

5-2017

Assessing Women's attitudes Towards Genetic Testing For Hereditary Breast Cancer

Taylor Jane Apostolico
University of South Carolina

Follow this and additional works at: <https://scholarcommons.sc.edu/etd>



Part of the [Genetics Commons](#)

Recommended Citation

Apostolico, T. J.(2017). *Assessing Women's attitudes Towards Genetic Testing For Hereditary Breast Cancer*. (Master's thesis). Retrieved from <https://scholarcommons.sc.edu/etd/4047>

This Open Access Thesis is brought to you by Scholar Commons. It has been accepted for inclusion in Theses and Dissertations by an authorized administrator of Scholar Commons. For more information, please contact digres@mailbox.sc.edu.

ASSESSING WOMEN'S ATTITUDES TOWARDS GENETIC TESTING FOR
HEREDITARY BREAST CANCER

by

Taylor Jane Apostolico

Bachelor of Science
Cornell University, 2015

Submitted in Partial Fulfillment of the Requirements

For the Degree of Master of Science in

Genetic Counseling

School of Medicine

University of South Carolina

2017

Accepted by:

Crystal R. Hill-Chapman, Director of Thesis

Sue P. Heiney, Reader

Ronald T. Murphy, Reader

Cheryl L. Addy, Vice Provost and Dean of the Graduate School

© Copyright by Taylor Jane Apostolico, 2017
All Rights Reserved

ACKNOWLEDGMENTS

Eternal thanks are owed to my family, especially my parents, Joe and Eileen, and my sister, Kaleigh, for their unconditional support, encouragement, and good humor throughout the more challenging points of this journey.

I would like to thank my thesis committee for their time, expertise, and contributions to this study. I am grateful to Dr. Crystal Hill-Chapman for helping develop this project and dedicating so much effort into making it feasible. This study would not have been possible without Dr. Sue Heiney, who's input was essential in moving forward with research design and participant recruitment, and Dr. Ronald Murphy, who made it possible to connect with the amazing men and women who staff Florence's Homeless Connection.

I am sincerely grateful to all those who have contributed to the foundation, implementation, and completion of this project, as well as to my training while at the University of South Carolina. Thank you to all of my instructors and mentors throughout this program who have provided me with both personal and professional standards to aspire to reach.

Last but certainly not least, to my classmates – thank you for the supportive shoulders, the open ears, and the countless laughs. I am privileged to have had the opportunity to call such an inspirational, compassionate, and motivated group of women my friends and peers for these last two years and cannot wait to see you grow and thrive as you begin your careers.

ABSTRACT

OBJECTIVES: Hereditary Breast and Ovarian Cancer (HBOC) is an autosomal dominant cancer predisposition syndrome with a 46-87% lifetime risk of breast cancer. Unaffected women who have HBOC are eligible for more screening procedures and prophylactic surgeries that may reduce the risk of developing cancer by up to 95%. The objectives of this study were to assess women's awareness of and interest in breast cancer genetic testing services, as well as women's attitudes and beliefs regarding the clinical utilization of HBOC genetic testing across demographic categories. **METHODS:** Two-hundred and sixty-eight women completed a 35-item survey designed to capture perceptions of HBOC and genetic testing, attitudes towards genetic testing for HBOC, and demographics.

RESULTS: Two-hundred and eight women met participation criteria. One-hundred and fifty-five (75%) indicated prior awareness of genetic testing services. One-hundred and forty-three (69%) indicated interest in genetic testing for HBOC. Black women, women with lower levels of education, and women with lower household incomes reported less awareness, but similar levels of interest in genetic testing services when compared to other participants. **CONCLUSIONS:** Women are interested in genetic testing for HBOC. Specific counseling on barriers to minority and low socioeconomic communities, such as cost and education regarding genetic testing procedures, may be beneficial in increasing utilization of genetic testing for HBOC in these communities. Women who experience more discrimination may have increased perceptions of their susceptibility to hereditary cancer syndromes and may benefit from personalized risk counseling.

TABLE OF CONTENTS

ACKNOWLEDGEMENTS	iii
ABSTRACT	iv
LIST OF TABLES	vii
LIST OF FIGURES	viii
CHAPTER I. BACKGROUND	1
1.1 Hereditary Breast Cancer	1
1.2 Hereditary Breast Cancer Screening	3
1.3 Genetic Testing for Hereditary Breast Cancer	4
1.4 Health Belief Model	11
1.5 Rationale	13
1.6 Purpose	16
CHAPTER II. ASSESSING WOMEN’S ATTITUDES TOWARDS GENETIC TESTING FOR HEREDITARY BREAST CANCER	17
2.1 Abstract	18
2.2 Introduction	19
2.3 Materials and Methods	20
2.4 Results	24
2.5 Discussion	40
2.6 Limitations and Future Studies	50
2.7 Summary	52

CHAPTER III. CONCLUSIONS	55
REFERENCES	56
APPENDIX A. Participant Resources	65
APPENDIX B. Invitation to Participants	66
APPENDIX C. Letter of Support.....	67
APPENDIX D. Background Information	68
APPENDIX E. Questionnaire	69

LIST OF TABLES

Table 2.1 Participant demographic information	26
Table 2.2 Demographic comparison of aware versus unaware participants	27
Table 2.3 Demographic comparison of interested versus uninterested participants	30
Table 2.4 Health Belief Model and comparison of groups	31
Table 2.5 Correlations between perceived discrimination, perceived susceptibility, and interest in genetic testing	38

