of Health (NIH, 2019) has established that family history is a woman’s single greatest predictor of
developing breast and ovarian cancer. In 1994, the \textit{BRCA1} gene (for BReast CAncer gene) was discovered, and in 1995, the \textit{BRCA2} gene was identified, suggesting an increased risk for breast and ovarian cancers in persons who have these mutations (NIH, 2019). Genetic counseling for suspected \textit{BRCA1}/\textit{2} mutations is only recommended for individuals with a high risk of having a mutation (Susan B. Komen, 2019; US Preventive Services Task Force, 2019). Qualification for genetic counseling (and insurance coverage) depends on various demographic, personal, and clinical variables. Having a family member test positive for \textit{BRCA1}/\textit{2} mutation(s) remains the most considerable risk factor or having family members who have been diagnosed with breast/ovarian cancer(s) at age 50 or younger. Personal histories of such cancers also place individuals at-risk, especially those who were diagnosed at age 45 or younger, and/or if they had cancer in both breasts, ovaries, pancreatic, or prostate (if the individual is male). Type of breast cancer also notably increases risk, especially having a history of triple-negative cancer (a type of breast cancer that is estrogen-receptor negative, progesterone receptor-negative, and HER2-negative) diagnosed at age 60 or younger. Certain ethnicities, such as being of Ashkenazi Jewish heritage also places one at an increased risk of having a \textit{BRCA1}/\textit{2} genetic mutation (Colombo et al., 2018; US Preventive Services Task Force, 2019). Therefore, each person within a family system has their own unique chance of inheriting \textit{BRCA1}/\textit{2} mutations, especially if another family member has tested positive, as families share some, not all, of their genetic code (Godet & Gilkes, 2017; Susan G. Komen, 2018).

Although \textit{BRCA1} and \textit{BRCA2} mutations are usually mutually exclusive events, but in extremely rare cases, they can co-occur. When this happens, the risk for breast/ovarian cancers increases threefold (Martin et al., 2006). With approximately 70\% of the function of the human genome fully understood (Jung, Kim, & Yi, 2019), there remains a possibility that additional
genetic involvement may be linked with breast and/or ovarian cancers that have yet to be discovered. For instance, \textit{BRCA1} and \textit{BRCA2} mutations reside on dominant genes, meaning that only one mutated gene from either parent is required to pass along the mutation to their son or daughter. The dominant nature of both \textit{BRCA1} and \textit{BRCA2} indicate a 50\% chance of inheritance for each child if one parent has a \textit{BRCA1/2} mutation, and a 100\% chance if both parents have the mutation (MSKCC, 2019; Scalia-Wilbur, Colins, Penson, & Dizon, 2016).

Although inheritance is the same between \textit{BRCA1} and \textit{BRCA2} mutations, cancer incidence differs. Individuals who test positive for \textit{BRCA1} mutations have an estimated 85\% chance of developing breast cancer and about a 44\% chance of developing ovarian cancer by age 70 (ACS, 2019; Kotsopoulos, 2018). Breast cancer risk associated with \textit{BRCA2} is similar to that of \textit{BRCA1}, but with a 16\% to 27\% increased risk for ovarian cancer over one’s lifetime, in addition to increased risks for pancreatic and melanoma cancers (Rosenberger et al., 2017). By age 70, seven out of 100 women \textit{without BRCA1/2} mutations will develop breast cancer compared to 45 to 65 per every 100 women amongst those who are \textit{BRCA1/2} positive (Godet & Gilkes, 2017). Exceedingly, the prevalence of \textit{BRCA1} and \textit{BRCA2} mutations remain heightened amongst specific ethnic groups. For instance, \textit{BRCA1} mutations are more common in Ashkenazi Jewish (8-10\%) and Hispanic (4\%) populations, whereas \textit{BRCA2} mutations are more prevalent in African American (3\%) and White (2\%; non-Ashkenazi Jewish) populations (Malone et al., 2006; NCI, 2018). Expanded preventive screening, in conjunction with genetic counseling, is imperative for women of these ethnic backgrounds, especially those who have a family history of breast and/or ovarian cancers.

\textbf{Screening and Risk Management}
For women who test positive for a BRCA1/2 gene mutation, imminent cancer risk remains at the forefront of their minds. Although each mutation and case are considered unique, treatment, management, and/or disease prevention differ among women. Following a positive genetic test result for one (or both) of these mutations, four clinical steps are suggested: 1) screen for early or current cancers on a biannual basis; 2) reduce cancer risk via prophylactic surgery or chemo-preventive treatment; 3) assess reproductive options (if of age); and 4) identify further risk to relatives and offspring (Petrucelli, Daly, & Pal, 2016; Rojas et al., 2019). For premenopausal women younger than 50 who are BRCA1/2-positive, most physicians would introduce reproductive options such as implementing fertility and mature oocyte cryopreservation for future pregnancy decisions (Chan et al., 2017; Pruthi, Gostout, & Lindor, 2010). All women with BRCA1/2 mutations should obtain clinical breast exams and breast magnetic resonance imaging (MRI) with contrast (or mammogram) every 6 to 12 months beginning at age 25, have an annual (or biannual) breast MRI with contrast and mammogram from ages 30 to 75, and consider a yearly 3D mammogram if available (Chan et al., 2017; Pruthi, Gostout, & Lindor, 2010). Increased screening among this population is necessary to identify early cancer growths for early treatment response to maximize survival and health-related quality of life (HRQoL).

Following a positive genetic test result, prophylactic health decisions remain the most recommended form of cancer prevention. The genetic counseling approach among BRCA1/2-positive women is to implement a combination of preventive treatments (i.e., chemoprevention) and prophylactic surgeries that greatly reduce the risk for breast/ovarian cancers (Morgan et al., 2018). Patients are given a number of options, dependent on their unique case, including double- or single-mastectomy and/or taking daily medication to reduce the growth of breast cancer.
abnormalities, known as chemoprevention (e.g., tamoxifen, raloxifene, and/or aromatase inhibitors [AIs]) (ACS, 2019; Pruthi, Gostout, & Lindor, 2010; Walker, Jacobson, & Sobel, 2019). Ovarian cancer risk management is often managed using oophorectomy, salpingectomy, and hysterectomy surgeries where the ovaries, fallopian tubes, and/or uterus are removed, respectively. These surgeries are usually recommended between ages 35 and 40 or upon completion of childbearing, and coincide with annual transvaginal ultrasounds (Chan et al., 2017; Pruthi, Gostout, & Lindor, 2010). Tertiarily associated cancers, such as pancreatic and melanoma cancers, do not have specified guidelines for surveillance (Pruthi, Gostout, & Lindor, 2010), but prevention and screening are uniquely based on genetic mutation (BRCA1, BRCA2, CHEK2 [a rarer female breast cancer gene]) as well as personal and family history. Dependent on individual characteristics, treatment and risk management strategies can vary drastically from case to case.

Screening for BRCA1/2 mutations allows genetic knowledge to guide the way for prevention, treatment, and surveillance strategies (Heshka et al., 2008). Unfortunately, it is usually not until symptoms or diagnostic patterns within a family emerge (often 3-4 family cancer diagnoses) that genetic counseling is undergone. Even then, most genetic services prioritize genetic counseling for the individual who is most “at-risk”, or the individual who has had multiple related cancer diagnoses. Focusing on one family member, therefore, may allow other “at-risk” members to be overlooked (GDF, 2010; Griffin et al., 2020). For women diagnosed with cancer because of these mutations, treatments are determined on a case-wise basis. Commonly, risk reduction strategies often occur immediately after genetic test results in succession, implementing biannual surveillance (i.e., ultrasounds, mammograms, pelvic exams, etc.), surgeries, and daily chemoprevention. Such risk management strategies are combined to
protect against future breast/ovarian cancer growth and spread, thus reducing cancer mortality and improving HRQoL. Due to the physical, emotional, social, and invasive impact of receiving genetic test results, additional side effects such as increased stress (Wenzel et al., 2012), familial conflict (Brunstrom, Murray, & McAllister, 2016), and nervousness/anxiety (Madlensky et al., 2017; Nordin et al., 2011; Schwartz et al., 2014), not relating to cancer itself, can surface. In response to the life-changing course that these mutations impose, there exist adverse, but highly variable, physical, psychological, and social consequences.

**Treatment and Recurrence**

Women who are *BRCA1/2*-positive and have been diagnosed with breast/ovarian cancers are likely to have a more difficult treatment experience regarding reaction to chemotherapy, radiation, and surgery (Godet & Gilkes, 2017; Susan G. Komen, 2019). Moreover, these women have a 25% to 30% increased chance of experiencing a recurrence within 10 years of their first cancer diagnosis due to their mutation(s) (MSKCC, 2019; Scalia-Wilbur, Colins, Penson, & Dizon, 2016). These rates create uncertainty for those living with these mutations, which may ultimately lead to adverse mental health effects (Graves et al., 2012).

Treatments for *BRCA1/2*-related breast and ovarian cancers are similar to that of the general population but maintain a higher risk of mortality due to higher recurrence rates (MSKCC, 2019; Scalia-Wilbur, Colins, Penson, & Dizon, 2016). Treatment is highly dependent on various aspects of the disease as it presents, including staging (I-IV), hormone receptor status (human epidermal growth factor receptor 2 [HER2], estrogen [ER], or progesterone [PR] negative or positive), patient age, prior health, menopausal status, treatment preferences, and genetic predispositions (*BRCA1/2*) (ACS, 2019; Walker, Jacobson, & Sobel, 2019). The most widely implemented treatments for breast cancer involve surgeries to remove affected tissue
(lumpectomy, mastectomy, etc.) in combination with radiation, chemotherapy, hormonal therapy, targeted therapy, and/or immunotherapy. These could also occur conjointly with chemoprevention once masses are removed to prevent future cancer regrowth. Treatment and awareness relating to *BRCA1/2* mutations directly are now becoming prominent due to increases in expanding genetic knowledge (GARD, 2018; Walker, Jacobson, & Sobel, 2019), media coverage, and access to direct-to-consumer (DTC) genetic testing. Breast and ovarian cancer treatments among this population are not without risks, including future adverse health conditions related to treatment such as lymphedema and osteopenia as well as associated psychosocial issues such as worsened health-related quality of life (HRQoL) (Hill et al., 2015). Whereas the impact of having a *BRCA1/2* mutation may be life-altering in many ways, it is important to realize that such awareness allows for action through additional family genetic counseling and support.
CHAPTER 2

REVIEW OF BRCA1/2 GENETIC COUNSELING LITERATURE

The history of genetic counseling for BRCA1/2 mutations in the United States has focused on individuals most at-risk of breast and/or ovarian cancer within a family unit (Hallowell et al., 2005). The medical genetics field emerged from the Human Genome Project in 2003, which identified genes across the human genome. Whereas the family unit is directly affected by these mutations, historically, genetic counseling systems have focused on individual family members, testing one member at a time (Hallowell et al., 2005).

The system has changed somewhat after 2010, when direct-to-consumer (DTC) genetic testing kits became readily accessible and affordable, producing a larger and quicker need for genetic tests and results. Prior to 2010, most of genetic counseling was done by licensed genetic counselors (Prucka, McIlvried, & Korf, 2008). In response to this increasing need, primary care physicians (PCPs), oncologists, and other providers have begun offering genetic testing through larger corporations such as Color Genes, Myriad, and GeneDx (Olaya et al., 2009; Prucka, McIlvried, & Korf, 2008).

Although genetic counseling is more available than ever through in-person visits, virtual medicine, and telemedicine, the current individualized approach lacks an understanding of the potential benefits of involving the biological family unit in the counseling approach. Therefore, very little has been published regarding BRCA1/2 genetic counseling regarding patient preferences in response to their own experiences. This chapter will review the BRCA1/2 genetic counseling literature, highlighting hereditary genetic counseling related concerns, psychosocial outcomes, and other challenges with genetic counseling.

Individualized v. Family-Based Genetic Counseling
Generally, genetic counseling occurs over two separate appointments. In the first appointment, a genogram (or family tree) is constructed which includes breast/ovarian risk, cancer(s) diagnosed, age at death, and age at diagnosis amongst biological relatives. Individual risk is then determined dependent on the information given considering insurance coverage and ACA requirements (Walcott & Dunn, 2015). If the patient is considered “at-risk” for a BRCA1/2 mutation, genetic counselors submit orders to genetic testing companies (GeneDx, Color Labs, etc.) to collect biospecimen samples for testing. Test results are sent to the genetic counselor, who schedules a second appointment to discuss results and referrals to appropriate health care providers, if the patient tests positive.

Individuals most “at-risk” are often those within a biological family unit who have either experienced breast/ovarian cancer below the age of 50 or has a first-degree family member who has had breast/ovarian cancer(s) at age 45 or younger (MSKCC, 2019; Scalia-Wilbur, Colins, Penson, & Dizon, 2016). The primary genetic counseling approach within the United States, individualized genetic counseling, solely tests the individual within the family unit identified to be at greatest risk for having a mutation. According to Lerman and colleagues (1995), patients undergoing individualized counseling may bring relatives to either the genetic counseling appointments, but it is important to note that these relatives are not being tested and are present for support alone. Within this approach, patients are expected to disseminate findings from results to the family unit (Lerman et al., 1995; Lerman et al., 1996).

An emerging genetic counseling approach, family-based genetic counseling, involves genetic counseling for all members deemed ‘at-risk’ by the genetic counselor for having a mutated gene to be tested at the same time (Druker et al., 2017). However, family-based genetic counseling remains rare, as genetic counseling approaches are dependent on individual genetic
counselors or practices, as well as what costs insurance will reimburse. Whereas past research has focused on attitudes and preferences regarding individualized genetic counseling approach (in-person, web-based, telephone, mail) and decision-making on when to be tested, these features have not focused within the family-based approach. Unlike individualized counseling, dissemination of family-based genetic counseling test results is completed and guided by a genetic counselor with the entire family unit together at one time (Druker et al., 2017). Examples of individualized and family-based genetic counseling approaches are given in Figure 1. It appears that family-based genetic counseling has been overlooked, despite the nature of these hereditary cancers (Samimi et al., 2017), and as a result, patients’ experiences and preferences for individual versus family-based hereditary cancer counseling are absent from the literature.

**Hereditary Genetic Counseling Related Concerns**

Although current genetic counseling approaches assist in making informed health-related decisions (WHO, 2018), but only focus on the risk of one individual out of the family unit. When someone tests positive for *BRCA1/2*, not only is that person affected, but all immediate biological family members as well (Daly et al., 2015; Hodgson et al., 2016). Involving family members in genetic counseling may introduce concerns regarding privacy/confidentiality (Miller & Tucker, 2017), the perceived utility of testing (Siegrist, 2002), familial persuasion, differences of opinion, and/or disclosure (Gilbar et al., 2016). Generally, individualized genetic counseling overlooks the inclusion of family members, which may negatively impact familial relations (Godino et al., 2016), stress (Gaff et al., 2005; Hamilton et al., 2009), and cohesion (Daly et al., 2016). Patient preferences have not been studied in the current genetic counseling approach (Tan et al., 2016), making it difficult to determine the best path to such testing.
Past research suggests that disease-related worry, in addition to anxiety, stress, and depressive symptomology, increase after genetic counseling (Oberguggenberger et al., 2016). It is unknown, however, whether these outcomes are in reaction to test results themselves, the counseling experience, or both. Individualized genetic counseling leaves patients with the burden of disseminating test results and educating family members about risk-reduction strategies alone, increasing anxiety and informational inaccuracies (Daly et al., 2016; Samuel et al., 2017). Among younger patients (<35 years of age), discussing prophylactic decisions and pressures from family conflict may also lead to increased stress and anxiety (Brunstrom, Murray, & McAllister, 2016). Many patients report frustration with the burdening task and moral hardship of disseminating test results to family members (Armstrong et al., 2015; Daly et al., 2016; Hallowell et al., 2005). Many women who were *BRCA1/2*-positive noted complications within individualized genetic counseling directly related to disseminating information to family members who were now considered at-risk (Claes et al., 2002; Wiseman, Dancyger, & Michie, 2010). Some studies have found lower patient satisfaction within current genetic counseling practices (Athens et al., 2017; Buchanan et al., 2015; Voils et al., 2018), with one study suggesting as much as 18% noting the experience as displeasing (Hesse-Biber & An, 2016). Thus, there remains an unmet need to better understand and support individuals and families who are at-risk for these mutations both during and after the genetic counseling experience (Forrest et al., 2008).

Very few studies have examined the newer genetic counseling approach, family-based counseling, within the United States, and even less have focused on *BRCA1/2* mutations. Most studies have focused on neonatal and prenatal genetic mutations that identify fetuses or infants at risk for genetic diseases. This lack of attention may be partly due to the absence of thorough
healthcare insurance coverage for many approaches of hereditary genetic counseling, the possibility of familial conflict over the nature of testing and results, and other unique health equity factors, such as lower socioeconomic status [SES], access to counseling, and even out-of-pocket cost for subsequent prophylactic measures. The potential for inaccurate (or any) dissemination of test results to family members provides a strong rationale for a change in the current genetic counseling approach. For instance, Mendes and colleagues (2015) posit that the most appropriate way to conduct genetic counseling for hereditary conditions (such as BRCA1/2 mutations) is through a family-centered approach. Other research has incorporated family-based genetic counseling in families that may have a hereditary condition, highlighting familial involvement and the importance of follow-up family therapy regardless of testing outcome (George, Kovak, & Cox, 2015). Thus, family-based genetic counseling is gaining attention due to the basic nature of BRCA1/2 mutations and related adverse psychosocial outcomes that follow individualized genetic counseling (Cicero et al., 2017; Gonzalez-Ramirez et al., 2017; Oberguggenberger et al., 2016). Although research has begun to examine family-based genetic counseling for other genetic conditions, it has not been widely used in BRCA1 and/or BRCA2 genetic counseling approaches.