LIST OF FIGURES

Figure 1.1 The Health Belief Model	12
Figure 2.1 Agreement with benefits of genetic testing	32
Figure 2.2 Agreement with barriers of genetic testing	32
Figure 2.3. Race vs. agreement with benefits	35
Figure 2.4 Race vs. agreement with barriers.....	35
Figure 2.5 Mediating role of perceived susceptibility in the relationship between perceived discrimination and interest in genetic testing	39

CHAPTER I. BACKGROUND

Hereditary breast and ovarian cancer (HBOC) is an autosomal dominant cancer predisposition syndrome caused by germline mutations in the breast cancer 1 (*BRCA1*) and breast cancer 2 (*BRCA2*) tumor suppressor genes. Individuals who have a pathogenic gene mutation in *BRCA1* or *BRCA2* have a 46-87% life time risk of developing breast cancer, compared to a general population risk of 12% (National Comprehensive Cancer Network, 2016). A prophylactic bilateral mastectomy in mutation carriers has been found to reduce the risk of breast cancer by at least 90% (Hartmann et al., 1999). An estimated 5-10% of all breast cancer cases are attributable to an inherited mutation (National Cancer Institute, 2015).

1.1 Hereditary Breast Cancer

Breast cancer is a cancer that develops from breast tissue. In the general population, one in eight women will develop breast cancer at some point in their lives, making it the second most common cancer in women (National Cancer Institute, 2014). The incidence of breast cancer has risen over the past 30 years, most frequently affecting white and black women (National Cancer Institute, 2013). Despite the high prevalence of diagnoses of breast cancer in white women, they are statistically the least likely to die once diagnosed with breast cancer (National Cancer Institute, 2013). Black women are more likely to be diagnosed at a later stage of breast cancer, and to die due to breast cancer (Siegal, 2015). Besides ethnicity, Some of the risk factors for developing breast cancer

include older age, higher breast density, a history of radiation therapy, a family or personal history of breast cancer, and genetic changes that predispose individuals to the development of breast cancer (National Cancer Institute, 2015).

Five to ten percent of all breast cancer cases are classified as ‘hereditary’, meaning that they are caused by an inherited genetic mutation that raises the risk of cancer development by ceasing or altering the function of cancer-preventing proteins (National Cancer Institute, 2016). The two most common genetic causes of breast cancer are the *BRCA1* and *BRCA2* genes, which are implicated in 20-25% of hereditary breast cancer cases (Pharoah et al., 2002). The prevalence of *BRCA* carriers in the general population is estimated to be 1 in 200-400. However, this estimate is based on data from breast cancer patient populations and may not reflect the true carrier frequency in the general population (Metcalfe et al., 2015).

When functioning properly, the *BRCA* genes work to produce proteins that repair damage to DNA and regulate transcription. When there are mutations in a *BRCA* gene, it results in the formation of a truncated protein that is unable to regulate cell growth, leading to tumorigenesis (Welsh & King, 2001). Mutations in the *BRCA* genes are associated with an estimated 46-87% lifetime risk of developing breast cancer, markedly higher than the 12% risk an average woman has of developing breast cancer (National Comprehensive Cancer Network, 2016). Currently, researchers have identified a network of over nine genes, including *BRCA1* and *BRCA2*, that are each implicated in raising the risk of developing breast cancer to at greater than 20% (National Comprehensive Cancer Network, 2016).

The vast majority of genetic changes implicated in hereditary breast cancer are inherited in an autosomal dominant pattern, meaning only one mutated copy of the two genes is required to cause a person to have an increased risk of cancer. However, tumorigenesis only occurs when both copies of the gene are pathogenically mutated. This happens naturally, or somatically, as a person ages either through errors in DNA replication or through environmental exposures such as tobacco smoke or UV light. In persons with no inherited change, both genes will have been somatically mutated throughout their life to cause cancer development. In persons with an inherited mutation in a cancer-susceptibility gene, only one detrimental environmental event that pathogenically harms the DNA of the un-mutated copy of the gene permanently is needed to cause the onset of tumorigenesis, a phenomenon termed the ‘Knudson Two-Hit Hypothesis’ (Anderson, 1992). Consequently, patients with an inherited mutation in a cancer-susceptibility gene tend to develop cancer at a younger age and at a greater frequency than those with no inherited mutation (Burke et al., 1997).

1.2 Hereditary Breast Cancer Screening

Women who are determined to have a high risk of developing breast cancer, either through a known genetic mutation or an extensive family history, are eligible for and encouraged to seek high-risk screening procedures to decrease this risk (National Comprehensive Cancer Network, 2016). These procedures involve earlier and more frequent utilization of physical examinations and imaging techniques (National Comprehensive Cancer Network, 2016). These screening techniques have been shown to detect a larger number of cases of breast cancer at an earlier stage of the disease

(DeSantis et al., 2013; Berg et al., 2012). Women who are considered to have a high risk of developing breast cancer may also elect to have risk reducing procedures such as a prophylactic mastectomy, which has been shown to reduce the risk of breast cancer by as much as 90-95% (Rebbeck et al., 2004). Compared to other cancers, breast cancer has a low mortality rate, as approximately 89.7% of women who develop breast cancer will still be alive five years after their initial diagnosis (National Cancer Institute, 2014).

1.3 Genetic Testing for Hereditary Breast Cancer

The *BRCA1* gene was first sequenced in 1994 in a collaborative effort by Myriad, a genetic researching company, the University of Utah, the National Institute of Health, and McGill University (Williams-Jones, 2002). Shortly after, Myriad filed for ‘composition-of-matter’ and ‘methods-of-use’ patents on the *BRCA1* gene, preventing other genetic researching institutions from sequencing the *BRCA1* gene for commercial use (Williams-Jones, 2002). The sequencing of *BRCA2* quickly followed and once again, Myriad was among the first to file a U.S. patent on the gene (Williams-Jones, 2002). Subsequently, Myriad gained control over the commercial market for genetic testing for all of the known hereditary breast cancer genes at the time.

The first commercial genetic test for hereditary breast cancer was *BRACAnalysis*, produced and marketed by Myriad in 1996 (Williams-Jones, 2002). Myriad used their control on the *BRCA* testing market to merge their testing service into multiple facets of health care and cancer research, partnering with various health insurance companies and institutions to allow the test to be covered by health insurance providers and accessible for research, at a price (Williams-Jones, 2002).

There was initial backlash against the commercialization of *BRCA* sequencing, with the opposition fearing that the test was less focused on the prevention of breast cancer and more focused on increasing patient anxiety and the creation of demand for the product (Williams-Jones, 2002). Furthermore, Myriad's patent on *BRCA* testing created barriers to clinical care and delays in research, leading to less effective and efficient testing at increased cost, ultimately creating wider health disparity gaps (Offit et al., 2013). Myriad's patent was eventually overturned by the Supreme Court in 2013, opening the commercial market for hereditary cancer testing and promoting greater accessibility to these tests.

Genetic testing for a hereditary breast cancer mutation can be beneficial to the patient in multiple ways. If a patient has already been diagnosed with breast cancer, genetic testing can help inform her treatment. A positive result for a mutation in a cancer susceptibility gene may encourage her to opt for a double-mastectomy to reduce her likelihood of developing a secondary cancer, as opposed to a cancer-removing lumpectomy, which would leave behind more breast tissue and increase the risk of recurrence (Wevers, 2014). In any patient with a family history of breast cancer, personally affected or not, knowledge of whether or not there is a hereditary mutation in the family can alter reproductive planning and help inform other family members if they are at risk as well (Donnelly et al., 2013). Unaffected patients who test positive for a mutation that increases their risk of developing breast cancer are eligible to begin following high-risk screening regimens to promote early detection of cancer if it does develop, and may begin to consider risk-reducing surgeries, like prophylactic mastectomies, to decrease their chance of ever having cancer (Scheuer et al., 2002).

Receiving genetic testing, whether the result is positive or negative, has been shown to increase utilization of mammography and breast self-exams after result disclosure (Botkin et al., 2003). Furthermore, women who know they have a mutation in the *BRCA2* gene are eligible to begin taking Tamoxifen, a chemoprevention drug that reduces the risk of breast cancer in women with *BRCA2* mutations by approximately 62% (King et al., 2001).

1.3.1 Patient Preferences

The impact of receiving a positive result for a *BRCA* mutation has been shown to have variable psychological impacts. Studies have suggested that a positive genetic testing result for a hereditary cancer syndrome initially leads to an increase in both general and cancer-specific anxiety for women with *BRCA1* or *BRCA2* mutations, but that this distress tends to subside significantly with time (Beran et al., 2008; Wilder-Smith et al., 2007). Women who report more intense emotional representations of hereditary cancer and difficulty understanding hereditary cancer are more likely to experience distress regarding hereditary cancer testing (Voorwinden & Jaspers, 2015).

Women cite many reasons as potential influences to sway them from pursuing genetic testing. Fear of discrimination or stigmatization by employers and health insurance companies by use of genetic information has historically been an influence on the decision to pursue genetic testing (Apse et al., 2004; Durfy et al., 1999). Women who perceive themselves to have a lower risk of developing cancer are less likely to accept an offer of genetic testing (Culver et al., 2001).

Women's decisions to undergo genetic testing for a hereditary cancer syndrome are complicated, with many factors influencing their reasoning for whether or not they

should pursue testing. Women who have children or a mother with a cancer history are more likely to choose to have testing sooner than those who do not (Hesse-Biber & An, 2016). Women also tend to factor in social factors when making decisions concerning their genetic testing and management (Hesse-Biber & An, 2016). Women are more likely to postpone genetic testing if they perceive themselves to have strong social supports (Hesse-Biber & An, 2016). For women who are not yet affected, the decision to pursue genetic testing for an adult-onset disease such as hereditary breast cancer is hypothesized to partially be a coping mechanism to combat the uncertainty of living at risk of developing a disease at some point in the future and to obtain control over an otherwise minimally controllable situation (Gooding et al., 2005)

1.3.2 Patient Demographics

Education has been found to be a predictor of women's awareness of and interest in genetic testing for hereditary cancer syndromes. Women who obtain higher levels of education are more likely to report awareness of genetic testing services for cancer (Mai et al., 2014; Tambor, Rimer, & Strigo, 1998). Reported interest in pursuing genetic counseling and testing services is also associated with higher levels of education (Culver et al., 2001; Lerman et al., 1994).