Not surprisingly, past research has yet to examine and compare the experiences, preferences, and psychosocial health outcomes in individuals receiving individualized versus family-based genetic counseling approaches. Similarly, increased stress (Gonzalez-Ramirez et al., 2017), anxiety (Di Lascio et al., 2017), and familial conflict (Dancyger et al., 2011; Fehniger et al., 2013) may compromise familial relationships. Although physical risks of genetic counseling remain low, there are known psychosocial risks both during and after the testing experience that remain long after genetic counseling has been completed.
**Genetic Counseling and Psychosocial Outcomes**

**Stress, anxiety, and perceived risk/health.** Throughout the individualized genetic counseling approach, anxiety and stress arise from different sources dependent on personal and situational factors. Varying from person-to-person, perceived cancer risk (Taber et al., 2015), familial conflict, comorbid conditions (Daly et al., 2016), and personal control can be overwhelming (Robinson et al., 2015). Within the individualized genetic counseling approach, stress and anxiety appear most often, and have the ability to worsen comorbid conditions such as heart disease, lymphedema, and hypertension (Madlensky et al., 2017; Nordin et al., 2011; Schwartz et al., 2014). In few cases, personal risk assessments occur prior to genetic counseling, most often with a trusted primary care physician (PCP). These assessments allow PCPs and genetic counselors to gauge patients’ perceived risk of a positive test result so steps can be taken to lessen their stress and burden. In a study conducted by Moyer (2014), a pre-test baseline measure of anxiety and stress was given before and after individualized genetic counseling to determine the utility of such a perceived risk screener. Undergoing genetic counseling is stressful because there is a possibility of receiving life-changing information (Nordin et al., 2011). Receiving such information in isolation, however, had the propensity to result in increased stress and anxiety, which is further worsened by the burden of disseminating such information to family members.

Overall, individuals who test positive for BRCA1/2 mutations experience increased stress, anxiety, fear, and uncertainty due to counseling experiences and potential consequences considering test results (Evans et al., 2001; Wenzel et al., 2012). Likewise, perceived overall health may also be adversely affected by the psychosocial outcomes described above, which in turn may cause stress and anxiety, therefore continuing this cycle. Moreover, patients are
expected to disseminate complicated test results to other at-risk family members, often neglecting the fact that patients may not be equipped to accurately answer questions that their families may pose (Healey et al., 2017). Even more intensive are prophylactic decisions for cancer risk reduction. Extraneous consequences relating to worrying about reproductive choices, insurance coverage, surgery recovery, and future genetic discrimination (ethical or otherwise) are often noted in the literature (Hall & Rich, 2000; Hoffman-Andrews, 2017; Klitzman, 2010). Therefore, stress and anxiety may worsen the overall health, both physical and psychological, not only during genetic counseling, but afterward as well (Madlensky et al., 2017).

**Health-related quality of life (HRQoL).** Although genetic counseling for *BRCA1/2* mutations allows opportunities for risk management, it has been shown to decrease general quality of life (QOL) (Harmsen et al., 2015; Tung & Garber, 2018). However, past literature has differentiated HRQoL among *BRCA1/2*-positive individuals by risk reduction strategies, such as surgery and chemoprevention (Friedlander et al., 2017). Unfortunately, little has aimed at understanding the subsequent risks that individualized genetic counseling has on HRQoL. Most research within this field has not focused on approach of genetic counseling, but on prophylactic measures relating to HRQoL. Prospective research has found reductions in HRQoL among *BRCA1/2*-positive women who have undergone risk-reduction mastectomies, finding that HRQoL worsened after surgery, but slowly improved with recovery (Metcalf et al., 2015). Recently, breast/ovarian cancer chemoprevention regimens have gained popularity, finding that among *BRCA1/2*-positive women, HRQoL has been shown to improve steadily, as those on these regimens perceive themselves as having “cancer protection” (Friedlander et al., 2018). Overall, it seems that among this population, little is known about HRQoL especially during genetic counseling and result dissemination periods.
Other Challenges with Genetic Counseling

Disclosing genetic test results has been reported to be stressful, difficult, and the main cause of familial disruption among those undergoing genetic counseling for BRCA1/2 mutations (Gilbar et al., 2016). Conflict amongst family members occurs for various reasons, all of which depend on individual perception, reaction, and preference for how test results are to be acted upon. Pressure from family members to make concrete prophylactic health decisions following a positive test result may increase familial disruption (Godino et al., 2016). Although family therapy may be suggested for highly disrupted family units, it is not required, and often left to genetic counselor discretion, leaving family members to find resources on their own. Additionally, some family members may not wish to know their test results (Gallo et al., 2009), which can cause issues when such information is discussed. Contention may also arise when familial communication is lacking, especially when a positive result demands time, effort, and commitment from the family unit (Gallo et al., 2009). Combined with an individual’s perception of cancer risk and test results, family members may disagree over the course of action regarding prophylactic measures. Although meeting with a genetic counselor, patients’ discussions of family history, symptomology, and likelihood of a positive result are brief and often overlook these potential challenges. A family-based approach to genetic counseling would involve the entire family unit, not just the individual, and therefore, the family becomes one cohesive patient group. Unfortunately, current standards do not include the option of family-based genetic counseling. By treating the entire family unit as one “patient” group, adverse psychosocial outcomes associated with dissemination of results would not fall on one family member. Family conflict, issues with communication and emotion, and isolation often occur with individualized
counseling but may be attenuated by implementing a choice for family-based genetic counseling (Galvin et al., 2015).

Ultimately, it remains unclear if one approach of genetic counseling (individualized versus family-based) may influence different psychosocial outcomes, including stress, anxiety, worry, and HRQoL in women testing positive for BRCA1/2 mutations (Cicero et al., 2017; Di Mattei et al., 2015; Rosenberg et al., 2016). Individualized genetic counseling comprises most of the genetic counseling in the United States today. Although research on genetic counseling for BRCA1/2 mutations and its psychosocial consequences continues to emerge, there remains unanswered questions about basic counseling approaches and pertinent patient preferences, as well as the potential for unintended consequences within those completing individualized genetic counseling (Cicero et al., 2017; Di Mattei et al., 2015; Graves et al., 2012; Rosenberg et al., 2016; Wevers et al., 2016).

Research Questions

The research questions driving the first objective were:

**RQ1.** What is the qualitative experience of women who have completed hereditary genetic counseling for BRCA1/2 (e.g., view of counseling, unique personal experiences)?

**RQ2.** What are women’s preferences regarding counseling approach (e.g., individual or family-based counseling) amongst women who have tested positive for BRCA1/2 genetic mutations?

The research question and two hypotheses of the second objective were:

**RQ3.** Do women who test positive for BRCA1 and/or BRCA2 genetic mutations differ from the general population on psychosocial outcomes such as anxiety, stress, and HRQoL?
**Hypothesis 3a.** Women who have received a positive hereditary breast cancer genetic result will have higher stress and anxiety than in the general population.

**Hypothesis 3b.** Women who have received a positive hereditary breast cancer genetic result will result in worse HRQoL and overall perceived health than those in the general population.
CHAPTER 3
RESEARCH DESIGN AND METHODS

The current project is a mixed-methods, cross sectional study, consisting of two primary objectives. The first objective was a qualitative exploration of women who have tested positive for \textit{BRCA1} and/or \textit{BRCA2} genetic mutations and their overall experiences surrounding genetic counseling, including their preferences for individualized versus family-based genetic counseling (see Figure 1). The second objective was an examination of psychosocial outcomes, including stress, anxiety, and health-related quality of life (HRQoL) among the current sample compared to the United States general female population.

Data collection was carried out in two steps. The first step asked participants to complete an online screener to determine eligibility and a subsequent survey for those eligible. The second step was an invitation to participate in a qualitative interview for a subset of survey participants.

\textbf{Participant Recruitment}

Participants were identified through national, online support groups: \textit{BRCA1} or \textit{BRCA2} Genetic Ovarian and Breast Cancer Gene group on Facebook (~6,800 members), Facing Our Risk of Cancer Empowered (FORCE; ~3,300 members; see Appendix A), \textit{BRCA} Genetic Sisters group on Facebook (~2,700 members), and \textit{BRCA} Strong group on Facebook (~1,300 members) (see Figure 2). One study recruitment post was posted per day on each support group website (\textit{BRCA} Strong Facebook group only allowed one post per week), with written permission obtained from the groups’ moderators. The post consisted of a brief announcement introducing the study, compensation for participation, and eligibility criteria as well as a link to an anonymous screener survey (see Table 1).
Before beginning the survey, participants were provided with a study information sheet, which they were asked to read and click “Next” at the bottom of the webpage. Due to the online nature of this survey, doing so was considered consenting. For compensation purposes, email addresses were collected at the end of the online survey. After completion of the online survey, one $20 Amazon e-gift card was emailed to each participant. Participants were then invited to schedule an interview (phone or webchat) with the study coordinator through the Facebook study webpage (see Appendix B), which had a scheduling option, where participants chose the time and date for their interview. The study coordinator then followed up with participant with desired mode of interview (phone or webchat), time, and date. Participants who completed the interview were compensated a second $20 Amazon e-gift card.

**Inclusion and Exclusion Criterion**

Participants were eligible if they were 18 years or older, female, and have tested positive for either (or both) *BRCA1* and/or *BRCA2* genetic mutations within the last five years. There was no maximum age set for the current study. The recruited sample focused on the adult cohort (18+ years old) due to current legislation banning children and adolescents from being genetically tested for these mutations, except if the child or adolescent has had cancer themselves (Saletta, Pozza, & Byrne, 2015). Completing testing within the past five years was implemented to ensure adequate sample size, as testing positive for *BRCA1* and/or *BRCA2* mutations is rare within the general population.

Participants were ineligible if they could not speak and/or read English, were male, were currently undergoing hereditary cancer counseling for *BRCA1* and/or *BRCA2* genetic mutations, and/or did not reside within the United States (obtained by Qualtrics© geolocation). Potential participants that were currently undergoing genetic counseling for the *BRCA1* and/or *BRCA2*
genes were not allowed to participate, because they did not receive their results. Although males remain at risk for inheriting *BRCA1* and/or *BRCA2* genetic mutations and experience breast cancer, it is rare, and comprise fewer than 1% of all breast cancer diagnoses (Mano et al., 2017).

One hundred and twenty-three potential participants responded to the 4-item online screener (see Table 1). Of the 123, 63 persons were eligible (51.2%). The most common reasons for ineligibility were not having completed *BRCA1/2* genetic counseling in full (*n*=29; 48.3%), not undergoing formal genetic counseling (*n*=11; 18.3%) and testing negative for *BRCA1* or *BRCA2* (*n*=10; 16.7%).

**Online Survey**

**Participant information.** Self-reported demographic information was collected from participants regarding age, relationship status, race, ethnicity, education level, employment, and other demographic areas of interest. Health insurance information and the extent genetic counseling was covered was also collected. Psychosocial variables were collected via Qualtrics© survey (see Appendix D). Most completed the survey in about 20-30 minutes (*M*=14.3; Range=5.95-45.7). Email addresses, IP addresses, geolocation, and names were deleted after completion of survey and subsequent compensation to retain confidentiality.

**Clinical cancer information.** The online survey also captured clinical cancer information including previous cancer diagnoses, type of cancer, treatment completion (if applicable), and recurrences (if any).

**Genetic counseling information.** Information concerning genetic counseling was also collected via online survey. Specifically, when counseling began and was completed, state that they received counseling, type of genetic mutations with positive result (*BRCA1*, *BRCA2*, both, or another mutation tested for in conjunction with a *BRCA1/2* mutation), location of counseling
(e.g., private, hospital, doctor’s office), if post-counseling therapy was offered, and whether family-based or individualized genetic counseling was completed.

**Psychosocial Outcomes Measures**

**Perceived stress.** The Perceived Stress Scale-10 (PSS-10; Cohen et al., 1983) was used to measure the perceived stress level of the participants at the time of genetic counseling. The PSS-10 was composed of 10 items on a 5-point Likert scale, ranging from 0 (never) to 4 (very often) (see Appendix D). Examples of items included, “In the last month, how often have you felt nervous and ‘stressed’?” and “How often have you felt that things were going your way?” Scores for four items (4, 5, 7, and 8) were reversed, and all items added for a total score ranging from 0 (lower stress) to 40 (higher stress) (Cohen et al., 1983). The PSS-10 has been used in breast cancer populations, showing that these groups had higher levels of stress when compared to the general population, remaining especially true if they were at increased risk for breast cancer (McDonough, Sabiston, & Wrosch, 2013) and/or were currently undergoing treatment (Golden-Kreutz, Browne, Frierson, & Andersen, 2004). According to Cohen and colleagues (1983), the PSS-10 had an acceptable reliability score of 0.78, and in the current sample it was 0.89, indicating good internal consistency.

**State and trait anxiety.** The State-Trait Anxiety for Adults Scale (STAI; Spielberger et al., 1983) was used to measure the state and trait anxiety levels of participants recalled from the time of genetic counseling. The STAI was comprised of 40 questions on a 4-point Likert scale, ranging from 1 (almost never) to 4 (almost always) and from 1 (not at all) to 4 (very much). Examples of these items included “I feel satisfied”, “I feel nervous”, and “I feel nervous and restless”. State and trait subscales were each comprised of 20 questions, all weighted from 1 to 4, with 4 indicating high levels of anxiety for the specific item. Scores were reversed for items 1, 2,
State subscale scores were added for items 1 to 20, and trait subscale scores from items 21 to 40, with each subscale score ranging from 20 (lower anxiety) to 80 (higher anxiety). This measure was standardized, with test-retest average reliability of 0.86, and a Cronbach’s alpha statistic of 0.86, indicating good reliability (Julian, 2011). Within the current sample, the reliability coefficient was shown to be excellent for both state ($\alpha=0.95$) and trait anxiety ($\alpha=0.91$). The STAI had been used widely in breast cancer patients and survivors, with mean scores being higher among these groups (Maass et al., 2015).

**Health-related quality of life and perceived overall health.** Health-related quality of life (HRQoL) was measured using the Medical Outcomes Scale Short Form-36 (SF-36). The SF-36 is a 36-item Likert scale survey used in groups with chronic conditions, such as breast cancer or a predisposition to cancer (Ware & Sherbourne, 1992). The SF-36 measures eight HRQoL domains, including physical functioning, bodily pain, role limitations due to physical health problems, role limitations due to personal or emotional problems, emotional well-being, social functioning, energy, and general health perceptions (Ware & Sherbourne, 1992). Each item was calculated from zero to 100 range, with lowest scores indicating poorest health. Coding scheme varied per subscale. A higher subscale score indicated higher functioning or better HRQoL.

Physical functioning, role-physical, and role-emotional item responses remained the same. Bodily pain item C22 was recoded (1=6, 1[if item C21 is 2-6] =5, 2=4, 3=3, 4=2, 5=1) and item C21 remained the same. General health items were recoded as follows: item C1 (1=5, 2=4.4, 3=3.4, 4=2, 5=1) and items C34 and C36 (1=5, 2=4, 3=3, 4=2, 5=1). Items C33 and C35 maintained their original coding scheme. Energy items C23 and C27 were recoded (1=6, 2=5, 3=4, 4=3, 5=2, 6=1) and items C29 and C31 stayed the same. Social functioning item C20 was
recoded (1=5, 2=4, 3=3, 4=2, 5=1) and item C32 remained the same. Emotional wellbeing items 9d and 9h were recoded (1=6, 2=5, 3=4, 4=3, 5=2, 6=1) but coding for items 9b, 9c, and 9f remained the same. Once items within the 8 subscales were recoded or retained (dependent on item), subscale scores were weighted (using general population normative scores) and summed to a zero to 100 scale using Ware and colleagues (1993) syntax. Two additional subscales (physical component score [PCS] and mental component score [MCS]) were also calculated by summing the following subscale scores: PCS (bodily pain + physical functioning + role limitations due to physical health issues + energy scores) and MCS (emotional wellbeing + social functioning + role limitations due to personal/emotional issues) (Ware et al., 1993). Both SF-36 component scores have standardized means of 50 and standard deviations of 10 (Ware, Snow, Kosinski, & Gandek, 1993). According to Stewart and colleagues (1992), all SF-36 subscales had good to excellent reliability, with Cronbach’s alpha statistics ranging from 0.78 to 0.93. In the current sample, SF-36 subscales’ reliability coefficients were as follows: physical functioning (excellent; \( \alpha =0.92 \)), role limitations due to physical health (excellent; \( \alpha =0.91 \)), role limitations due to emotional problems (good; \( \alpha =0.84 \)), energy (good; \( \alpha =0.89 \)), emotional well-being (good; \( \alpha =0.81 \)), social functioning (good; \( \alpha =0.89 \)), pain (good; \( \alpha =0.84 \)), general health (acceptable; \( \alpha =0.77 \)), PCS (acceptable; \( \alpha =0.76 \)) and MCS (acceptable; \( \alpha =0.78 \)). Furthermore, the SF-36 had primarily been used in populations with chronic conditions, including those predisposed to, under treatment for, and survivors of breast/ovarian cancers (Razdan, Patel, Jewell, & McCarthy, 2016; Treanor & Donnelly, 2015).

Similarly, perceived overall health was measured using a subscale of the SF-36 measure called “General Health”, comprised of five items (see above) relating to perceived current health, likelihood of falling ill, and comparing one’s health to others (Ware & Sherbourne, 1992).
According to Ware and Sherbourne (1992), the General Health subscale of the SF-36 has shown good reliability, with a Cronbach’s alpha statistic of 0.78, but in the current sample it has shown acceptable reliability ($\alpha=0.77$).

**Perceived breast/ovarian cancer risk.** Perceived breast/ovarian cancer risk was examined by asking participants several questions based on previous research assessing individual risk. Four questions including the belief about current tumor growth (de Castro e Silva et al., 2013), belief of risk of developing an ovarian tumor (de Castro e Silva et al., 2013), belief of risk compared to others (Skinner et al., 1998), and breast/ovarian cancer risk as compared to family history (Audrain et al., 1995) were presented. All of the above questions had responses ranging from zero (low perceived risk) to 10 (high perceived risk) on four separate breast cancer-related items. Item scores were interpreted separately, with higher scores indicating greater perceived breast/ovarian cancer risk and vice versa in response to prompt, which ranged from 0 (low perceived risk) to 10 (high perceived risk).

**Data management.** Data management was an ongoing process from the beginning of recruitment until the study was completed. Data management was conducted in conjunction with cleaning both during recruitment and afterward. At each ten-participant interval, Qualtrics© data were downloaded into SPSS 25© to be screened, helping to ensure reliability and possibly foresee potential issues in recruitment. The cleaning of data post-recruitment included recoding (i.e., reverse, weighting, dummy-coding) and scoring of outcome scales.

**Analytic Plan**

**Preliminary descriptive and correlational statistics.** Sample means and corresponding standard deviations were calculated for continuous variables, in addition to frequencies and percentages regarding for categorical variables. The sample’s distribution, as well as evidence of
kurtosis and skewness regarding outcome measures, were assessed via SPSS©. Normality relating to skewness and kurtosis was as follows: skewness statistics less than -1.0 or greater than +1.0 would be considered highly skewed, and appropriate variable transformations would be conducted to normalize the variable. Kurtosis statistics less than -2.0 (platykurtic) or greater than +2.0 (leptokurtic) would be transformed to mirror a normal distribution through standardization. Overall, the current study had an ongoing approach to potential missing data, as a data management and cleaning plan was constructed prior to beginning recruitment. Cases would have been eliminated if missing more than 20% of quantitative data, as suggested by Dong and Peng (2013). Using basic frequency statistics, item-level outcome missingness was identified, ranging from 0.0% to 6.7% missing. Therefore, out of the 60 participants in the total sample, none were missing over their data, so no cases were eliminated from analysis.

**Covariates and variable structure.** Exploratory analyses identifying possible confounding variables were planned in addition to the research questions. These were identified using a method by Henderson and Velleman (1981), where covariates were chosen based upon a combination of statistical analyses, past literature, and applicability to the current analyses. Using this method, a number of variables were tested to identify possible covariates and underlying relationships, including but not limited to demographic (e.g., age, education, ethnicity, income, state of residence, insurance), clinical variables (e.g., cancer occurrence/recurrence, treatments), and genetic variables (e.g., years since genetic counseling, avenue of genetic counseling, follow-up services offered).

Several demographic and genetic counseling variables were dichotomized for further analyses and to ensure variability for analyses. Education level was dichotomized from a five-level variable (0=less than high school, 1=high school graduate or GED, 2=some college or
technical vocation, 3=college graduate, 4=some graduate school, 5=graduate degree) to a
dichotomous variable (0=less than undergraduate degree, 1=undergraduate degree or above).
Similarly, marital status was originally a four-level variable (0=married or living as married,
1=divorced, 2=separated, 3=widowed, 4=single and never married) to a dichotomous variable
(0=not married, 1=married or living as married). Employment status was also a multi-level
variable (0=working full-time, 1=working part-time, 2=full-time homemaker or family caregiver,
3=retired, 4=student, 5=unemployed, 6=other) to a dichotomous variable (0=not working,
1=working). The variable relating to genetic counseling facility (0=private genetic counseling
office, 1=hospital, 2=PCP, 3=DTC) was dummy coded for further analyses.

**Psychosocial health and wellbeing of the current sample.** Secondary objectives
examined the differences between psychosocial outcomes (e.g., stress, anxiety, HRQoL, overall
health, and perceived breast/ovarian cancer risk) in the current sample and the general female
United States (US) population. This research question was addressed by first compiling general
female population data from previous research that had used the same standardized measures
(PSS [Cohen et al., 1983], STAI [Spielberger, 1983], SF-36 domains, the General Health
subscale, and PCS and MCS components [Ware & Sherbourne, 1993], to create a baseline
comparison group to the current sample. Equivalent general female population subscale score
means and standard deviation statistics for the outcomes listed above were entered into SPSS
25©. The data were analyzed through group mean scores on the PSS, STAI, and SF-36 subscales
(and associated General Health subscale), comparing current sample scores to the previously
published general female population normalized scores on the same measures. Using
independent sample t-test analyses and frequencies, group differences were identified on the
PSS, STAI, and SF-36 subscales. Perceived breast/ovarian cancer risk was unable to be
compared to general female population scores because there was no standardized measure that mirrored the compilation of questions informed by past literature (noted above). Additionally, to our knowledge, perceived breast/ovarian cancer risk (specifically from genetics or personal/family history) is not normally collected upon a population-wide basis. Therefore, this score was analyzed considering the current sample only.

**Semi-Structured Interviews**

Before collecting data, a process was defined to ensure quality and rigor. This process highlighted 1) what specific approach will be employed to guide research (e.g., thematic analysis); 2) the type of method used to collect data (e.g., interviews), and 3) how data will be recorded and transcribed to aid the thematic coding of data (e.g., telephone, web-based video chat) (Berger, 2013). The current study’s qualitative portion was comprised of three stages: 1) planning the interview process, 2) data collection and transcription, and 3) data analysis and coding.

**Interview procedure and protocol.** A total of 60 participants completed the online survey (described above), and at the end of the survey, were asked whether they would like to take part in a follow-up interview concerning their experiences in genetic counseling, the primary research question of the current study. A total of 35 (58.3%) participants agreed to having a follow-up interview and scheduled a time to talk (or virtually “meet”) during the following week.

A semi-structured interview protocol was created (see Appendix E), modeled after a research protocol used by Augestad and colleagues (2017). Augestad and colleagues (2017) focused their interview questions on what healthy persons without cancer or cancer-related mutations believe they can do to prevent or screen for different types of cancer. Using their script
protocol as a model (Augestad et al., 2017), the current study focused on women’s attitudes and preferences: timing of genetic information and testing, experiences about deciding whether to be tested, mode of delivery (in-person, web-based, telephone, mail), positive and/or negative aspects about the mode of delivery, test result format (in-person, web-based, telephone, mail, etc.), situation when receiving positive test results, emotional response to test results, and/or long-term reaction of test results (see Appendix E).

Before beginning the interview, verbal consent was obtained in two parts: 1) interview consent, and 2) permission to record the interview. Recorded interviews were copied and saved in two different places in case file corruption occurred. All interview questions were open-ended and novel, indicating that no preconceived meaning or patterns were expected from participant interviews (Münster, 2013). The interviewer used memo-writing (Creswell & Poth, 2018) to review different aspects of previous interviews to prepare for future interviews. Building rapport was also another practice that was used to establish trust within the interviewer/participant relationship that is necessary for optimal comfort as well as the ability to share personality. The interviewer established rapport by exercising her ability to listen and synthesize research agenda simultaneously, being able to ask comprehensive follow-up questions, being conscientious, and letting the participant guide the interview at their own pace (Seidman, 2019). The interview process was semi-structured, but flexible, depending on what information participants introduced and where the discussion led.

Out of the 35 participants who agreed to partake in a follow-up interview, 34 (97.1%) completed a full interview. One interview was stopped prematurely because the participant was disconnected abruptly, despite the interviewer calling back. This interview was 2.35 minutes long and used only yes/no responses, so this interview was not included in analyses. Past
literature identifies (Faulkner & Trotter, 2017; Guest, Bunce, & Johnson, 2006; Morgan et al., 2000) data saturation to guide the number of participant interviews collected with the ceiling usually between 20 and 40 interviews. Saturation is further identified by a redundancy inferring that no new themes would emerge. The total subsample to qualitatively analyze was 34 interviews, fitting between the previously suggested interview collection range discussed above.

**Interview transcription.** The transcription team was comprised of four research assistants (RAs). To ensure accuracy, the principal investigator (PI) organized the four RAs into two teams (two RAs on each team). The PI then split the 34 recorded interview audio files in half, so that each RA team was assigned 17 interviews to transcribe fully and verbatim. This acted as a failsafe to catch possible mistakes by both individuals and/or transcribing teams. The PI then compared each of the interview transcriptions for accuracy and clarity against the mirroring transcription. The PI then met with the transcription team to discuss results of the crosscheck and to discuss transcription concerns and nuances. These meetings were conducted at fixed intervals throughout the transcription process. After accurate transcriptions were confirmed, physical copies were printed and given to the coding team to begin thematic analysis.

**Coding team and data analysis plan.** The coding team consisted of two expert coders (Coder 1, Coder 2) who independently identified themes within each interview question. Physical copies, provided by the RA transcription team, were given to Coders 1-2 so that they could read and analyze and identify potential themes using thematic analysis. Thematic analysis generally identifies themes by topics, ideas, and patterns that repeat across participant interviews (Christians & Carey, 1989). Prior to thematic analysis, the method of “pawing” (otherwise known as “eyeballing”) was implemented after reading each interview transcription for the first time. “Pawing” involves reading through interview transcriptions, marking up with different
highlighter pens to observe material and key phrases that are of interest and make sense for the overall objective or research question (Bernard, 2000). By rereading each interview transcription multiple times on multiple occasions, both coders were able to get a feel for the data (Bogdan & Biklen, 1992).

Coders 1 and 2 met on four different occasions. Initially, four interviews were coded together to gain a coding cadence and to start a preliminary codebook. Between meetings, both coders separately “pawed” through a subset of interview transcriptions when initial key phrases and quotes were identified. Each time they met, key phrases and themes were discussed compared to previously recorded quotes and in relation to possible themes. On the third meeting, coders reached data saturation at the 28th interview, but decided to code the remaining six interviews because there were so few left to add richness through key phrasing. On the final meeting, main themes and subthemes were confirmed.

Sample saturation. A total of 34 interviews were analyzed using thematic analysis. Upon meeting several times, Coders 1 and 2 noticed that no new themes emerged after 28 interviews, but there were only six interview transcriptions remaining, both coders analyzed them as well. Although 28 interviews were the point where no new themes were exemplified in interviews and was considered “saturated” (Morgan et al., 2000), this total did not identify a redundancy in information (Faulkner & Trotter, 2017; Guest, Bunce, & Johnson, 2006). Therefore, the remaining six transcribed interviews were analyzed to collect any key phrases or quotes that added depth and richness to the analyses. As mentioned previously, data saturation most commonly occurs between 20 and 40 participants (Kerr, Nixon, & Wild, 2010), which supports the decision to analyze all 34 transcribed interviews, allowing for the flexibility and impact of analyzed themes and subthemes.
**Coder inter-rater reliability.** Thematic analyses regarding interview transcription was completed in the following steps: 1) independently identified themes within each question; 2) if the majority of transcriptions have an apparent theme (>50%) (coders had 97.1% inter-rater reliability), that theme was included as a main theme; and 3) Coders 1-2 compared themes for each question. Subthemes of main themes were also noted, but did not follow any previously set majority percentage, but were based on how often they appeared, how pervasive it is across different experiences, and how impactful or forceful it appeared (Ryan & Bernard, 2003).

Qualitative coder inter-rater reliability (IRR) was also calculated by using the following formula (Marques & McCall, 2005):

$$\text{IRR} = \frac{(\text{Total # agreements})}{(\text{Total # observations})} \times 100$$

If the IRR result fell below 85%, the Coders 1-2 discussed and identified discrepancies between themes and identified an agreeable coding resolution to be implemented for all future coding.

Overall, there was a 97.1% agreement between Coder 1 and Coder 2.

Interview data was entered into SPSS© from Interview Question 8b: “Based on your experiences, which form of genetic counseling would you have preferred?” (see Appendix A) to answer the second research question. Using SPSS©, a categorical variable for counseling preference was created (0=individualized, 1=family-based). Using descriptive statistics, frequencies were analyzed based on what percentage of the sample reported preference for either individualized or family-based genetic counseling.

**Validity.** The concept of “reflexivity” within qualitative research can be defined as the understanding of a concurrent relationship between the group being interviewed and the experience of the researcher (Gilgun, 2008; Probst, 2015). Such a recursive relationship infers that researchers’ subjective responses may enact trust, rapport, and preconceptions from the
participant, promoting truer answers and the continuance of discussion (Probst, 2015). Therefore, reflexivity is an essential part of the entire research design, from planning to writing, not only for the quality of information shared, but the meaning shared as well (Palaganas et al., 2017). Thus, reflexivity was incorporated within every aspect of the current research design. For instance, after every interview was completed, I took the time to reflect on what was discussed and what topics were introduced within the interview to prepare for, and enrich, subsequent interviews. I took notes during each interview on topics that could inform future interviews. Using my own past experiences regarding breast cancer and BRCA1/2 genetic counseling, I used reflexivity to understand how participants may feel from diagnosis, treatment, or test results to better conceptualize their experiences.

Navigating “insider” vs. “outsider” status. Within qualitative research, there exists terminology called “insider” and “outsider” status. Although it has been discussed widely within ethnography, observation, and field research, it has been increasingly attributed to interviewers and interviewees, especially within minute cultures (Dwyer & Buckle, 2009). Breast/ovarian cancer survivors and “pre-vivors”, as those with BRCA1/2 mutations call themselves, belong to a culture of their own, based upon the increased risk for chronic disease(s) due to genetic predispositions. Within the current study, individuals who have had breast/ovarian cancers and/or have tested positive for BRCA1/2 mutations are part of the “insider” group. Individuals, such as researchers and clinicians for example, who have not experienced these life occurrences, belong to the “outsider” group (Tinker & Armstrong, 2008). Most qualitative research is conducted from the “outsider” perspective (Tinker & Armstrong, 2008), with the researcher on the outside looking in, attempting to learn more about a population.
Personally, I have undergone BRCA1/2 genetic counseling, but my results were negative. Therefore, I identify as being between an “insider” and “outsider” regarding the current sample. On one hand, I have had multiple family members diagnosed with breast and/or ovarian cancers, allowing me to understand and share their struggle. I also have experience in researching those with breast, ovarian, and other cancers. I have not, however, been diagnosed with cancer nor a genetic mutation relating to cancer, so I cannot identify as having pure “insider” status. I have not experienced what this sample has, but I am not ignorant of the breast/ovarian cancer and genetic counseling worlds, making me neither “insider” nor “outsider”, landing somewhere in between. I view these aspects of my own personal experiences and research as beneficial, especially when interviewing women of this population. During the interviews, the majority of the sample (n=16; 47.0%) seemed to know that I was somehow connected to the BRCA1/2-world, per say, asking me at interview end what experiences I had to lead me to research this topic specifically.
CHAPTER 4

RESULTS

Sample Characteristics

A total of 60 participants completed the online survey. The mean age of the sample was 43.3 years ($SD=10.9$, Range=24-71 years), with the majority completing an undergraduate degree or above ($n=43, 71.1\%$). Most were married or living as married ($n=44, 73.3\%$), employed ($n=43, 71.1\%$) and were primarily from the Northeast ($n=18, 30.0\%$) and Midwest ($n=18, 30.0\%$) regions of the United States. The mutation(s) in which participants tested positive were similar, as $45.0\%$ ($n=27$) had a $BRCA1$ mutation and $48.3\%$ ($n=29$) had a $BRCA2$ mutation. Most received genetic counseling from a hospital genetic counseling program ($n=24, 40.0\%$) or private genetic counseling office ($n=19, 31.7\%$). Only $16.7\%$ ($n=10$) of the current sample had experienced breast cancer and $6.7\%$ ($n=4$) experiencing ovarian cancer, and five ($8.3\%$) participants experiencing cancer recurrences. Additional demographic, clinical, and genetic counseling variables are displayed in Table 2. There was no variability in ethnicity or race, therefore these variables were not included in subsequent analyses. Genetic counseling approach also had little variability.

A subset of the total sample completed an interview ($n=34$). The demographic characteristics of the subsample were similar to the total sample. The mean age was $43.3$ years ($SD=9.94$; Range=28-40 years). The majority of interviewees were White ($n=32; 94.1\%$), non-Hispanic ($n=34; 100.0\%$), married (or living as married) ($n=25; 73.5\%$), worked full-time ($n=24; 70.6\%$), and earned an undergraduate or above ($n=29; 85.3\%$). Most lived within the Midwest ($n=11; 32.4\%$) and Northeast regions ($n=8; 23.5\%$) of the U.S. Clinically, most had no previous cancer diagnoses ($n=26; 76.5\%$) and only three participants ($8.8\%$) experienced a recurrence.
Overall, the average number of years since genetic counseling was 2.29 years ($SD=11.7$; Range=3 months - 4 years) and all had insurance at the time of testing. Most received genetic counseling at a hospital ($n=15; 44.1\%$) or through their primary care physician (PCP) ($n=9; 26.5\%$). Mutations tested positive for was split, with 50% ($n=17$) testing positive for $BRCA1$ and 50% ($n=17$) for $BRCA2$. The majority ($n=23; 67.6\%$) was not offered follow-up mental health counseling services after genetic results were given. All demographic, clinical, and genetic demographic characteristics group differences (total sample, interviewed sample) were compared using independent samples $t$-tests and chi-square analyses. Educational level (0=less than undergraduate degree; 1=undergraduate degree or above) was the only variable to differ between groups, $\chi^2(1, 60)=7.16, p=.007$, as those who completed the interview had higher educational attainment than those who did not. Detailed demographic information for the interviewed subsample can be found in Table 2.

**Research Question 1: Women’s Experiences of Genetic Counseling**

The primary objective of this research was to examine the lived experiences of women who have completed hereditary genetic counseling for $BRCA1/2$ mutations. Exploratory data was collected through 34 independent, semi-structured interviews with women who have completed genetic counseling and tested positive for $BRCA1/2$ mutations within the United States. Participants were asked questions related to their unique personal genetic counseling experiences, and where they viewed counseling fell short.