There are significant differences in the ethnic composition of women who choose to pursue genetic testing for cancer. Ethnic minorities are less likely to be aware of genetic testing services and less likely to refer themselves to genetic counseling (Mai et al., 2014; Glenn, Chawla, & Bastani, 2012; Hutson, 2003). While it is hypothesized that the mutation prevalence of the *BRCA* genes is similar across ethnicities, women of European ancestry make up the majority of consumers of genetic testing services, while

women of African, Asian, and Latina ancestry comprise a smaller portion, even when their numbers are combined (Hall et al., 2009; Susswein et al., 2008). Minority women tend to face more socioeconomic barriers to accessing genetic services, such as time limitations, difficulty accessing providers and referrals, geographic barriers, and language barriers (Forman & Hall, 2009; Thompson et al., 2003). Ethnicity and race also play a role on women's attitudes and perspectives on cancer genetic testing. Minority populations typically have greater levels of distrust in the medical community. Latina and African American women have previously been shown to be more concerned about testing abuses of genetic information when compared to Caucasian women (Glenn, Chawla, & Bastani, 2012; Thompson et al., 2003).

Income and insurance coverage both play a role in determining how likely a woman is to pursue genetic testing. Women who have higher income and utilize some form of health insurance are more likely to have had genetic testing (Lerman et al., 1996). Women who have lower incomes are more likely to agree with perceived disadvantages of genetic testing, such as fear of confidentiality, concern about the impact the result may have on the family, and a fear of being singled out, among others (Thompson et al., 2003). Recent research has suggested that women in low income communities are interested in receiving genetic counseling and testing, but face challenges related to access (Komenaka et al., 2015).

Whether a person lives near an urban community or within a rural community predicts their awareness of and access to genetic testing services. Physicians in rural practices are less likely to refer for hereditary breast cancer than physicians practicing in

urban or suburban locations, due to distance, lack of awareness, lack of effective cancer risk reduction, and lack of patient interest (Koil et al., 2003).

1.3.3 Genetic Testing: Climate Changes Impacting Awareness and Interest

The Genetic Information Nondiscrimination Act (GINA) was approved in 2008. The main purpose of GINA was to prevent insurance companies and employers from discriminating based on genetic testing results (Feldman, 2012). Violations of GINA are punishable by monetary penalty. (Erwin, 2008). Misuse of genetic testing in the 1970s to discriminate against African Americans with sickle cell disease in the workplace and through insurance coverage fostered a mistrust of the clinical use of genetic information, and still remains a serious concern of many citizens today (Feldman, 2012). GINA was developed to encourage patients to receive genetic testing when it is deemed to be a helpful investigative tool to manage their health care, without fear of this information being used against them in the future. While knowledge about GINA is associated with a reduction in concern regarding health insurance discrimination, consumer knowledge about GINA is still limited (Allain, Friedman, & Senter, 2012).

The Patient Protection and Affordable Care Act (ACA) became law in 2010, and was fully implemented on January 1, 2014. One of the main purposes of the ACA was to guarantee access to universal, affordable, health insurance coverage. In 2007, an estimated 56 million American citizens were considered “medically disenfranchised”, meaning that their most basic health and wellness needs were not being met (National Association of Community Health Centers, 2007). The ACA is projected to double the number of patients who are medically served (Rosenbaum, 2011). Furthermore, the ACA

allows for the reduction of the cost and the provision of Medicaid coverage for clinical preventative services, including advanced screening and genetic counseling and testing for women who are at high risk of developing breast cancer. (Koh & Sebelius, 2010). However, the ACA does not provide coverage for all screening techniques recommended, even for women with a known *BRCA1/2* mutation, raising concerns that the coverage of genetic testing alone would have limited benefit in vulnerable populations (Walcott et al., 2014).

In 2013, awareness of genetic testing, for the *BRCA* genes specifically, increased dramatically due to media coverage of celebrity Angelina Jolie's essay sharing her experiences with genetic testing and her subsequent decision to have a prophylactic mastectomy (Evans et al., 2014). The aftermath of this event led to global increases in referrals to genetic testing services and in requests for *BRCA1/2* testing (Evans et al., 2014). Women who identified with Jolie in some way were found to be especially motivated to pursue genetic testing (Kosenko, Binder, & Hurley, 2016). In general, research suggests that celebrity experience and endorsement raises positive public perception of the subject and increases information seeking behaviors (Hoffman & Tan, 2013).

Studies have suggested that up to 50% of female *BRCA* mutation carriers have no significant family history of suggestive of HBOC (Moller et al., 2007). Based on current guidelines, these females would not be considered suitable candidates for genetic testing until they themselves had an early cancer diagnosis, removing the option of prophylactic action. Population screening for *BRCA1/2* has been proposed as a possible alternative to capture this population (Gabai-Kapara et al., 2014). Population screening has been

shown to be more cost-effective than family-history based approaches, but will still prove to be a costly and time-consuming process in a multi-million-person population such as the United States (Manchanda et al., 2015; Clain et al., 2015). General population screening of *BRCA1* and *BRCA2* may be a cost effective option to identify carriers, and seems to be a possibility within our grasp as small-scale implementations are already in effect (Wang, 2016).

1.4 Health Belief Model

The Health Belief Model (HBM) was created out of psychological and behavioral theory in 1952 as a measure to assess factors impacting the utilization of preventative health services, such as screening and immunizations (Rosenstock, 1974). The original HBM consisted of four key dimensions – perceived susceptibility of developing a condition, perceived severity of the condition, perceived benefits of preventative action and perceived barriers to preventative action (Janz & Becker, 1984). The interactions of each component are summarized in Figure 1.