**Thematic patterns.** A total of six overarching themes were identified: 1) sources affecting perceived risk ($n=34$); 2) preventive concerns and decisions ($n=34$); 3) experiences in healthcare ($n=34$); 4) emotional reactions to genetic counseling ($n=33$); 5) future recommendations ($n=31$); and 6) family support and communication ($n=29$) (see Figure 3).
Overall, these themes were comprised of participant experiences, reactions, and views of the \textit{BRCA1/2} genetic counseling. The following section discusses each of these themes, in order from most- to least-prevalent. Subthemes will be discussed in order of overall impact within each theme’s section.

\textit{Theme 1: Sources affecting perceived risk.} The first theme, sources affecting perceived risk, was identified across all participant interviews. This theme encompassed how participants perceived their own risk (and family members’ risk) of being diagnosed with breast and/or ovarian cancer(s) due to their positive \textit{BRCA1} and/or \textit{BRCA2} genetic status. The full diagram is featured in Figure 3, but this section’s subthemes are depicted below.

Most women who were interviewed highlighted the importance of perceived risk following a positive \textit{BRCA1/2} test. Women perceive “every illness, pain, and symptom” as being associated with their risk of cancer. One woman stated that she feels as though she is “living life on the edge” because she is “hyperaware” of her risk. Women, on average, seemed to note increased perceived risk of breast/ovarian cancers in the future if they experienced multiple sources of risk. For instance, having many instances of ovarian cancer within the family was perceived as higher risk than women with less sources. Sources of perceived risk seemed to be mitigated by “active awareness” of cancer (e.g., biannual surveillance, self-breast exams, etc.). Therefore, women who felt “confident” in the risk assessment given to them by genetic
counselors or oncologists, coupled with their active awareness of cancer seemed to have a less strenuous perceived risk of cancer overall. Participants shared the following:

“I was diagnosed with breast cancer, my father had prostate melanoma cancers, and his mother had ovarian cancer. I started to wonder if maybe this was relevant to me and my cancer risk.” – Participant 16

“My mother had breast cancer twice; she’s a survivor. Her sister had it twice, my aunt, but the second time it killed her. Their mother passed away from ovarian cancer at an early age, and I was starting to think there’s something wrong with the women in my family.” – Participant 17

“I’ve opted for preventive maintenance like alternate between mammograms and MRIs every six or eight months in addition to tamoxifen to prevent against cancer, since I’ve had it in the past.” – Participant 30

The sources of perceived risk associated with having a BRCA1/2 genetic mutation, to self and family members were also highly prevalent. Overall, women reported perceiving their risk in a different and more meaningful way. Some women explained that living their life (post-test results) had caused them to “stress about cancer”, when they would not have before, because they were now “made aware of it”. Although the most common source affecting perceived risk was having multiple familial histories of cancer, especially breast and/or ovarian, it is also important to note that many participants reported more than one source affecting perceived risk within their interview, such as having a personal and familial history of breast cancer.

**Theme 2: Preventive concerns and decisions.** Women’s unique experiences within this theme portrayed how and why, considering a positive BRCA1/2 test result, they may have chosen specific surgeries, treatment(s), and/or medications, and their plans for undergoing such measures. The decision for opting in or out of surgery or medication therapies was discussed in response to emotion, potential side effects, views of disfigurement, and/or the cause of infertility. The impact of these decisions may be enough, in some, to outweigh choices that were medically advised, especially amongst younger women. Although these women may have chosen surgery,
surveillance, or a combination, it was clear that they knew that they would “have to undergo all risk-reduction surgeries eventually” to reduce their risks of breast and ovarian cancers. Participants explained feeling “overwhelmed” with the information given at the time of results, stating that such feelings impacted how they felt about prophylactic surgery ($n=29$), medication therapies ($n=3$), and/or biannual surveillance ($n=21$). Women stated that they felt “inundated” with all this information, not knowing what to do with it. Themes and subthemes are also depicted in Figure 3 and below.

**THEME 2: PREVENTIVE CONCERNS & DECISIONS $n=34$**
- Surveillance ($n=21$)
- Worry about side effects ($n=2$)
- Hormone replacement ($n=3$)
- Surgery ($n=29$)
- Oophorectomy ($n=17$)
- Double mastectomy ($n=23$)
- Salpingectomy ($n=10$)
- Hysterectomy ($n=13$)

Women had varying responses to the recommendation of immediate prophylactic surgery ($n=29$), the standard for women who are $BRCA1/2$-positive. Most medical professionals recommended consecutive bilateral double mastectomies and total hysterectomies, especially if the woman has already had children. Although undergoing all prophylactic surgeries may be the final “hurdle” to reduce once risk, women held varied reactions about them. Some women did not question the recommendation to elect surgery, but a subset of the sample had a more difficult time deciding, identifying a need for “reflection and time”. A minority of premenopausal women noted being approached for “freezing their eggs” so they could undergo surgery now. Women responses varied from positive to negative to these recommendations. Some felt as though it was “too much, too fast”, whereas others wanted to “take the bull by the horns” and “get it over with”. Women explained feeling “overwhelmed”, “disrespected”, and “dehumanized”, but at the
same time, felt “empowered” and “strong”, suggesting the co-existence of positive and negative reactions. Response to prophylactic surgery often was dependent on one’s previous history of cancer, previous familial histories of cancer, perceived cancer risk before genetic counseling, and whether the participants had children. These issues can be illustrated in the following quotes:

“I didn’t want to go into menopause that way...I wanted to go naturally. At first, I was afraid of the side effects of surgery, of a hysterectomy and so forth, then the whole thought of having a mastectomy, just felt like amputations.” – Participant 3 (underwent surgery but regretted it)

“I’ve had it in my head since day one that if I’m positive I’m going through all of the surgeries to give myself a fighting chance.” – Participant 39

“I do what I can to be proactive – I don’t want the same fate and I especially don’t want my children to have to see their mom go through cancer treatment if I have choices to prevent it.” – Participant 48

“I had the bilateral double mastectomy. The right breast was fine but in the left breast there was a tiny node of cancer. That scared me into getting the oophorectomy. I had that to buy me some time before having the hysterectomy in a couple of years. The door into that place of innocence is gone.” – Participant 52

“It’s taking the bull by the horns...if it were just me, I might have a totally different approach, but I’ve got a six-year-old daughter and eight-year-old son, and I need to be around to help if either of them have to go through something like this. There’s no question for me that getting [the surgery] done is the right thing. My initial response was that I didn’t even blink as soon as my doctor told me what the results were, I said, ‘gut me off and get them off.’” – Participant 57 (underwent all preventive surgeries back-to-back)

Some participants exhibited fearful responses (n=10) and worry about side effects of such major surgeries (n=2) but viewed them as positive because of their risk-reducing benefit and overall necessity long-term (n=19). Women who completed prophylactic surgeries did so to reduce their risk of having cancer, which in turn, increased their perceived “control over their mutation” and the way their bodies were “working against them”. Although all participants noted that prophylactic surgeries were a “means to an end”, some felt that they were being “forced into
something that they did not want to do”, feeling “shamed” or “pressured” by medical professionals. Others underwent surgeries back-to-back, identifying this experience as the “most important for their health as well as their families”. Participant opinion varied due to age, reproductive choices, and personal situation, exhibited with an array of emotions when it came to the different surgeries and choices available.

Another preventive concern noted by many of these women was the “struggle” of ongoing surveillance. These women approached surveillance and risk management in different ways: opting out of having surgery altogether ($n=4$) or continuing surveillance in conjunction with limited surgery ($n=17$). Active surveillance often included biannual mammograms, magnetic resonance imaging (MRI), gynecologic exams, and transvaginal ultrasounds. These surveillance methods often co-occurred with one another and additional tests for associated cancers such as those for the skin, pancreas, and colon. The impact of having to “juggle” surveillance appointments, oftentimes with several specialists, was almost “too much to handle”. Overall, women with $BRCA1/2$ believed they “do not truly have a choice” in long-term surveillance, even after they undergo risk-reducing surgeries, because it really is not “preventing anything”. Women reported feeling “like a deer in the headlights”, “helpless”, and “cornered” regarding the immediacy and necessity of prophylactic surgeries. Women expressed that although they may undergo biannual surveillance, cancer could occur between surveillance appointments, so they are “just doing it to catch it early”. The time that this takes, for instance, was noted to be a reason why women opted for “one-and-done” surgeries. There was also a small group of women who did not want to be “disfigured” or “defeminized” ($n=3$) by surgery. Again, most women who opted for long-term surveillance methods often did not have children yet (and
wanted them), were not finished having children yet, or did not find “comfort” in surgery. The struggles these women faced are exemplified below:

“I was recommended to have an oophorectomy and then a bilateral mastectomy two days later. I chose not to because I’m so young, but I am starting colon and pancreatic cancer screenings because those run in my family too. Endoscopic ultrasounds twice or once per year don’t sound fun.” – Participant 5

“I ended up doing a double mastectomy and removing my ovaries and tubes so now I don’t have the risk of ovarian cancer. But tomorrow, I’m actually starting monitoring for pancreatic cancer, so it really just never stops.” – Participant 16

“We do the screens [mammograms] every 6 months and the MRI as well. I wasn’t able to do the MRI in October because I was pregnant, but when they did the prenatal ultrasounds, I always asked if they could check my ovaries too, just for peace of mind. I called them my ‘pre-scans’.”. – Participant 49

“The breast doctor was like you can get the double mastectomy, reconstruction, and total hysterectomy within the next 6 months and then you’ll be set. I must’ve hesitated, because she told me, ‘well, you don’t have to decide today, I guess.’ I’m set on having surveillance every 6 months with a breast MRI and mammogram. It just felt like almost being pushed to get the surgery and get it done now. It’s really scary to think that I was being forced into making a life-altering decision.” – Participant 59

These women also faced the anxiety of impending biannual surveillance, some of whom have already had some form of prophylactic surgery (e.g., oophorectomy, double mastectomy, salpingectomy, etc.).

A subset of women described taking medication therapies (n=3) to prevent cancer growth. This approach was used in conjunction with surgery with hormone replacement to elevate estrogen to a premenopausal level. Although two women noted having concerns regarding side effects of all preventive measures, both women have completed surgery and continued surveillance. Although only a handful of participants reported using this approach long-term, both stated that these side effects were both physical (due to surgeries/medication) and psychological (stress, anxiety, etc.) but were “manageable for now”. Overall, they
understood the importance of these procedures and routine surveillance despite the side effects and concerns because both identified the risk of future cancers as “too great” to “not do anything”.

Theme 3: Experiences in healthcare. This theme included women’s perceptions of how the healthcare providers handled genetic counseling for breast and/or ovarian cancers (n=34), how patients were treated (n=23), insurance coverage issues (n=16), and the limited accessibility of genetic counseling within some regions of the United States. Themes and associated subthemes are further depicted in Figure 3 and below.

The majority of women explained the different ways that one was genetically tested, such as through a genetic counselor (n=26), primary care physicians (PCP) (n=9), oncologist (n=7), or direct-to-consumer (DTC) testing (n=2). (see Figure 3 or above). Although most participants underwent genetic counseling through a private, licensed genetic counselor, PCPs and oncologists were also found to be common. Depending on who conducted the testing, experiences varied. Of note, out of the 26 women tested through private genetic counselors, 10 noted having negative experiences with counseling, describing problems of provider
“insensitivity” and “ignorance”, making the patient feel “misunderstood” and “overlooked”. No other negative experiences of such magnitude were noted within any other type of healthcare provider.

Doctor-patient communication experienced during genetic counseling was mentioned by 23 participants, with 14 negative and 9 positive experiences noted. Such experiences ranged from “sympathetic”, “knowledgeable”, and “conscientious” to “fear-inducing”, “ignorant”, “rushed”, and “rude” when asked about the counselor/physician. Comfortability and rapport seemed to sway positivity in the eyes of the participants, whereas being “unemotional” or “pushy”, especially relating to surgeries, prompted disapproval. Women reported feeling “illogical” and “unvalidated” during conversations with medical professionals who did not listen to their thoughts and concerns. Participants noted feeling “confident in their medical choices” when physicians took their time to “explain recommendations” and when they were knowledgeable. Participants shared examples of such doctor/patient interactions:

“At first, my mammogram and reconstructive surgeries weren’t covered by insurance, but my gynecologist [who did the genetic testing] wrote a letter to the insurance company and called them until they approved my claim wasn’t just ‘cosmetic’. She went above and beyond to help me and take the time to help me.” – Participant 6

“I had originally gotten my results from 23andMe. My doctor had me retested through a licensed genetic counselor, who found the same result. She got me in for active surveillance the next day – I kid you not – until I could get in to see a surgeon. She probably saved my life.” – Participant 33

“This technician, whether she was trying to be nice or not, was really like hell on wheels. I left that mammogram very aware of my own mortality like I’m going to die tomorrow. Although my mammogram was clear, she took it upon herself to, because of the BRCA, scare me with statements, telling me to immediately get a hysterectomy before age 40, a double mastectomy, and find an oncologist to get me on tamoxifen right now. She also told me to put all my paperwork regarding BRCA in a fireproof safe so that when I’m gone, the information doesn’t die with me. She told me to put a letter with the results with my daughter’s name on it in
Insurance-related issues (n=16) were also identified under this theme and were primarily described as “detrimental” to the genetic counseling experience. Oftentimes this was linked to doctor-patient communication and recommended preventive measures. The most widely cited limitation regarding insurance was the “limited amount of genetic counseling” that is covered by healthcare insurance (n=10). Insurance ranged from full coverage to no coverage. Most women in the sample were recommended to have immediate prophylactic surgeries and if not, then biannual surveillance. A small group (n=6) noted that some biannual surveillance methods (e.g., transvaginal ultrasounds, MRIs) were also not covered by their health insurance, despite being BRCA1/2-positive. One woman described having reconstruction surgery after a double mastectomy that was denied by insurance because she “did not have active cancer” and therefore it was “cosmetic” even though her oncologist wrote a letter to the insurance company stating that she had a BRCA1 mutation.

Losing healthcare insurance coverage, on the other hand, either during the genetic counseling experience or in preventive stages (n=4) was also noted but occurred for reasons not related to genetic counseling such as change or loss of employment. In response, some participants reached out to state resources, but found they were “inadequate” (e.g., no assistance for those losing insurance, no chemoprevention coverage, etc.). To “beat the system” with respect to life insurance, most genetic counselors recommended participants enroll before testing took place to avoid being denied once results were finalized. Only two women were denied life insurance coverage due to a positive BRCA1/2 test result (see Figure 3). Participants shared:

“I had to wait a year to get tested because my parents were in-between insurance coverages. I took the time to come up with a plan if I was positive. At the end, I was comfortable with my plan.” – Participant 42
“At the time [I was going to be tested] I didn’t have insurance. I didn’t want to pay out-of-pocket to go to the doctor and have them tell me my results, so I contacted the company that ran my sample to get the results myself.” – Participant 50

**Theme 4: Emotional reactions to genetic counseling.** This theme was discussed by 33 participants and illuminated a wide range of emotional reactions to genetic counseling and test results. These emotional reactions ranged from extremely negative to extremely positive, highlighting what participants had experienced throughout the genetic counseling experience. Positive emotional reactions to genetic counseling often co-existed with negative counseling reactions. Due to the vast array of emotional reactions, this section will first present the positive and then negative responses.

Self-advocacy and empowerment (n=19) were highly prevalent in this theme, illustrated by the experience of “strength” and “encouragement” that was needed to progress from genetic test results to surgery, surveillance, and/or chemoprevention. Women noted that knowledge had given them the “power to make their own decisions for their future”, including being able to “protect” themselves and their families. Some participants explained how they “share their stories” with others who have experienced multiple personal or familial cancer diagnoses, urging
them to be tested so that they too, can make “informed” preventive health decisions. A few quotes that illustrate these issues are noted below.

“I thought it was all fun and games until that test came back positive. I guess everything happens for a reason.” – Participant 20

“It’s made me more aware and bolder because now I’m not afraid to talk to people about it. If I hear someone has a relative with cancer, I’m like, ‘we need to talk’. I try to at least let people ask questions if they want and act as a resource for them, which makes me feel better, in return.” – Participant 31

“I feel find of empowered by the information that I found by myself…I can take control of my own destiny really. I chose to take the head-on approach.” – Participant 42

“I’m going to worry about it but I’m not going to let it cause anxiety or crush me.” – Participant 55

A total of 16 participants had a feeling or sense of “intuition” before they received results of their genetic counseling, knowing that the test would show a positive result. Such “intuition” was also seen in conjunction with higher perceived sources of risk (own personal or family histories of BRCA1/2 mutations or cancer diagnoses). Some of these women (n=5) also reported having a “plan ahead of time” (i.e., surgery, chemoprevention, or continuous surveillance) for what they would do in terms of a response to such result. Women further described this as impacting their views on genetic counseling and “comfortability”.

“When it came time to get my results, I pretty much already knew. I think I’ve known for a long time and was able to come to terms with it quickly.” – Participant 31

“When I went in for the testing, I felt really comfortable with the time, and comfortable with what I would do if I received a positive result – I had a plan.” – Participant 42

Feelings of acceptance and relief were identified by nine participants, finally “having an answer” for their associated fears and “intuition”. These experiences were attributed to having a positive reaction to the genetic counseling experience, noted by a minority of the sample. Some
viewed receiving their test results as a “breath of fresh air” whereas others noted that they were merely “accepting” their fate. Those who had felt “acceptance” and “relief” after results were given also explained that they were “empowered” by their “new-found knowledge”. Having knowledge and a plan moving forward had further added to their perceived “control” over their own genetic code and survival through a genetic predisposition relating to cancer.

Participants also identified being “thankful” and “lucky” (n=8) after receiving test results. Despite the counseling experience producing a negative emotional response in most participants, a minority of the sample stated that they were “thankful” and “lucky” to have this information, because they would be able to use it to reduce their own risk of cancer. These feelings related to previous notions of knowledge to reduce risk of cancers and taking “control of their own destiny”. Some felt as though being BRCA1/2-positive was a “family curse”, others have reflected on the other end of the spectrum.

“Now I look at this test result as a blessing. I get to do something about my risk of cancer.” – Participant 38

“I’m leaving this experience with perkier breasts than I’ve ever had in my life, so, don’t feel bad for me.” – Participant 42

Negative emotions were more common than positive emotions, discussed by 10 participants in relation to their own mortality (see Figure 3). A subset identified themselves as “pre-vivors” (n=9) because they did not “fit” into cancer patient or survivor categories, not having or surviving cancer. The term “pre-vivor” was a term that is consistent with other recent studies that have examined women with BRCA1/2 mutations (Dean, 2016; Dean et al., 2017; Getachew-Smith et al., 2019; Herndl, 2014). These women explained that this was the way they described their “situation” to others – that they were removing their breasts and “mutilating” themselves in response to their high risk of cancer. Likewise, feelings of “anxiety” and “fear”
were mentioned because of the overall nature these results meant for women in the both short- and long-terms. Likewise, women who tested positive for BRCA1/2 mutations had “difficulty explaining their risk” and “situation” to others, including family members. This term angered some participants, not being able to identify a “place” or “define their experience” in such a way. However, many found solace in separating themselves from those with cancer, determined not to ever develop cancer at all. These women viewed themselves as living in “constant fear of developing cancer” and “live with an incredible uncertainty” relating to impending cancer risk.

“It’s a hard thing to explain to people. It’s like, ‘I don’t have cancer, I just have a really, really high chance of getting cancer in the future, so I’m going to have this really radical surgery.’ That was one challenge -I didn’t know how or what to tell people.” – Participant 23

“I just don’t want to live my life waiting for cancer. There were many days at the beginning that I was despondent, cried a lot. I would go in the shower, taking about half an hour, and sob because I didn’t want my kids to know what I was crying about. It affects them too.” – Participant 38

Participants were also “shocked” and/or “overwhelmed” (n=17) by test results, prophylactic measures, and their impact on personal and familial relationships. They noted feeling “floored”, “stunned” and “knocked off-kilter” when test results were positive for mutations, especially when the individual had little or no cancer history personally or among family members. Feeling “shocked” is considered “normal” for this population as found in previous literature. Oftentimes women explained feeling surprised that they were felt this way.

“I was so overwhelmed. I was sitting there, a perfectly healthy woman who at 33, knew she was going to lose her breasts. It took me weeks, if not months, to come to terms with.” – Participant 6

“The news that I was positive shook me to the core. I literally felt like I was punched in the stomach, and that feeling did not go away.” – Participant 54

The emotions “anger, guilt, and frustration” were also mentioned within the current sample (n=8). Most described the emotions above as “confusing” because testing positive was
through no fault of their own. Women’s feelings were centered around their personal situation (i.e., receiving a positive genetic test result), issues with the genetic counseling experience, familial issues due the nature of test results, and other experiences that may cause reactions such as these. These feelings were also considered “normal”. Women believed it was “out of their control” to be predisposed to having cancers such as these. These feelings were particularly strong in women who did not exhibit any risk factors of having a hereditary BRCA1/2 mutation, but “stumbled across testing”, nonetheless. They were not only angry at themselves because they felt as though they had “asked for it” but also felt “guilty” because they had introduced this information to their families. Several of them mentioned that they would have rather “lived without knowing” their test results.

“Aggravation fell by to devastation.” – Participant 15

“My first reaction wasn’t guilt – that came later. Anger is a very big part of it – there’s a lot of anger.” – Participant 22

“I was able to grieve, and you know like, really let out my emotions. At one point, I think I screamed and punched a pillow.” – Participant 38

Unfortunately, some women expressed feeling a combination of “depression” and “loss” (n=8) (see Figure 3). Reactions to test results were viewed as negative but mixed regarding women’s genetic counseling experience. Prophylactic surgeries and the “push” for immediacy after test results were given was reported to be a main cause for these feelings. Women felt that they “did not know what to do”, “who to go to for help”, or “how to feel”. One woman described depression post-results as feeling “nothing”. One participant who experienced reactions such as these explained “feeling depressed” and “lost for days to months” after receiving such results:

“I started therapy after my surgery [on my own accord], but I wish I started it in advance. Because I think people don’t really prepare you for the loss. People don’t give you a space to talk about how it is a loss and a painful situation.” – Participant 23
Linked closely with depression and loss was participants’ post-surgical body image. For a small group of women ($n=3$), post-surgical “rebounding” was more difficult, especially for those undergoing double mastectomies at premenopausal ages. This was defined by negative feelings focusing on the “elimination of their femininity” and “womanhood”. However, several women viewed their post-surgical bodies as “empowering”, noting that it is “just a body, not my soul” and that she has never “had perkier breasts in her entire life” after reconstruction surgery. These were described especially after double mastectomies, oophorectomies, and hysterectomies, which are often seen as the premise of “womanhood”.

“I feel lucky, but it’s still a little strange to walk in the bathroom and look at myself, not recognizing my own body. That’s the process, you know. It takes a long time. I realized that it’s just my body, not my life.” – Participant 17

“I wanted to be someone who had a positive experience and was willing to share their information...but after the mastectomy...that was the hardest part for me, for a woman, to lose her breasts.” – Participant 42

Negative emotions were also discussed in relation to preventive surgeries and sources of perceived cancer risk. One participant described it was “scary to think” that she was being forced into making a life-altering decision so quickly” in reference to immediate, back-to-back prophylactic surgeries. Women who had reported these emotions had also explained that they felt pressured” into prophylactic surgeries and treatments that they did not believe they were ready for. Similarly, a few participants ($n=3$) believed that they needed additional time to “adjust” to their emotions to test results. Although genetic counseling and test results were impactful and new, some women felt as though they needed time to “sit back and think” about what they want to do with this information.

Although the time between results and referrals to specialists (e.g., oncologist, surgeon) may vary, some women felt as though they were “not given enough time to ask appropriate
questions” regarding their own personal risk, taking in account age, cancer history, and family history. These feelings of “misbelief” and “incredulity” were in response to receipt of a positive test result. When such feelings occur, women suggested it is difficult to think logically and propose questions that are necessary, leading to feelings relating to concern and worry. Lastly, a few participants felt “disrespected” by their genetic counseling experience ($n=2$) (see Figure 3). Women felt as though providers were “insensitive” and “rude” by not listening to what they preferred prophylactically, going against women’s decisions. was small, but important, to the overall genetic counseling premise because it highlighted the importance of respect within this genetic counseling approach, and closely tied into recommendations for future care, discussed in the next section.

**Theme 5: Future recommendations.** This theme identified different ways in which the genetic counseling experience can be improved. Thirty-one women gave opinions on how genetic counseling can be improved for women who undergo $BRCA1/2$ genetic counseling for breast and/or ovarian cancers in the future.

Suggestions for future recommendations included: 1) “guidance and standards” in genetic counseling practice (e.g., mental health therapy referrals, prophylactic decision-making meetings, follow-up appointments, etc.) ($n=24$); 2) “additional education” for both the public and healthcare providers on the risks associated with having $BRCA1/2$, testing approach, and prophylactic measures ($n=20$); 3) more “sympathetic” medical professionals ($n=10$); 4) and
“more genetic counselors” \( (n=4) \) especially in rural areas of the country (see Figure 3).

Participants shared:

“After what I had just gone through, I needed a doctor or medical staff or whoever to understand what had just happened – what this meant for me. I desperately wanted someone to know where I was coming from, and how fearful I was. There was nothing like that for me.” – Participant 13

“I like the knowledge is power standpoint. What can I do now, knowing this information, and be proactive?” – Participant 21

“I know doctors who are ignorant who tell patients ‘oh there’s always this sort of risk; men can’t pass it on…I mean, doctors are telling patients this, and to me, it seems bizarre.” – Participant 52

The desire for additional guidance and standards for \( BRCA1/2 \) genetic counseling across the country \( (n=24) \) were also highlighted, with important elements to be included: 1) information on “local support groups” \( (n=15) \); 2) “additional follow-up genetic counseling visits” (that are covered by insurance) to ask detailed questions that may have not been thought of at the initial appointment \( (n=12) \); 3) post-result “family- or talk-therapy” \( (n=11) \); 4) “information on available preventive choices” \( (n=5) \); 5) “specific risk assessments” to those who test positive based on personal and clinical characteristics \( (n=4) \); 6) and “reproductive counseling”, especially for younger patients \( (n=1) \). Twelve women felt as though they were given test results and were “expected to know what questions to ask” at that meeting, right at that moment, whereas others “never received a chance to ask” such questions. Women, therefore, were expected to “process their results and emotions simultaneously” and be able to ask necessary questions about future care and treatment. The following quotes highlight the experiences of women who received test results and were expected to make “the big decisions” afterward.

“I think follow-up genetic [counseling] appointments are a great idea. You get all this information, and not to mention, life-changing news, and you’re supposed to be ready to ask questions after? I just sat there and shut down. Having that time to process would have been great for me.” – Participant 3
“That’s how I knew I had it – from a freaking webpage. I was sitting there at this computer, I was at work, and I just started bawling! It’s a moment I’ll never forget, looking at the screen and it said, ‘BRCA2’ and I was like, ‘holy shit’. – Participant 6

“So, it’s like, what the fuck? Every time I feel like I made my mind up about surgery, I find some other research that says something different.” – Participant 33

“You should be hooked up with someone that walks you through the process and you know, follows you until the end.” – Participant 58

Some participants suggested updated standards and guidance regarding the need for mandated therapy:

“I wanted some immediate support in place, some kind that they offer you so that you’re not walking away with only doctor’s numbers, and people to call who are going to, as I was putting it for years, mutilate me.” – Participant 22

“It’s probably one of the hardest things that you have to hear in your life. Maybe genetic counselors should be more aware and willing to offer a referral to psychological services that are available or support services.” – Participant 42

“I wish there was a way to identify counselors with different specialties. They call us ‘previvors’, which I don’t care for as somebody who has this positive test result, but I wouldn’t feel comfortable going to a counselor who treats people with cancer because now I can see them sitting there going, ‘you really think you got problems’?” – Participant 57

Most women made it clear that if individuals receive serious and “life-altering” test results such as these, that there should be “mandatory support services in place to help” women no matter genetic test result to assist them and their families. Due to this life-altering change, some found it “incredulous” that no services were offered to them, having to find them by themselves if at all.

Theme 6: Family support and communication. Familial support and communication were themes that emerged from the interviews (n=29). This theme was comprised of many subthemes relating to familial relationships resulting from a shared genetic code, and therefore inheritance of BRCA1/2 mutations, and their interaction during and after genetic counseling. This
section also focused on how family support and communication (or lack thereof) affected women with BRCA1/2 mutations.

Family “solidarity” \( (n=23) \) was a common occurrence within this sample and was identified as one of the main supports for testing positive. Many family members noted feeling a sense of “solidarity” because of this mutation that they have not felt before. They were “no longer alone”, but “going through this process and worry together”, using each other as their own support system.

“My older brother is positive too, and he kind of looks at it more along the lines of, ‘Well, we know how we’re going to die.’ I keep talking to him about it, telling him it’s not a guarantee… I’m pretty positive about it and I’m glad to know that I’m able to do things and try to look on the bright side.” – Participant 39

“Between my two sisters and I, who are positive, I didn’t want to be left out. I can’t believe I’m saying this, but I would have felt left out if I didn’t have BRCA.” – Participant 49

“I have a brother and sister, who are both positive, so that makes all of us. It’s our Ashkenazi heritage.” – Participant 33

A small number of participants noted having “lost touch with family members” \( (n=6) \) which reportedly increased their stress and anxiety regarding genetic counseling, because they themselves are the “bearers” of test results. These women described themselves as “nervous and anxious” because they had to approach family members that they did not speak to (mostly due to previously unrelated familial conflict), so they could be made aware of such information and
tested. Although disseminating test results to close family members was stressful, explaining test results to family members who had lost touch due to conflict was even more so.

Noting the sanctity of familial “legacy” was introduced by a small, but persistent group of participants (n=4) that introduced the importance of their own familial legacy. They did not want BRCA1/2 mutations to be known as the “family curse” to their children, grandchildren, and so on. These women did not want to be remembered for having BRCA1/2 or being “tainted”. Several women also worried that they would “die from cancer before having the chance to have children” or being concerned about “passing the mutation down” to their children. One participant stated:

“You find out your genetics is bad, you’ve got to wrap your head around that and think about my children, my grandchildren, the family line.” – Participant 15

Gender misconceptions were also noted by nine participants explaining that their family members held misconceptions and stereotypes relating to breast cancer, BRCA1/2 mutations, and how it is inherited. Seven participants had male family members that thought “men could not inherit the BRCA1/2 genetic mutation”, whereas four believed that these mutations “do not affect males directly”. Lastly, three participants held the belief that “males who are BRCA1/2-positive cannot pass on this mutation to their children” (see Figure 3).

Research Question 2: Women’s Preferences for Genetic Counseling

Research question two examined women’s preferences regarding genetic counseling approach. The interview question, “Based on your experiences, which form of genetic counseling would you have preferred (i.e., individualized or family-based genetic counseling) and why?” (Appendix E) was used to assess these preferences.

Among 34 participants, the majority had undergone individualized genetic counseling (n=31; 91.2%), with only three (8.8%) having family-based genetic counseling. However, when
asked which approach of genetic counseling participants would have preferred, the majority (n=21; 61.8%) noted that family-based genetic counseling would have been favored (see Table 3). Independent samples t-tests and chi-square analyses were conducted to identify sample characteristic differences between those who had preferred individualized versus family-based genetic counseling. No differences were found (see Table 3).

“If I could have gotten tested with my brother and sisters, it could’ve saved me and them a lot of trouble. We would all get it over with together instead of having to be tested one-by-one” – Participant 13

“Having my family there, even if they turned out to be negative, would have been important to me. We could all lean on each other for support and future care” – Participant 47

Of the 13 women that preferred individualized genetic counseling, eight (61.5%) noted that they did not have any familial connections that would warrant family-based counseling, such as family members who have already tested positive for BRCA1/2 genetic mutations. The remaining five individuals (38.4%) reported being the first to undergo genetic counseling within their family. Details can be viewed in Tables 2-3.

**Research Question 3: Psychosocial Wellbeing of Women with BRCA1/2 Mutations**

Research question 3 hypothesized that women who have received a positive BRCA1/2 genetic test result will have higher stress and anxiety, and worse HRQoL and overall perceived health than those in the general population. To address the third research question, these data were analyzed through group mean differences via independent samples t-tests between the current sample (N=60) and previously published, general female population on the same standardized measures.

Overall, hypothesis 3a was supported. Women in the current sample scored significantly worse on state (M=46.0, SD=13.1) and trait anxiety (M=44.2, SD=11.9) than the general female
population ($M = 35.7, SD = 10.6; M = 34.9, SD = 9.22$, respectively). Perceived stress was also significantly higher within the current sample ($M = 18.9; SD = 6.77$) than the general female population ($M = 13.7, SD = 6.6$). Details are presented in Table 4. Perceived breast cancer risk, as we measured in our study, could not be compared to scores in the female general population. The items used in the current study was compiled from numerous questions from past literature (Audrain et al., 1995; de Castro e Silva et al., 2013; Skinner et al., 1998) with no standardized scales to compare mean scores to. Instead, the current analyses mirrored how Augestad and colleagues (2017) conducted their analyses. The current study highlighted item-level value scores on an item-level range from 0 (low risk) to 10 (high risk). Item-level scores hovered around the median (e.g., 5) but were slightly higher ($M = 6.47, SD = 3.61$) on the question relating to the perceived risk of getting breast/ovarian cancer in relation to others your age and sex. The remaining item-level variables are displayed in Table 5.

Hypothesis 3b was also partially supported, as the current sample reported significantly higher HRQoL SF-36 scores than the general female population published by Ware and colleagues (1993), depending on the subscale (see Table 4). Women in the current sample scored significantly worse on physical role limitations ($p < .05$), emotional role limitations ($p < .05$), energy ($p < .01$), emotional well-being ($p < .01$), social functioning ($p < .01$), and mental component scores ($p < .05$) than in the general population, as hypothesized. On the other hand, there were no significant differences between groups on scores relating to physical functioning, bodily pain, general health, and physical component scores, so this hypothesis was not completely supported. Overall, the current sample scored worse on more emotionally driven domains of HRQoL, consistent with previous literature among those who have just tested positive for $BRCA1/2$ mutations or in those reporting long-term psychosocial effects relating to the genetic counseling
process or the stress of cancer risk (Kiechle et al., 2016). Schwartz and colleagues (2004) found that more physical distress relating to the genetic counseling (and follow-up) processes occurs more often after prophylactic surgeries are completed, consistent with current findings. Therefore, HRQoL domain scores may vary in response to time since genetic counseling and preventive measures undergone (Schwartz et al., 2004). See Table 4 for mean scores and group difference statistics.
CHAPTER 6

DISCUSSION

The purpose of the current study was to explore the lived experiences of women who have received genetic counseling and tested positive for BRCA1/2 mutations. Six main themes emerged in relation to the participants’ own experiences of undergoing the genetic counseling approach: 1) sources affecting perceived risk, 2) preventive concerns and decisions, 3) experiences with healthcare, 4) emotional reactions to genetic counseling, 5) future recommendations, and 6) family support and communication. The current study also introduced the type of genetic counseling approach (i.e., individualized, family-based) BRCA1/2-positive women preferred. Although most women completed individualized genetic counseling, they would have preferred family-based genetic counseling if presented that option. Of note, most women who preferred individualized genetic counseling also stated that they did not have any at-risk family members that would warrant family-based counseling. Psychosocial outcomes relating to participants’ genetic counseling experience were reported and compared to the general female population. Overall, physical role limitations, emotional role limitations, energy, emotional wellbeing, social functioning, and summed mental component scores were significantly worse in the current sample compared to the general female population. Although this was not expected, it remains important to include in future research. The remainder of the discussion integrates current study findings within the context of existing literature, and discusses implications, and future areas of research and practice.