Perceived susceptibility encompasses a range of perceptions of risk from complete denial to imminent risk (Strecher & Rosenstock, 1997). Some individuals without a family history of breast cancer may not feel that they are susceptible to cancer themselves. Other individuals with a family history of breast cancer may feel that they are highly likely to develop breast cancer themselves. Perceived severity is defined as an individual's assessment of the impact a diagnosis or condition may have on their lifestyle and emotional being (Strecher & Rosenstock, 1997). Perceived benefits, or the positive aspects of particular actions, may include physical and psychosocial gains and may

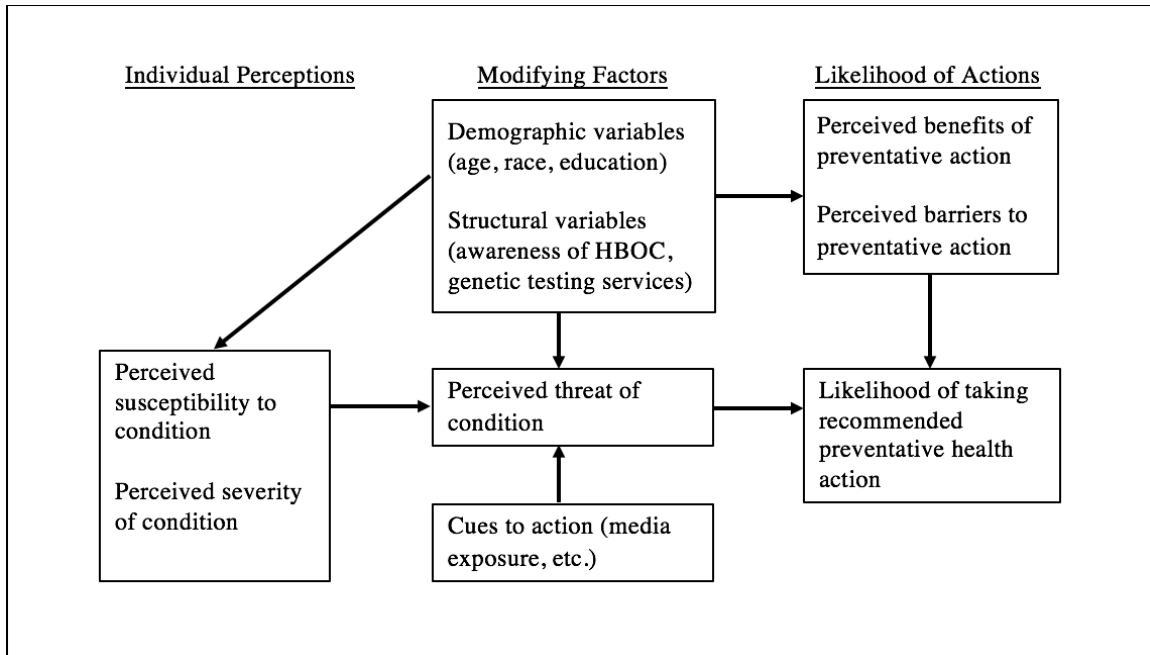


Figure 1.1 The Health Belief Model (Becker, 1974)

influence actions related to management (Strecher & Rosenstock, 1997). Lastly, perceived barriers, or the negative aspects of an action, may include financial costs, inconvenience, or pain incurred by the action and also may influence actions (Strecher & Rosenstock, 1997). Previous studies utilizing the HBM to examine women's attitudes and perceptions towards *BRCA1/2* testing have shown a relationship between perceived susceptibility, perceived severity, and concerns about being a mutation carrier with testing decisions (Wang et al., 2007). Women who tend to be more concerned about being mutation carriers, and report high perceived susceptibility and low perceived severity were found to be more likely to undergo genetic testing (Wang et al., 2007). Women who perceived the benefits of testing to be high were more likely to pursue genetic testing than women who perceived the benefits of testing to be low (Wang et al., 2007).

In the HBM, perceived susceptibility and severity are thought to motivate an individual to action. The reduction of perceived barriers is thought to provide easier accessibility for an action. If an individual believes they are at high risk for a health issue, such as developing breast cancer, an action to reduce risk is not likely to take place unless the individual believes there are substantial benefits associated with their efforts (Strecher & Rosenstock, 1997). Therefore, expressed interest in utilizing preventative services, such as *BRCA1/2* genetic testing may be related to whether or not the perceived benefits of this action outweigh the perceived risks.

Along with the original four items of the Health Belief Model, perceptions of self-efficacy, or belief in one's ability to successfully navigate tasks and challenges, have been proposed as an independent variable to partially explain interest in preventative health behavior (Rosenstock et al., 1988). Individuals must not only feel that the benefits of preventative action would outweigh the costs, but also feel that they are competent to overcome perceived barriers to take action.

1.5 Rationale

The first objective of this study is to identify factors that predict women's interest in genetic testing for hereditary breast cancer syndromes. Previous literature suggests that women thought to be at risk for breast cancer based on their family history may benefit from pursuing genetic testing. Genetic counseling and subsequent testing for cancer-predisposing mutations is linked to increased screening behaviors, which help early diagnosis and management of cancer if it does develop (Botkin et al., 2003). Genetic testing may promote patient empowerment as some women report feeling more control

over their health after deciding to pursue genetic testing (Gooding et al., 2005). Many studies following the initial rise in commercialized genetic testing suggested that education and perceived risk of developing breast cancer were the biggest predictors in women's reported interest in genetic testing services, despite ethnicity and income. African American women and Hispanic women of low socioeconomic status have reported high interest in learning more about cancer genetic testing (Komenka et al., 2015).