Women’s Experiences with Genetic Counseling

The primary objective of the study was to examine women’s experiences of genetic counseling in the U.S. and was identified within the first research question. Key findings of this
objective included the identification of six themes (i.e., sources affecting perceived risk; preventive concerns and decisions; experiences with healthcare; emotional reactions to genetic counseling; future recommendations; and family support and communication). Within the first theme, women noted how family and personal histories of cancer are significant sources in perceiving their own cancer risk. Although this finding mirrors those of previous literature, it was only in relation to cancer diagnoses among limited family members, those who do not fit the criteria of being “at risk” for a mutation, not those who are BRCA1/2-positive (Katapodi et al., 2004; Taber et al., 2015). Interestingly, women in the current sample explained having higher perceived cancer risk if they had family or personal histories of cancer considering a positive BRCA1/2 genetic test result. Therefore, future research should explore the concept of perceived breast and ovarian cancer risk within this population, as this information could be used to inform educational resources for patients and at-risk family members. A more thorough understanding of women’s perceived risk and sources of risk may help inform healthcare providers conversations with women undergoing genetic counseling for BRCA1/2 mutations. Resources can be provided to patients and at-risk family members to help understand genetic test results. For instance, Keohane and colleagues (2017) found that in a pre-post app-based educational intervention, accuracy of perceived cancer risk among those living with BRCA1/2 mutations was 30% more accurate after the intervention than the control group. New online resources, such as AssessYourRisk.org (2020), provide women with a personalized prevention plan, showing them if they have average, increased, or high risk of either (or both) breast/ovarian cancers, their key risk factors, what they can change to lower risk, and what steps they can do to protect themselves in the future. With additional information readily available to these populations, these individuals and their families can help manage cancer worry by having more accurate risk appraisals.
The second theme women discussed was the notion of preventive decision-making and related concerns. Women’s beliefs were that prophylactic surgeries are necessary within this population, as other options are viewed as “just buying time”. Women felt as though healthcare providers were “pushing” prophylactic surgeries on them immediately after receiving genetic test results, causing them to feel “overwhelmed”, “disrespected”, and “dehumanized”. Women described “having no choice” and feeling “trapped” to follow-through with prophylactic surgeries because ongoing surveillance does not “prevent anything”. Those opting for surveillance reported this immediacy from medical professionals as well. Although immediacy was noted in a previous study by Myklebust, Gjengedal, and Stromsvik (2015), few studies have discussed pressure by provider type felt among this population (Caiata-Zufferey et al., 2015; Klitzman & Chung, 2010). Although preventive choice varied by reproductive age (premenopausal=biannual surveillance, possible double mastectomy; postmenopausal=surgeries, chemoprevention), it is possible that there are additional factors that were not identified. Therefore, future research should focus on understanding these patterns at a deeper level. Similarly, research may want to explore differences in preventive experiences by prophylactic decision and/or by surgery type to further inform resources for patients, their families, and doctors alike. Additional education and sensitivity training may be beneficial for medical professionals who work closely with women of this population. Knowing women’s possible reactions to such choices as well as the emotional aspects of the decision-making process, medical professionals can improve communication with BRCA1/2-positive women, presenting more sensitive, appropriate risk reduction options, considering age and reproductive choices. Additionally, referrals to mental health professionals and therapists specifically trained providing support to women who test positive for BRCA1/2 may be beneficial to introduce as standard
practice in genetic counseling programs. Prophylactic decision-making remains an important aspect of risk management, it is important to support women and their families during the decision-making process. The introduction of family therapy and other mental health resources may act to mitigate a portion of the stress and anxiety that rest on individuals during these decision-making processes. Resources involving decision-making tools for prophylactic measures may be beneficial such as online or in-person support groups that allow for open discussion of preventive decision-making and other BRCA1/2-related questions or concerns, as suggested by Hoskins and colleagues (2017). Such decision-making tools exist, as published by Jabaley, Underhill-Blazey, and Berry (2019), Krassuski and colleagues (2019), and some are currently being tested in clinical trials by the National Cancer Institute (NCI, 2019), but should be implemented within this population to ease the decision-making processes.

The third theme explored women’s experiences in the healthcare system during and after testing for BRCA1/2 mutations. Most women noted that they desired improvements in doctor-patient communication, noting some providers as “ignorant”, “pushy”, or “unemotional”. Although this finding has been discussed in previous research (Dean, 2016; Dean et al., 2017), the current study highlights women’s varied experiences (positive and negative) by provider type (i.e., genetic counselor, PCP, gynecologist, oncologist). For instance, it is possible some providers who initiate genetic counseling by collecting family history and presenting results, may not be as versed in the genetic factors related to BRCA1/2 mutations as a trained genetic counselor; hence, partially explaining some of the variability in women’s experiences. Future research should explore the role different types of healthcare providers have on women’s experiences and different psychosocial health outcomes. It may be important to examine how many providers, outside of genetic counseling, are providing counseling to their patients, to
inform future policy and field standards. Medical professionals may not have specific training in conducting genetic counseling may not be able to explain what a positive test result means for patients. Therefore, education and sensitivity training approaches may be beneficial for all who genetically counsel patients regarding BRCA1/2 mutations (Roter et al., 2008), what a positive test result truly means for a woman at differing ages, how to properly (and accurately) reduce risk, and how to present patients and their families with this news. Similar interventions were implemented by Hanoch and colleagues (2014), finding that by offering online training opportunities for genetic counselors on how to present test results, some negative emotional reactions were reduced among patients. Clinically, it may also be beneficial for women who test positive for BRCA1/2 mutations to have a choice how they receive their results (i.e., in-person, email, mail, over the phone, webcam) as to mitigate negative responses to doctor/patient communication within this population. It also may be beneficial for families to be included in these communicative processes, as identified in past literature (Elrick et al., 2017) and the last theme of family support and communication. Both are additional areas for future research.

The fourth theme highlighted women’s emotional reactions to the genetic counseling experience. Most of the current sample described a range of emotions relating to genetic counseling approaches and subsequent test results, varying from extremely positive (i.e., “empowerment and self-advocacy”; “thankful and lucky”) to extremely negative (i.e., “depression and loss”; “anger, guilt, and frustration”). Women in this sample also described themselves as “pre-vivors” because they did not know how to explain to others why they were undergoing drastic prophylactic measures when they appeared “perfectly healthy”. Participants could not categorize themselves as cancer patients or cancer survivors, so to easily field questions about being BRCA1/2-positive and the steps being taking to minimize cancer risk, they
identified themselves as being a “pre-vivor”, which is consistent with previous studies (Dean, 2016; Dean et al., 2017; Getachew-Smith et al., 2019; Herndl, 2014). Previous research has also found similar range of emotions in this population (Cicero et al., 2017; Gonzalez-Ramirez et al., 2017; Hesse-Biber & An, 2016; Oberguggenberger et al., 2016; Wenzel et al., 2012). Like recent literature (Heiniger et al., 2015; Zimmermann et al., 2020), the experience of “intuition” was described by women in the current study. Women reported knowing that they would test positive before receiving their BRCA1/2 results, mostly in women within high-risk families (those who have many cancer diagnoses and occurrences and recurrences) (Heiniger et al., 2015). Also, these same women reported having a “plan laid out for future procedures” (e.g., surgery, chemoprevention, continuous surveillance, or a combination) “quite a bit” before their test results were given to them. Planning various trajectories helped them feel as though they were in “control” of their own cancer risk and destiny. Although intuition seems to be a relatively “new” way of describing ones feeling about cancer risk prior to diagnosis, other research has similarly discussed something similar, called “controlled-fate” (Zimmermann et al., 2020), which is related to individuals’ previous life philosophy. It is unknown why this feeling occurs, whether it occurs in relation to sources of perceived risk, or a combination of factors. Future research could gain insight into understanding the role of intuition on women’s perceived risk and subsequent psychosocial health outcomes. Although emotional reactions are not mutually exclusive, and can co-occur, it is imperative that clinicians and medical professionals working with this population be aware of this distinction. Perhaps, clinicians who provide genetic counseling can use the positive emotional aspects as a starting point to and entry way to support women through the common and expected negative emotions (Scherr et al., 2015). Referrals to mental health counseling or family therapy (Hoskins et al., 2017) and information on support groups
(Landsbergen et al., 2010) for those that are struggling emotionally have been found to mitigate long-term patterns such as these. Considering the emotional reaction that immediately follows a genetic test result, resources such as infographic tools to outline possible questions for genetic counselors regarding next steps may assist women in making decisions despite feeling overwhelmed. Like the information published by the Illinois Department of Public Health (2020) and beBRCAware.com (2020), infographic tools can present possible questions and prompts for women who plan or are undergoing genetic counseling or have tested positive for *BRCA1/2* mutations.

The fifth theme represented how the future of genetic counseling could be improved to help women and their families through this challenging time. Better clinician knowledge and understanding of women’s risk was imperative for every woman in the current sample. The need for additional education and sensitivity training, as well as for the need guidance and standards relating to the genetic counseling experience, were noted. In fact, these findings mirrored suggestions that have been discussed in previous literature, including access to genetic counselors (Markens, 2017), finding applicable reproductive options and specialists (Petrucelli, Daly, & Pal, 2016; Rojas et al., 2019), finding support groups (Dean et al., 2017), referrals for post-result family therapy (Godino et al., 2016), and the choice to have follow-up genetic counseling appointments after results are presented to ask additional questions (Kolor et al., 2017). The current study supports previous literature by introducing two suggestions: 1) sensitivity training for medical professionals and 2) offering personalized risk assessments based on the individual and not population-based estimates. Sensitivity training has been suggested in the past by Roter and colleagues (2008) to mitigate negative emotional reactions to counseling results. Mays and colleagues (2012) conducted an intervention that offered personalized cancer...
risk assessments to low-income women with \textit{BRCA}1/2 mutations, finding that those who received assessments utilized preventive measures (e.g., surgery, surveillance, chemoprevention) more often than those who did not receive an assessment. Additionally, genetic counseling was also limited geographically. Accessibility can be expanded by applying telemedicine or web-based genetic counseling that allow secure, video or text chats with counselors, which may be especially important for women living in rural areas of the United States (McDonald et al., 2014). Recent telehealth interventions have applied genetic counseling for \textit{BRCA}1/2, finding that such applications were used mainly by younger individuals (>60 years) (Goldstein et al., 2018) and that these practices have the ability to reduce geographical barriers for those living in rural areas (Rhoads et al., 2020) as well as provide an increasingly-available alternative option for patients (Vrecar, Hristovski, & Peterlin, 2017).

The sixth theme identified how family support and communication (or lack thereof) may affect women with \textit{BRCA}1/2 mutations during and after the genetic counseling experience. Women mentioned the importance of “family solidarity”, identified by the strength and support family members provide one another through this trying time. Such findings are consistent with previous literature, noting that the availability of family support through genetic counseling is imperative for mental health (Mendes, Chiquelho, Santos, & Sousa, 2010). Future research should identify women who may lack family support and ultimately provide appropriate resources to mitigate the adverse effects on mental health. Current study findings highlight the need for assistance in disseminating test results, especially among women who have lost touch with family members who now may be at risk for \textit{BRCA}1/2 mutations themselves. Assistance for disseminating test results could be given in a brochure or interactive webpage format to provide examples of how to begin conversations with family members about \textit{BRCA}1/2 risk, as suggested
by BeBRCAware.org (2020) and NCI (2020) resource websites. Genetic counselors could disseminate test results to all family members deemed at-risk (such as in the family-based genetic counseling approach), so that no family member would be left unaware and the burden of disclosure would not be placed on one individual, but this has been the topic of ethical discussions. Montgomery and colleagues (2013) offered an alternative, providing information to individuals tested positive for *BRCA1/2* mutations on how to disseminate test results in several ways: 1) identify relatives who could benefit from the information; 2) choosing communication format (e.g., phone, in-person, email, text, web-based video); and 3) assessing how much families might want (or do not want) to know. These findings may support testing family-based counseling approaches both during and after genetic counseling or providing additional guidance to individuals to families on how to disseminate such information as to mitigate negative emotional reactions.

**Women’s Preferences for Genetic Counseling Approach**

Most women preferred the family-based genetic counseling approach over the individualized genetic counseling approach. The *BRCA1/2* mutations are a hereditary, familial mutation, so it only seems appropriate to approach genetic counseling for these mutations in a similar way. Although it was clear that more women in the current sample completed individualized genetic counseling, it is possible that they only did so because it was the only option given or it was the only option covered by insurance. This finding and recommendations have not been explored in the existing research, but its importance cannot be overstated. Therefore, future research should examine potential differences in patient satisfaction and overall patient experience between the two genetic counseling approaches. Future studies can focus on how test result dissemination occurs within each genetic counseling approach as well as
understanding familial support during this experience. The prophylactic decision-making process can be further explored by including family members in discussions.

**Psychosocial Health & Wellbeing of the Current Sample**

Women who have *BRCA1/2* genetic mutations have higher stress and anxiety than the general population. This finding is consistent with past literature as those who are predisposed to cancer, have had cancer, and have survived cancer have higher levels of stress and anxiety (Madlensky et al., 2017; Nordin et al., 2011; Schwartz et al., 2014). The current study findings suggest that genetic counseling for *BRCA1/2* can be a life-changing, stressful event and experience. Understanding more about the stresses and anxiety associated might lend insight into future areas of research and care that could assist women testing for these mutations. Therefore, it would be increasingly relevant to prospectively follow women from test results as they progress through prophylactic surgeries and/or surveillance, identifying major points of stress and anxiety. This in turn, would inform future practice and interventions. Research should focus on implementing larger, population-based studies to collect information involving prophylactic surgery and treatments, *BRCA1/2*-related cancer diagnoses, and recurrences to understand how prevalent feelings of stress and anxiety are within this population. Additional training for medical professionals who work with this population should occur on a regular basis to further inform these individuals how best to care for women undergoing genetic counseling and preventive measures. Lastly, to mitigate anxiety and stress related to the lack or incongruence of information about *BRCA1/2* mutations, resources and a referral to mental health services can be provided to women that are identified as struggling.

Health-related quality of life (HRQoL) varied in the current sample. Physical and emotional role limitation, energy, emotional wellbeing, and social functioning domains were
worse compared to the general U.S. female population. Changes in HRQoL domains such as these may depend on if BRCA1/2-positive women are actively undergoing prophylactic treatments or not (Schwartz et al, 2004). This is valid, as undergoing prophylactic surgeries for BRCA1/2 risk reduction are considered major and are very time- and energy-intensive, as is any surgery. As shown in previously literature (Metcalf et al., 2015), HRQoL follows an up-and-down pattern over time, relative to when choices are made regarding prophylactic surgeries and if any cancer diagnoses occur. Similarly, Young and colleagues (2017) found that when BRCA1/2-positive women are actively undergoing prophylactic treatments or struggling with genetic counseling information or processes, their energy levels are lower due to increased worry and anxiety. As HRQoL has been shown by the current study to vary, it is important to understand when to help target future resources at critical times during the genetic counseling. Mental health counseling referrals for patients who are identified as struggling may be important to mitigate decreases in emotional health and HRQoL (Hoskins et al., 2017). Considering these findings, population-based prospective studies are needed prior to genetic counseling to identify factors that impact the domains of HRQoL more negatively to further inform practice and resources. These results can further inform clinical practice as to how HRQoL may impact (if at all) the genetic counseling and prophylactic treatment experiences following a BRCA1/2-positive test result. As suggested above, future practice can introduce resources such as mental health and/or family counseling to mitigate decreases in emotional and social functioning and improved overall HRQoL.

Limitations

It is important to interpret the current study’s findings considering its limitations. The current study was cross-sectional in nature, which does not allow causal relationships between
genetic counseling or psychosocial health. Recall bias also may have been an issue, as participants were asked to recall information up to five years earlier regarding their genetic counseling experience. Self-report survey information was used for a portion of this study, which could introduce response bias in participants. It is not extremely common to be tested for \(BRCA1/2\) mutations, so a widened five-year limit was set to ensure that participant saturation was met. Generalization was limited to educated, mostly insured, non-Hispanic White women who had tested positive for \(BRCA1/2\) genetic mutations within the United States. These participants were recruited from online support groups, which may introduce bias by being more open and willing to share experiences than others not in support groups. Moreover, certain personality type, extroverts, may seek out these groups and participate in research (Brown, Tang, & Hollman, 2014; Houlihan & Tariman, 2017), more so than introverts. Lastly, follow-up interviews could have been included so that researchers could gain clarification or ask additional questions that arose during analyses.

**Need for Education, Best Practices and More Research**

Although the current study was primarily exploratory in nature, much was learned from its results. This study suggests that women have unique experiences during genetic counseling for \(BRCA1/2\) mutations. Women highlighted their need for more knowledge and information for themselves, their families, and healthcare providers. Therefore, education resources should be created and disseminated to individuals, family members and healthcare professionals regarding what to expect during the genetic counseling experience. Following a positive test result, education resources for women with \(BRCA1/2\) mutations can be in the form of phone or computer applications (i.e., apps) that allow video chats or text chats for support, in place of support groups (Landsbergen et al., 2020). App-based educational resources can also be used to
increase cancer perception accuracy within this population, as shown by Keohane and colleagues (2017). Online and easily accessible breast/ovarian cancer assessments can help provide women with key factors that increased and decreased cancer risk, and what they can do to protect themselves and become more aware of their own situation (AssessYourRisk.org, 2020). Women should also have access to educational resources that assist in having conversations with family members about their BRCA1/2 risk. Related tools can be created and/or disseminated, to be used as a guide to help women ask questions during genetic counseling appointments, especially considering intense, albeit normal, emotional reactions. Tools such as these have been published by the National Cancer Institute (NCI, 2020) and BeBRCAware.com (2020), highlighting the importance of accessible resources for asking questions to inform themselves and family members. In response to participant concerns regarding joint prophylactic decision-making, roadmap websites, featuring quizzes, applications, or infographics may be important to implement and widely distribute, so that participants can have a resource to refer to when making decisions outside of oncologists’ offices. Some of these tools have been published by Jabaley, Underhill-Blazey, and Berry (2019), Krassuski and colleagues (2019), and the NCI (2020), focusing on an educational, patient-focused decision aid for families to use when making prophylactic decisions due to a positive BRCA1/2 test result. Research can also examine the subthemes relating to pre-vivors, intuition, and the hard truth. Overall, future research should aim to include women and families of racial, ethnic, and sexual minority populations as well as women from disadvantaged geographic, health, and socioeconomic backgrounds.