However, literature has also shown that these services are primarily utilized by women who have had more education, have greater annual household incomes, have some form of health insurance, and are Caucasian (Lerman et al., 1996). Currently, services are reported to be underutilized by ethnic or racial minorities and women with lower socioeconomic statuses, despite the fact that hereditary breast cancer is no more or less likely to affect these populations, and is more likely to be the cause of death if it does develop either due to later diagnoses or more aggressive manifestations. The second goal of this study will be to attempt to clarify whether or not services are underutilized by these populations because they are unaware of these services or if the perceived utility of genetic testing in these populations is not high enough to outweigh concerns related to accessibility and psychological impact, or possibly a combination of the two.

The third goal of this study is to compare historical factors that predict interest in genetic testing to those that are expressed in this study. Since the first utilization of breast cancer genetic testing, many aspects of this process have changed. Legislation has now made genetic testing more affordable by making it possible to be covered by public insurance. Genetic information is now protected by law, potentially alleviating fears of

discrimination and stigma. Celebrity use and publication of experiences with genetic testing have increased awareness of services and aided in normalizing the process. These factors together suggest that there may be differences in the way the general population of women now perceives genetic testing than they might have in its early days. The healthcare barriers and perceived disadvantages that women feel concerning genetic testing in 2016 may be very different than those that were identified in the early 2000s.

Despite all that has changed since *BRCA* testing was first commercialized in 1996 to make this service more accessible to underserved populations, research suggests that vulnerable and historically discriminated against populations still are not utilizing genetic testing services at as great a frequency (Glenn, Chawla & Bastani, 2012). However, in previous studies, women in these communities have expressed interest in these services. This raises a question of what factors are specifically preventing women from seeking genetic testing. Therefore, we hypothesize that while participants of all backgrounds will express similar levels of interest in genetic testing services, women from underserved populations will be more likely to cite concerns regarding accessibility and utility, and less likely to agree with the proposed benefits of genetic testing.

1.51 Potential Risks

A potential concern of this study is that presenting the risk of breast cancer may raise individual's anxiety concerning their chance of having a hereditary cancer syndrome. To address this, participants were provided with a fact sheet containing instructions for contacting a local genetic counselor if they find themselves experiencing increased anxiety about their personal risk of having a hereditary cancer syndrome (Appendix A). Information gained from this study may not benefit the participants

directly. However, the results of the study may benefit current and future patients with hereditary breast cancer syndromes. Information gleaned from participants' responses will aide in developing educational tools and outreach strategies to better include underserved populations.

1.6 Purpose

The current study explores the attitudes and beliefs of women across demographic characteristics concerning genetic testing for hereditary breast cancer. The first goal of this study is to identify factors that predict women's interest in genetic testing for hereditary breast cancer. The second goal of this study is to clarify women's perceptions of the personal and clinical utility of genetic testing for hereditary breast cancer. The third goal of this study is to compare factors that predict interest in genetic testing for hereditary breast cancer in this population against factors that have predicted interest in previous studies. This study aims to provide insight into women's perceived utility and knowledge of genetic testing services. It also aims to identify populations that may benefit from further education about hereditary breast cancer genetic testing services, as well as what concepts and concerns educational outreach should address.

CHAPTER II. ASSESSING WOMEN'S ATTITUDES TOWARDS GENETIC TESTING FOR HEREDITARY BREAST CANCER¹

¹Apostolico, T.J., Hill-Chapman, C.R., Heiney, S.P., & Murphy, R.T. To be submitted to *Journal of Community Health*

2.1 Abstract

OBJECTIVES: Hereditary Breast and Ovarian Cancer (HBOC) is an autosomal dominant cancer predisposition syndrome with a 46-87% lifetime risk of breast cancer. Unaffected women who have HBOC are eligible for more screening procedures and prophylactic surgeries that may reduce the risk of developing cancer by up to 95%. The objectives of this study were to assess women's awareness of and interest in breast cancer genetic testing services, as well as women's attitudes and beliefs regarding the clinical utilization of HBOC genetic testing across demographic categories. **METHODS:** Two-hundred and sixty-eight women completed a 35-item survey designed to capture perceptions of HBOC and genetic testing, attitudes towards genetic testing for HBOC, and demographics. **RESULTS:** Two-hundred and eight women met participation criteria. One-hundred and fifty-five (75%) indicated prior awareness of genetic testing services. One-hundred and forty-three (69%) indicated interest in genetic testing for HBOC. Black women, women with lower levels of education, and women with lower household incomes reported less awareness, but similar levels of interest in genetic testing services when compared to other participants. **CONCLUSIONS:** Women are interested in genetic testing for HBOC. Specific counseling on barriers to minority and low socioeconomic communities, such as cost and education regarding genetic testing procedures, may be beneficial in increasing utilization of genetic testing for HBOC in these communities. Women who experience more discrimination may have increased perceptions of their susceptibility to hereditary cancer syndromes and may benefit from personalized risk counseling.

2.2 Introduction

Hereditary Breast and Ovarian Cancer (HBOC) is an autosomal dominant cancer predisposition syndrome caused by germline mutations in the breast cancer 1 (*BRCA1*) and breast cancer 2 (*BRCA2*) tumor suppressor genes. Individuals who have a pathogenic gene mutation in either *BRCA1* or *BRCA2* have a 46-87% life time risk of developing breast cancer, compared to a general population risk of 12% (National Cancer Institute, 2016). A prophylactic bilateral mastectomy in mutation carriers has been found to reduce the risk of breast cancer by at least 90% (Hartmann et al., 2001).

Women who are determined to have a high risk of developing breast cancer, either by known genetic variant or family history, are eligible and encouraged to seek high-risk screening procedures to decrease this risk (National Comprehensive Cancer Network, 2016). These procedures, including earlier and more frequent utilization of physical examinations and imaging techniques, have been shown to detect a larger number of cases of breast cancer at an earlier stage of disease (DeSantis et al., 2013; Berg et al., 2012). Women found to be at higher risk of developing breast cancer are also offered the option of having a risk reducing prophylactic procedure, such as a mastectomy, which may reduce the risk of breast cancer by as much as 90-95% (Rebbeck et al., 2004).

Genetic testing for a hereditary breast cancer mutation has multiple proposed benefits. Asymptomatic patients who learn they have a mutation that increases their risk of developing breast cancer can begin to follow high-risk screening regimens to promote early detection, and may consider risk reducing surgeries (Scheuer et al., 2002). Receiving genetic counseling and testing, whether the result is positive or negative, has

been shown to increase utilization of mammography and breast self-exams post result disclosure (Botkin et al., 2003).

Historically, genetic testing services have been underutilized by individuals who are minorities and individuals who are in lower socioeconomic classes (Glenn, Chawla, & Bastani, 2012; Thompson et al., 2003). Women reporting higher levels of education are more likely to report awareness and interest in genetic testing services (Lerman et al., 1994). However, within the past decade the climate surrounding genetic testing has shifted, potentially increasing not only the general population's awareness of genetic testing services but also their ability to access these services. Our study aimed to determine women's awareness of and interest in breast cancer genetic testing services, as well as women's attitudes and beliefs regarding the potential clinical and personal utilization of genetic testing for HBOC across multiple demographic categories.

2.3 Materials and methods

2.3.1 Design and participants

Women, age 18 or older, who had not yet undergone genetic testing for hereditary breast cancer syndromes in the United States were surveyed. Potential participants were required to meet eligibility criteria in order to proceed with the questionnaire to confirm that only the opinions of the targeted population would be captured by the study. Males were excluded from this study as the cancer risks and medical management recommendations differ significantly between male and female *BRCA1/2* carriers. Women who had already undergone genetic testing for hereditary cancer syndromes were excluded from the study as their opinions concerning benefits and barriers would have

already been informed by the testing and counseling process. Women who were not from the United States were excluded as other countries not only differ in their guidelines and recommendations, but also in their national health insurance programs. Individuals who met exclusionary criteria were disqualified from participating in the questionnaire via the skip logic function of the questionnaire programming software provided by SurveyMonkey.com. Based upon G* power program, we determined that at a statistical significance level of $\alpha=0.05$, we would require 60 participants. The threshold for G* power was exceeded for all statistical analyses.

Responses were collected from November 1st, 2016 to February 8th, 2017. In order to capture a diverse sample, women were invited to participate through three channels: Facebook.com, student classes, and homeless shelters. The invitation to participate can be found in Appendix B. Individuals were invited to participate in the study through the social network, Facebook.com. Students enrolled in Introductory Biology, Introductory Chemistry, Introductory Nursing, and Introductory Psychology at a small public liberal arts college in the southeastern United States were invited to participate in the study in return for extra credit in their coursework. Individuals utilizing services provided by shelters assisting those in need in South Carolina were also invited to participate verbally in return for a \$5 food coupon. Signed letters of support were obtained from directors of any shelter that participated prior to the distribution of the questionnaire (Appendix C). A flyer advertising the study and providing standardized background information on hereditary cancer and genetic counseling was distributed to women at the shelter (Appendix D). Women who expressed interest in completing the questionnaire sat in

private with the principle investigator, who assisted the participants in reading the background information and questionnaire, and documenting responses.

2.3.2 Instrument

An original questionnaire utilizing skip logic was developed through SurveyMonkey.com. The questionnaire (Appendix E) was constructed by the principle investigator and contained thirty-five quantitative items used to measure categorical information from the participants. This measure was reviewed and edited by all members of the committee for face validity. The questionnaire utilized a series of multiple choice questions designed to assess the participant's prior awareness of breast cancer genetic testing services and interest in genetic testing for hereditary breast cancer. One continuous item was utilized to elucidate participants' perceptions of their cancer risk (Gurmenkin Levy et al., 2006). Validated scales were utilized to ascertain participants' perceived self-efficacy (Schwarzer & Jerusalem, 1995), perceptions of daily discrimination (Krieger et al., 2005), and prior general knowledge regarding *BRCA1/2* (Lerman et al., 1996). The questionnaire contained a version of the Health Belief Model (HBM) modified specifically for this questionnaire to assess perceived susceptibility and severity of hereditary breast cancer, and perceived benefits and barriers of preventive genetic testing (Wang et al., 2007; Jacobsen et al., 1997).

Cronbach's alpha scores were calculated for each scale. For the perceived self-efficacy scale, $\alpha = 0.99$. For the daily discrimination scale, $\alpha = 0.71$. For the perceived susceptibility to hereditary breast cancer scale, $\alpha = 0.80$. For the perceived severity of a hereditary breast cancer scale, $\alpha = 0.84$. For the proposed benefits scale, $\alpha = 0.78$, and for the proposed risks and barriers scale, $\alpha = 0.61$. As the Cronbach's alpha calculated

for participants' prior knowledge of hereditary breast cancer was $\alpha < 0.50$, these items were not analyzed as a scale and instead analyzed separately.

Demographic information including age, gender, race, level of education, relationship status, region of residence in the United States, number of children, annual household income, and current method of health insurance was obtained. The survey also contained questions specific to breast cancer, including personal and family histories of breast cancer and genetic testing in order to better characterize whether or not the participant would be considered at high risk of having a hereditary cancer syndrome. The demographic and categorical data provided variables for statistical analysis.