Findings from the current study suggest several considerations for clinical practice and additional research. Effective doctor/patient communication requires further understanding, focusing on the emotional needs of women who test positive for BRCA1/2 mutations. Providers
should also be aware, through sensitivity training, of the emotional range that \textit{BRCA1/2} test results may evoke among women, as understanding the emotional side is necessary for the comfort of women undergoing testing (Hanoch et al., 2014; Scherr et al., 2015). Providers conducting genetic counseling should also give patients the option of how they would like to receive their results, by phone, email, mail, webcam, or in-person. Providers should also have the resources listed above readily available for patients undergoing genetic counseling and preventive measures related to \textit{BRCA1/2} mutations. Lastly, adding support staff to genetic counseling programs to field follow-up appointments and questions may be beneficial for all parties. The growth of telemedicine genetic counseling (through phone or video chat) may be the future of the field, focusing on expanding access to patients across the nation. These services and applications would be especially true for women who live in more rural areas with limited access to resources and services (McDonald et al., 2014). Although the reactions, needs, and expectations of women with \textit{BRCA1/2} mutations may vary, it is imperative to note that most require emotional assistance throughout and after the genetic counseling processes. Additional actions may be introduced from a family systems perspective, one that seems to mirror the hereditary nature of \textit{BRCA1/2} mutations. Care for this population may want to focus on what is known from the current and previous research – that women with these mutations require additional emotional supports. Family-inclusive mental health counseling, for instance, involve the entire family, increasing communication and support, to possibly reduce the burden and struggle of women testing positive for these mutations.

\textbf{Conclusion}

This study provides new and important information for understanding women’s experiences and psychosocial health during the genetic counseling experience. Several themes
emerged, some consistent with previous literature and some new themes. Women discussed the importance of being tested as a family unit, identifying places that could use improvement, and psychosocial issues relating to counseling. This recommendation can serve to inform future research including testing women’s experiences and outcomes of individual vs. family based genetic counseling approaches. Although genetic counseling may only be conducted for a minority of the general population, these women’s experiences, thoughts, and suggestions are important to understand. With genetic counseling becoming more available, it is possible that women may need more post-counseling resources that are not readily available (i.e., genetic counseling, support groups, therapy, specialists, etc.). The possibility of changing the genetic counseling approach, as suggested in this study, is multi-faceted, requiring additional research testing the two different approaches.
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Table 1.
Eligibility 4-item Screening Survey

<table>
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<tr>
<th>Question</th>
<th>Answer</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Have you undergone genetic testing for <em>BRCA1</em> and/or <em>BRCA2</em> genetic mutations within the past five years?</td>
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</tr>
<tr>
<td></td>
<td>You do not fit the criteria of the current study. Thank you for your interest at this time.</td>
</tr>
<tr>
<td></td>
<td>Yes</td>
</tr>
<tr>
<td></td>
<td>Continues to Question 2</td>
</tr>
<tr>
<td>2. Have you tested positive for either <em>BRCA1</em> and/or <em>BRCA2</em> genetic mutations?</td>
<td>No</td>
</tr>
<tr>
<td></td>
<td>You do not fit the criteria of the current study. Thank you for your interest at this time.</td>
</tr>
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<td></td>
<td>Yes</td>
</tr>
<tr>
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</tr>
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<td>3. Are you female?</td>
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</tr>
<tr>
<td></td>
<td>You do not fit the criteria of the current study. Thank you for your interest at this time.</td>
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</tr>
<tr>
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<td>Continues to Question 4</td>
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<td>4. Are you currently undergoing hereditary breast cancer counseling for these mutations?</td>
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<tr>
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<td>You do not fit the criteria of the current study. Thank you for your interest at this time.</td>
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<td>No</td>
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<td>Enrolled in study</td>
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Table 2.
Demographic Characteristics of the Total and Subset Samples

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<th>Survey Sample (N=60)</th>
<th>t-test</th>
<th>( \chi^2 )</th>
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<td>M</td>
<td>SD</td>
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<td>Marital status</td>
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<td>Not married</td>
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<td>26.7</td>
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<td>71.1</td>
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<td>3</td>
<td>5.0</td>
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<td>Midwest</td>
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<td>18</td>
<td>30.0</td>
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<td>West</td>
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<td>20.6</td>
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<td>Previous cancer diagnoses</td>
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<td>Ovarian cancer</td>
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<td>8.8</td>
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<td>82.4</td>
<td>44</td>
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<tr>
<td></td>
<td>N (%)</td>
<td>Mdn</td>
<td>Q1</td>
<td>Q3</td>
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<tr>
<td>----------------</td>
<td>-------</td>
<td>-----</td>
<td>-----</td>
<td>-----</td>
</tr>
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<td>3</td>
<td>8.8</td>
<td>11</td>
<td>18.3</td>
</tr>
<tr>
<td><strong>1 or 2</strong></td>
<td>3</td>
<td>8.8</td>
<td>5</td>
<td>8.3</td>
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<td>Avenue for genetic counseling</td>
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<td></td>
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<td>19</td>
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<td>44.1</td>
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<td>40.0</td>
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<td>21</td>
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<td><strong>Genetic counseling result</strong></td>
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<td>BRCA2</td>
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<td>47.1</td>
<td>29</td>
<td>48.3</td>
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<tr>
<td>Both BRCA1 &amp; BRCA2</td>
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<td>1.7</td>
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<tr>
<td>BRCA &amp; CHEK</td>
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<td><strong>Mental health therapy offered</strong></td>
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</table>

*Note.* $p<.05^*; p<.01^{**};$ Pearson chi-square analyses were unable to be conducted for the ethnicity and preferred approach of genetic counseling because they had no variability.
Table 3.  
Demographic Characteristics Based on Approach of Genetic Counseling Completed

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<th></th>
<th>Individualized</th>
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<th>Total</th>
<th>t-test</th>
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<th>SD</th>
<th>M</th>
<th>SD</th>
<th>M</th>
<th>SD</th>
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<td>43.2</td>
<td>7.71</td>
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<td></td>
<td></td>
<td></td>
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<td></td>
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<td>Education</td>
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<td></td>
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<td>.991</td>
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<td></td>
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<td>17</td>
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<td>Undergraduate or above</td>
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<td>73.2</td>
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<td>50.0</td>
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<td>71.1</td>
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Avenue for genetic counseling

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<th>Number</th>
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Genetic counseling result

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<td>29</td>
<td>48.3</td>
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Mental health therapy offered

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<td>22</td>
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Note. Pearson chi-square analyses were unable to be conducted for the ethnicity and preferred approach of genetic counseling because they had no variability.
### Table 4.
Comparison of Current Sample and General Population Mean Scores on Psychosocial Outcome Subscales (N=60)

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<td>M</td>
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*Note. p<.05*, *p<.01***; Used general female population scores from Cohen et al. (1983) for stress on the PSS, Spielberger (1983) for state and trait anxiety on the STAI, and Ware & Sherbourne (1992) for HRQoL on the SF-36

<table>
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<tr>
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<th>Emotional Role Limitations</th>
<th>Energy</th>
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<th>Social Functioning</th>
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*Note. p<.05*, *p<.01***; Used general female population scores from Ware & Sherbourne (1992) for HRQoL on the SF-36

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<th>Health Change</th>
<th>SF-36 PCS</th>
<th>SF-36 MCS</th>
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<td>M</td>
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<td>24.2</td>
<td>70.6</td>
<td>21.5</td>
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</table>

*Note. p<.05*, *p<.01***; Used general female population scores from Ware & Sherbourne (1993) for HRQoL, general health subscale, and physical and mental component scores on the SF-36; Physical component score (PCS) = physical functioning + physical role limitations + bodily pain + general health; Mental component score (MCS) = emotional role limitations + energy + emotional wellbeing + social functioning
### Table 5.

*Perceived Breast/Ovarian Cancer Risk Item-Level Variable Descriptive Statistics (N=60)*

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<th>SD</th>
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<td>1. What do you believe your risk of developing a tumor on your breast is currently?</td>
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<tr>
<td>2. What do you believe your risk of developing a tumor on your ovaries is currently?</td>
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<td>3.48</td>
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<tr>
<td>3. Compared to others your same age and sex, how would you rate your risk of getting breast/ovarian cancer within the next 10 years?</td>
<td>Range from 0 (low risk) to 10 (high risk) for each item</td>
<td>6.47</td>
<td>3.61</td>
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<tr>
<td>4. Based on your family’s history of breast/ovarian cancer diagnoses, how would you rate your risk of developing breast cancer within the next 10 years?</td>
<td></td>
<td>4.97</td>
<td>3.50</td>
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</table>

*Note. The above items were compiled from past literature by Augestad et al. (2017) and Audrain et al. (1995)*
Figure 1.  
Comparison of the Constructs and Examples of Individualized and Family-Based Genetic Counseling

**Individualized Genetic Counseling**  
*(Standard)*

**DEFINITION**
The current standard - hereditary genetic counseling that focuses on the breast/ovarian risk of the individual only. Patients are expected to disseminate findings from test results to the family unit without the help of a counselor.

**EXAMPLE**
A woman has an appointment for hereditary cancer counseling based upon her gynecologists’ medical opinion, in addition to her history of breast/ovarian cancers. She goes through genetic counseling at a local hospital alone, receives her positive results, and informs family members that may be affected as well. She urges them to undergo testing themselves.  
*Note. It is possible to bring friends/family members to individualized counseling, but they are not tested.*

**Family-Based Genetic Counseling**  
*(Proposed)*

**DEFINITION**
The proposed future care - hereditary genetic counseling that focuses on the family unit who are at risk for sharing a mutated gene and undergo counseling as a family unit. Dissemination of results are completed by a genetic counselor with the entire family unit.

**EXAMPLE**
A woman has been determined at increased risk of a *BRCA1/BRCA2* genetic mutation for breast/ovarian cancer due to family history. When making her genetic counseling appointment, she is asked to bring in specific family members for counseling who are also at risk so that they can go through the process together. The family undergoes testing, and the genetic counselor meets with them as a group, identifying if other family members may also be at risk. Further steps are taken for family members who test positive, including therapy, additional oncological appointments, and surveillance.
Figure 2.
Recruitment CONSORT Flow Diagram

**ENROLLMENT**
- FORCE.org \(n=3,330\)
- FB BRCA Strong \(n=1,299\)
- FB BRCA Sisters \(n=3,218\)
- FB BRCA1/2 Genetic \(n=7,520\)

Open for screening \((n=15,367)\)

**SURVEY**
- Assessed for eligibility \((n=123)\)
- Redirected to online survey \((n=63)\)
  - Completed survey \((N=60)\)
  - Did not complete survey \((n=3)\)

**INTERVIEW**
- 34 interviews thematically analyzed:
  - 35 interviews in total completed (1 removed due to hanging up after 2 min of yes/no answers)

**ANALYSES**
- Objective 1 analyses (RQs 1-2) \((n=34)\)
- Objective 2 analyses (RQ3) \((N=60)\)

Did not meet inclusion criteria \((n=60)\)
- Did not undergo genetic counseling \((n=11)\)
- Did not test positive for BRCA1/2 \((n=10)\)
- Are not female over 18 years \((n=9)\)
- Did not complete BRCA testing \((n=29)\)
- Did not complete screener \((n=1)\)
Figure 3. Qualitative Thematic Analytic Figure (n=34)
Appendix A.
*Image of the Current Study FORCE Posting Webpage*
Appendix B.

Image of the Current Study Facebook Private Group Webpage, UConn BRCA1 and BRCA2 Genetic Testing Study
Appendix C.
Current Study Informational Sheet for Objective 2 Survey Presented to Participants

Informational Sheet

Principal Investigator (PI): Dr. Keith M. Bellizzi
PI Phone Number: 860-486-0663
Title of Research Study: Genetic Counseling for BRCA1/2 Mutations: Patient Experiences, Preferences, and Outcomes of Counseling
Expected Duration of Subject’s Participation: 30 minutes (survey) and 30 minutes (interview)
IRB Number: E18-562
Sponsor/Funding Entity: Connecticut Breast Health Initiative (CT BHI)

Purpose of this Research Study
This research study is about understanding the experiences, preferences, and outcomes of genetic hereditary breast cancer counseling in those who test positive for BRCA1 and/or BRCA2 genetic mutations. Evidence suggests that there are unintended consequences (i.e., anxiety, stress, and decreased quality of life) in individuals who test positive for these mutations, partly due to the expectation to inform family members of their test results. The purpose of this research is to see if women who have tested positive for BRCA1 and/or BRCA2 genetic mutations would have preferred a more family-centered approach to counseling versus the individualized standard that is in place today.

Voluntary Participation
You are invited to take part in this study because you have tested positive for hereditary breast cancer genetic mutations BRCA1 and/or BRCA2 and have completed genetic testing via a licensed genetic counselor.

It is important that you know that participation in this study is voluntary. Before making your decision, please read this information form. If you choose to participate in this research study, you are free to withdraw from it at any time.

We estimate that 40 people will enroll.

Study Procedures
If you choose to participate in the study general time commitments for study participation are as follows: You will be asked to complete a survey and give your email address so that your compensation can be emailed to you via online $20 Amazon e-gift card. You will respond to the
survey at one time (no more than 30 minutes long), and you will be contacted via email to schedule a telephone or Skype (or preferred video conferencing application) with a graduate research assistant for a short, 30-minute interview at another time, after which you receive another $20 Amazon e-gift card via email. You will also be given the option to take only the survey, but not take part in the interview.

More details about the study procedures are provided below.

- **Survey Administration:** You will be asked to complete a brief online survey about your health and well-being. The survey will include questions about your clinical breast cancer information, counseling information, stress, overall mental and physical well-being, and anxiety. If you choose, you can participate solely in the survey portion of this study.

- **Telephone or Skype Interview:** You will be contacted via provided email address to schedule a telephone or Skype (or similar video conferencing application) to interview you about your experiences and preferences for individualized or family-based counseling. You will be asked to describe your experience undergoing hereditary breast cancer genetic testing by examining what you viewed as positive/negative, and how it may be improved. You will also be asked about your preference for communication approach concerning test results (individualized or family-based).

**Risks:**

Before deciding, it is important for you to know about the risks of participation in this research. We have summarized the most important information below. You should review all information about risks before making your decision.

**Common risks:** Common means that more than 10% of people have experienced the risk (i.e., at least 11 out of 100 people). Common risks of this research include

- emotional upset when responding to the survey questions or interview questions
- minimal possibility of breach in confidentiality

**Safeguards:** The risks described above will be minimized by providing counseling referrals or information on support groups. You may skip a question within the survey if it is upsetting to you in any way. If you do not wish to be interviewed, you have the option of partaking in the survey portion of the study only. To ensure quality control, a secure, password-protected database will be constructed to record the names of the participants and emails, with corresponding assigned numerical codes. Once data has been coded and participants paid, identifying information will be deleted.

**Benefits**

No direct benefits to study participants are anticipated, although it is common for participants to find completing questionnaires about their thoughts, feelings, and experiences helpful. Results of the study will provide important information regarding patient-reported outcomes associated with hereditary breast cancer counseling and preferences for future intervention and policy.
Payment

You will be paid for each portion of the study completed. For instance, a $20 Amazon e-gift card will be emailed to you upon completion of the online survey and an emailed $20 Amazon e-gift card upon completion of the scheduled interview. If you choose to complete the survey only, you will still receive the $20 Amazon e-gift card. If you terminate the survey or interview before completion, you will not be compensated.

Privacy and Confidentiality

The confidentiality of your information will be protected as follows:

- All records, including the survey, will be locked in a locked study laboratory at UConn-Storrs accessible only to the research team.
- All electronic files (e.g., database, spreadsheet, recordings etc.) containing identifiable information will be password-protected. Any computer hosting such files will also have password protection. Any file stored on a portable computer device like a laptop will also be encrypted, meaning only people with a valid identification and password can access the specific files on the computer. As each participant is compensated, all identifying information will be deleted from the study files (i.e., data from surveys, recordings) and only the unique 5-digit code will be used.
- At the conclusion of this study the researchers may publish their findings. Information will be presented in summary format and you will not be identified in any publications or presentations.

We will do our best to protect the confidentiality of the information we gather from you, but we cannot guarantee confidentiality. There is always a risk of a breach of confidentiality.

You should know that to ensure that the study is being done correctly the following parties may also inspect your records:
- Administrative oversight areas of UConn Storrs, including representatives of the Institutional Review Board and Human Subjects Protection Office, and Corporate Compliance.

Study Results and Incidental Findings

We do intend to submit our overall findings for general publication and/or presentation. If any of the study procedures reveal other information that is not needed for the study but is important for you to know, we will share that information with you.

Withdrawing

As noted above, you can withdraw at any time without penalty or loss of benefits. If you decide to withdraw we ask that you let us know. Please call Dr. Keith Bellizzi at 860-486-0663 for guidance. You also have the option of withdrawing by mail by writing Dr. Keith Bellizzi, 348 Mansfield Road U-1058, Storrs, CT 06269-1058 or by email at keith.m.bellizzi@uconn.edu or Kate Dibble, the study graduate assistant, at kate.dibble@uconn.edu. If you choose to withdraw
from this study the data that has already been collected will continue to be used and remain in the study database.

Questions

Dr. Bellizzi and Ms. Dibble are willing to answer any questions you have about the research. You are encouraged to ask questions before deciding whether to take part. You are also encouraged to ask questions during your study participation. If you have questions, complaints or concerns about the research, you should call or email Dr. Bellizzi at 860-486-0663/keith.m.bellizzi@uconn.edu or Ms. Dibble at kate.dibble@uconn.edu.

If you have questions about your rights as a research subject you may contact a coordinator at the Institutional Review Board at 860-679-1019, 860-679-4851, or 860-679-4849. You may also contact the Research Subject Advocate at 860-679-3276. You may also call a coordinator at the Institutional Review Board if you want to talk to someone who is not a member of the research team in order to pass along any suggestions, complaints, concerns or compliments about your involvement in the research, or to ask general questions or obtain information about participation in clinical research studies.

Please do not call the IRB number for medical-related issues or to schedule or cancel an interview appointment.
Individuals undergoing genetic counseling for breast/ovarian cancer represent an important population of those undergoing genetic counseling. In recent years, increased attention is being paid to genetic counseling and its outcomes but has thus forth ignored the process itself. This survey contains questions about your well-being, genetic counseling outcomes, as well as demographic information. We know of no better way to learn about these issues than to ask women who have tested positive for BRCA1 and/or BRCA2 themselves. There are no right or wrong answers, so please respond by giving the answer that best describes your situation. You may find some of the questions to be personal or difficult, and you may not have thought about some issues before. Even if you feel you must skip a question, please indicate this by clicking “SKIP” option in that question and proceed to the next question. Your answers to other questions will still be important to us and we will appreciate your responses very much.

All the information you provide is confidential and will not be disclosed to your health care providers or others. The information obtained will be analyzed as grouped data without any personal identification. When you are completing this survey, if any issues concern you about your health, please discuss these with your health care provider. You are also free to contact us by telephone to discuss any issues related to the survey material. If you have any questions, please contact the study coordinator, Ms. Kate Dibble at 860-681-9668 or kate.dibble@uconn.edu, or the Principal Investigator of the study, Dr. Keith Bellizzi at 860-486-0663 or keith.m.bellizzi@uconn.edu. We are very grateful to you for the time you will be taking to complete this survey and for helping us learn how to improve the testing experience of women who are going to undergo genetic hereditary breast cancer counseling in the future.

A. BACKGROUND & CLINICAL INFORMATION

Participant’s Name: ____________________________________________
A1. Today’s date: _____/___/____
   Month    Day    Year
A2. What is your birthdate? _____/___/____
    Month    Day    Year
A3. What is the highest level of formal education you have completed?
   □ Less than high school
   □ High school graduate or GED
   □ Some college or technical / vocational school
   □ College graduate
Some graduate school
Graduate degree

A4. Do you consider yourself to be…?
- Hispanic or Latino
- NOT Hispanic or Latino

A5. Which of the following best describes your race?
- American Indian or Alaska Native
- Asian
- Black or African American
- Native Hawaiian / Other Pacific Islander
- White

A6. What is your current marital status?
- Married or living as married
- Divorced
- Separated
- Widowed
- Single (never married)

A7. What best describes your current employment status?
- Working full-time
- Working part-time
- Full-time homemaker or family caregiver
- Retired
- Student
- Unemployed
- Other, please specify
  ____________________

A8. What is your state of residence? (List of all states)

A9. Did you have health insurance at the time of genetic counseling?
- Yes
- No

A10. What portion did your health insurance cover of your genetic tests?
- All of the fees were covered
- A portion of the fees were covered
- None of the fees were covered

A11. Have you ever been diagnosed with breast or ovarian cancer in the past?
- I have been diagnosed with breast cancer
- I have been diagnosed with ovarian cancer
☐ I have been diagnosed with another type of cancer _____________________
☐ Neither breast or ovarian cancer

A12. Have you completed treatment for your breast, ovarian, or other cancer?
☐ Yes
☐ No

A13. What type of treatment(s) did you undergo for your cancer? Select all that apply.
☐ No cancer treatment
☐ Lumpectomy
☐ Partial mastectomy
☐ Total mastectomy
☐ Partial hysterectomy
☐ Total hysterectomy
☐ Chemotherapy
☐ Radiation
☐ Aromatase inhibitor (AI) therapy (e.g., Tamoxifen, Arimidex, Femara, etc.)
☐ Immunotherapy (e.g., Yervoy, Opdivo, Keytruda, Imfinzi, etc.)
☐ Targeted therapy (e.g., monoclonal antibodies, small-molecule drugs)
☐ Stem cell transplant
☐ Precision medicine (e.g., genetic medicine)

A14. How many recurrences did you experience?
☐ None
☐ 1 or 2
☐ 3 or 4
☐ 5 or more

B. GENETIC CANCER COUNSELING INFORMATION

B1. When did hereditary breast/ovarian cancer counseling begin? ______ / ____ / ______
                                      Month  Day  Year

B2. When did you receive your genetic testing results? ______ / ____ / ______
                                      Month  Day  Year

B3. What state did the genetic counseling take place in? (List of all states)

B4. What was the outcome of your genetic testing?
☐ Negative
☐ Positive

B5. Where did you receive your genetic counseling?
☐ Private genetic counseling office
Hospital
Primary care physician
Direct-to-consumer (DTC) online test

B6. What genetic mutations do you have, according to your genetic test results? Select all that apply.
- BRCA1
- BRCA2
- Both BRCA1 and BRCA2
- Other mutation(s)

B7. Was therapy offered to you after you received your test results, no matter the outcome?
- No, it was not offered.
- Yes, it was offered for those with positive results
- Yes, it was offered for everyone, no matter results
- I don’t know

B8. Individual-based counseling, the norm in most of the United States, is when an individual patient (you) undergoes genetic counseling to test for a specific mutation(s), where you alone receive results. Family-based counseling is when at-risk family members undergo genetic counseling together, all being physically tested for specific mutation(s), and all receiving their results together. With this in mind, did you undergo individualized or family-based counseling?
- Individualized genetic counseling
- Family-based genetic counseling

C. HEALTH & WELLBEING

For each item below, choose one option.

C1. In general, you would say your health is:
- Excellent
- Very good
- Good
- Fair
- Poor

C2. Compared to one year ago, how would you rate your health in general now?
- Much better now than one year ago
- Somewhat better now than one year ago
- About the same
- Somewhat worse now than one year ago
- Much worse now than one year ago
Instructions. The following items are about activities you might do during a typical day. Does your health now limit you in these activities? If so, how much?

C3. Vigorous activities, such as running, lifting heavy objects, participating in strenuous sports

C4. Moderate activities, such as moving a table, pushing a vacuum cleaner, bowling, or playing golf

C5. Lifting or carrying groceries

C6. Climbing several flights of stairs

C7. Climbing one flight of stairs

C8. Bending, kneeling, or stooping

C9. Walking more than a mile

C10. Walking several blocks

C11. Walking one block

C12. Bathing or dressing yourself

During the past 4 weeks, have you had any of the following problems with your work or other regular daily activities as a result of your physical health?

C13. Cut down on the amount of time you spent on work or other activities

C14. Accomplished less than you would like

C15. Were limited in the kind of work or other activities

C16. Had difficulty performing the work or other activities (for example, it took extra effort)

During the past 4 weeks, have you had any of the following problems with your work or other regular daily activities as a result of any emotional problems (such as feeling depressed or anxious)?

C17. Cut down the amount of time you spent on work or other activities

C18. Accomplished less than you would like

C19. Didn’t do work or other activities as carefully as usual
C20. During the past 4 weeks, to what extent has your physical health or emotional problems interfered with your normal social activities with family, friends, neighbors, or groups?
- Not at all
- Slightly
- Moderately
- Quite a bit
- Extremely

C21. How much bodily pain have you had during the past 4 weeks?
- None
- Very mild
- Mild
- Moderate
- Severe
- Very severe

C22. During the past 4 weeks, how much did pain interfere with your normal work (including both work outside the home and housework)?
- Not at all
- A little bit
- Moderately
- Quite a bit
- Extremely

Instructions. These questions are about how you feel and how things have been with you during the past 4 weeks. For each question, please give the one answer that comes closest to the way you have been feeling.

<table>
<thead>
<tr>
<th>How much of the time during the past 4 weeks…</th>
<th>All of the time</th>
<th>Most of the time</th>
<th>A good bit of the time</th>
<th>Some of the time</th>
<th>A little of the time</th>
<th>None of the time</th>
</tr>
</thead>
<tbody>
<tr>
<td>C23. Did you feel full of pep?</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>C24. Have you been a very nervous person?</td>
<td></td>
<td></td>
<td></td>
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<td></td>
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</tr>
<tr>
<td>C25. Have you felt so down in the dumps that nothing could cheer you up?</td>
<td></td>
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</tr>
<tr>
<td>C26. Have you felt calm and peaceful?</td>
<td></td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>C27. Did you have a lot of energy?</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
C28. Have you felt downhearted and blue?

C29. Did you feel worn out?

C30. Have you been a happy person?

C31. Did you feel tired?

C32. During the past 4 weeks, how much of the time has your physical health or emotional problems interfered with your social activities (like visiting with friends, relatives, etc.)?

☐ All of the time
☐ Most of the time
☐ Some of the time
☐ A little of the time
☐ None of the time

How TRUE or FALSE is each of the following statements for you.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Definitely true</th>
<th>Mostly true</th>
<th>Don’t know</th>
<th>Mostly false</th>
<th>Definitely false</th>
</tr>
</thead>
<tbody>
<tr>
<td>C33. I seem to get sick a little easier than other people</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>C34. I am as healthy as anybody I know</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>C35. I expect my health to get worse</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>C36. My health is excellent</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

D. PERCEIVED RISK

Instructions. Please answer the following questions to the best of your ability.

D1. Have you ever been diagnosed with breast and/or ovarian cancer in your lifetime?

☐ Yes
☐ No

D2. What do you believe your risk of developing a tumor on your breast is currently?

☐ Low ☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 ☐ 8 ☐ 9 ☐ High

D3. What do you believe your risk of developing a tumor on your ovaries is currently?

☐ Low ☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 ☐ 8 ☐ 9 ☐ High
D4. Compared to others your same age and sex, how would you rate your risk of getting breast/ovarian cancer within the next 10 years?

- Low
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- High

D5. Based on your family’s history of breast/ovarian cancer diagnoses, how would you rate your risk of developing breast cancer within the next 10 years?

- Low
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- High

### E. PSYCHOSOCIAL OUTCOMES

*Instructions.* The questions below will ask you about your feelings and thoughts **during the last month.** In each case, you will be asked to indicate **how often** you felt or thought a certain way.

<table>
<thead>
<tr>
<th>In the last month…</th>
<th>Never</th>
<th>Almost Never</th>
<th>Sometimes</th>
<th>Fairly Often</th>
<th>Very Often</th>
</tr>
</thead>
<tbody>
<tr>
<td>E1. How often have you been upset because of something that happened unexpectedly?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E2. How often have you felt that you were unable to control the important things in your life?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E3. How often have you felt nervous and “stressed”?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E4. How often have you felt confident about your ability to handle your personal problems?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E5. How often have you felt that things were going your way?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E6. How often have you found that you could not cope with all the things that you had to do?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E7. How often have you been able to control irritations in your life?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E8. How often have you felt that you were on top of things?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E9. How often have you been angered because of things that were outside of your control?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
<tr>
<td>E10. How often have you felt difficulties were piling up so high that you could not overcome them?</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
<td>□</td>
</tr>
</tbody>
</table>
**Instructions.** A number of statements which people have used to describe themselves are given below. Read each statement and then choose the appropriate choice to the right of the statement to indicate how you feel **right now**, that is, **at this moment**. There are no right or wrong answers. Do not spend too much time on any one statement but give the answer which seems to describe your present feelings best.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Never</th>
<th>Sometimes</th>
<th>Often</th>
<th>Almost Always</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>E11.</strong> I feel calm</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E12.</strong> I feel secure</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E13.</strong> I am tense</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E14.</strong> I feel strained</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E15.</strong> I feel at ease</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E16.</strong> I feel upset</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E17.</strong> I am presently worrying over possible misfortunes</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E18.</strong> I feel satisfied</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E19.</strong> I feel frightened</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E20.</strong> I feel comfortable</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E21.</strong> I feel self-confident</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E22.</strong> I feel nervous</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E23.</strong> I am jittery</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E24.</strong> I feel indecisive</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E25.</strong> I am relaxed</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E26.</strong> I feel content</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E27.</strong> I am worried</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E28.</strong> I feel confused</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E29.</strong> I feel steady</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E30.</strong> I am pleasant</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Instructions.** A number of statements which people have used to describe themselves are given below. Read each statement and then choose the choice to the right of the statement that indicates **how you generally feel**. There is no right or wrong answer. Do not spend too much time on any one statement but give the answer which seems to describe how you generally feel.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Never</th>
<th>Sometimes</th>
<th>Often</th>
<th>Almost Always</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>E31.</strong> I feel pleasant</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E32.</strong> I feel nervous and restless</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>E33.</strong> I feel satisfied with myself</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
E34. I wish I could be as happy as others seem to be
E35. I feel like a failure
E36. I feel rested
E37. I am calm, cool, and collected
E38. I feel that difficulties are piling up so that I cannot overcome them
E39. I worry too much over something that really doesn’t matter
E40. I am happy
E41. I have disturbing thoughts
E42. I lack self-confidence
E43. I feel secure
E44. I make decisions easily
E45. I feel inadequate
E46. I am content
E47. Some unimportant thoughts run through my mind and bothers me
E48. I take disappointments so keenly that I can’t put them out of my mind
E49. I am a steady person
E50. I get in a state of tension or turmoil as I think over my recent concerns and interests

Contact for Compensation & Objective 1 Interview

Name: _______________________

Email address for Amazon E-gift Card: _______________________

Participants can follow the following link to the study Facebook site and click “Book Now” for Objective 1 Interview, or email kate.dibble@uconn.edu to schedule.

https://www.facebook.com/uconnBRCA12genetictestingstudy/

This is the end of the survey. THANK YOU for taking the time to fill out this survey.
Appendix E

Objective 1 current study interview script and outline

UCONN GENETIC COUNSELING INTERVIEW SCRIPT

INTRODUCTION

Thank you for agreeing to assist us with this project, as your input is very important. This interview should take up to 30 minutes to complete. If you do not have that time now, we can easily reschedule for another date.

Before we begin, I’d like to review the informational sheet (*see Informational Sheet document) to go over a little information regarding the study before we begin.

*After reading Informational Sheet to participant*

As part of this research study, the University of Connecticut may record your likeness and/or voice through a medium, such as audio or visual recordings, including but not limited to video, audio, photographic, digital, and electronic mediums during your participation in this interview. This choice is completely up to you. Your name, or any other identifying information, will not be present in these recordings. The photo/videos will not be used for commercial purposes and will only be accessed by the research team. You are also given the option of signing a physical consent form instead of verbal consent, completed via email. Please let me know if this is preferred.

Can I ask for your verbal consent that you understand all of the risks and benefits of this study, as well as the release of video recordings, and understand what is being asked of you as a participant?

*IF USING TELEPHONE INTERVIEW ➔ Can I ask your permission to record the audio conversations from this telephone interview for research purposes only?

*IF USING SKYPE INTERVIEW ➔ Can I ask your permission to record the video conversations from this Skype interview for research purposes only?

Thank you. We can now begin the interview. If any question makes you feel uncomfortable in any way, we can skip it. Please let me know if any uneasy feelings arise stemming from interview questions.

INTERVIEW QUESTIONS

1. You have undergone genetic hereditary breast/ovarian cancer counseling and tested positive for either BRCA1 and/or BRCA2 genetic mutations. How long ago were you tested?

2. How did you decide to be tested? For example, did your primary care physician or gynecologist refer you to genetic counseling, or was it on your own accord?
3. Did you feel as though this was the right choice, or did another aspect such as family or future childbearing influence the decision?
   a. If you began testing using direct-to-consumer (DTC) methods such as 23andMe or Ancestry.com, you received formal genetic counseling afterwards. How did you feel about going into formal testing?

4. If you have any biological children, how many do you have? Are they female or male?

5. Do you know if they are BRCA-positive or carriers as well?
   a. If they are under the age of 18, do you think they will get tested when they are old enough?
   b. Are you nervous about telling them about your predisposition?

6. Were your test results explained to you so that you can understand them fully?

7. If you were dissatisfied on how your results were given to you, how would you change them?

8. Right now, there are two main types of genetic counseling. Individualized counseling occurs when you, the patient, undergo physical genetic testing and results yourself. Family-based counseling involves at-risk family members undergoing the process together, including the testing and results. Which form of genetic counseling did you undergo?
   a. Based on your experiences, which form of genetic counseling would you have preferred?
   b. Why do you think you would have preferred this type of counseling?

9. Are you happy with how your experience of genetic counseling went? What were some positive aspects of counseling?

10. Was there anything about your experience of genetic counseling that was not positive? What were some of the negative aspects of counseling?

11. When it was time to be given your test results, how were they presented to you? For instance, some individuals get results over the phone, in an email, or in a separate counseling appointment in-person.

12. Were you satisfied with how your test results were given to you? If not, how would you want your test results presented?

13. How did you feel when your test results were given to you? What were your emotions at that time?

14. How have your emotions changed since then?

15. Were you given guidance on next steps or future preventative measures to help decrease your chances of developing breast or ovarian cancers?

16. If you have undergone any preventative measures, what were they and how did you feel about them?
17. Since it has been some time since you were given your genetic results, what was your long-term reaction to this information?

18. If you could change the genetic counseling process, how would you change it?

19. Are there any other comments or concerns that you would like to bring up about your experiences or preferences undergoing genetic counseling?

**CONCLUSION**

Before we wrap up our interview, are there any last-minute comments you have regarding what I asked you before?

As I mentioned earlier, we are planning to speak with up to 40 individuals across the nation who have tested positive for *BRCA1* and/or *BRCA2*. Our goal is to gain inside knowledge on genetic counseling experience, preferences, and outcomes when a positive result is given. Thank you for your participation. Please do not hesitate to call or email if you have any concerns or additional questions.