Prior to beginning the questionnaire, all participants were provided with a brief and succinct definition of hereditary breast cancer and genetic counseling (Appendix D). Consent agreement was provided on the first page of the questionnaire in accordance with the protocol approved by the University of South Carolina Institutional Review Board.

2.3.3 Data Analysis

For quantitative data analysis, responses were compared as a whole, as well as divided into groups for further investigation. Data analysis was performed using Statistical Package for the Social Sciences, Version 24. One-way ANOVA tests examined the statistical significance ($p < 0.05$) of associations between demographic factors and levels of interest and awareness of genetic testing services. Spearman and Pearson correlations were utilized to further explain associations between variables such as age and education and awareness of and interest in genetic testing for hereditary breast cancer.

To analyze responses to the HBM, average ratings were calculated for each item regarding perceptions of disease susceptibility and severity, and the benefits and barriers of testing using the Likert scale format (strongly disagree = 1; disagree = 2; agree = 3; strongly agree = 4). Mann-Whitney U tests were used to examine the statistical significance of associations between reported awareness and interest in genetic testing services and items of the HBM. Kruskal-Wallis H tests were used to examine the statistical significance of associations between reported demographic factors and items of the HBM.

2.4 Results

2.4.1 Demographic Information

Of the 268 responses to the four-item section of the questionnaire that determined eligibility, 238 responses met criteria. Ineligible participants included men ($n = 6$), minors ($n = 4$), individuals who had already had genetic testing for *BRCA1/2* ($n = 17$), and individuals who lived outside of the United States ($n = 3$). Of those who met criteria, 30 did not complete more than 80% of the survey and were excluded from data analysis, resulting in a total of 208 eligible responses.

Of the 208 responses, 65% of participants described themselves as non-Hispanic white ($n = 135$) and 26% of participants described themselves as black ($n = 54$). The average age of participants was 34.11 with a range of 18 to 65 years of age and a standard deviation of 14.86. The majority of participants reported receiving a Bachelor's degree or higher ($n = 121$). 64.4% of participants reported utilizing employer-provided health insurance ($n = 134$). The majority of participants were single ($n = 93$), had no children (n

= 119), and lived in the eastern region of the United States ($n = 177$). Participants had a mode annual household income reported between \$125,000 to \$149,999 ($n = 17$).

Demographics of individual respondents are shown in Table 2.1

2.4.2 Awareness of Genetic Testing Services

The majority of participants (74.5%) indicated that prior to this survey they were aware of genetic testing services for hereditary breast cancer. Statistical significance ($p < 0.05$) was noted in differences between average awareness of services within multiple demographic factors, including race, highest level of education, health insurance coverage, and annual household income. A demographic comparison of women who reported awareness and those who did not can be found in Table 2.2.

Black women were statistically significantly less likely to have heard of genetic testing for hereditary breast cancer than non-Hispanic white women ($p = 0.03$). Women who reported utilizing government-provided health insurance were less likely to be aware of services than women who reported utilizing employer provided private insurance ($p < 0.01$). Women who reported an annual household income of less than \$10,000 were less likely to be aware of services than women who reported an annual income of \$200,000 or more ($p = 0.03$).

Age had a weak but statistically significantly relationship to awareness of genetic testing services ($r = 0.23, p < 0.01$). Younger women were less likely to be aware of services than older women. Education also had a moderate but statistically significantly correlation with awareness of testing services. Women who reported obtaining higher levels of education were more likely to be aware of genetic testing services than those who reported lower levels of education ($r = 0.33, p < 0.01$).

Table 2.1 Participant demographic information

<i>Individual Participant</i>	N =	(%)	<i>Individual Participant</i>	N =	(%)
	208			208	
Race			Marital Status		
Asian	10	(4.9)	Married	82	(39.4)
Black American	54	(26.2)	Divorced/Separated	7	(3.4)
Hispanic	7	(3.4)	Cohabiting	26	(12.5)
Non-Hispanic White	135	(65.5)	Single	93	(44.7)
Highest Education			No. of children		
Less than high school	2	(1.0)	0 children	119	(57.2)
High school/GED	30	(14.4)	1 child	10	(4.8)
Some college	50	(24.0)	2 children	37	(17.8)
Associate's degree	5	(2.4)	3 children	20	(9.6)
Bachelor's degree	76	(36.5)	4 or more children	19	(9.2)
Graduate degree	45	(21.6)	Income		
Age			< \$10,000	20	(9.6)
< 20 years	28	(13.5)	\$10,000 - \$24,999	11	(5.3)
20 – 24 years	69	(33.2)	\$25,000 - \$49,999	11	(5.3)
25 – 29 years	17	(8.2)	\$50,000 - \$74,999	23	(11.1)
30 – 34 years	4	(1.9)	\$75,000 - \$99,999	19	(9.1)
35 – 39 years	13	(6.3)	\$100,000 - \$124,999	14	(6.7)
40 – 44 years	8	(3.8)	\$125,000 - \$149,999	17	(8.2)
45 – 49 years	13	(6.3)	\$150,000 - \$174,999	7	(3.4)
50 – 54 years	32	(15.4)	\$175,000 - \$199,999	5	(2.4)
55 – 59 years	18	(8.7)	> \$200,000	48	(23.1)
60 – 64 years	5	(2.4)	Prefer not to answer	33	(15.9)
> 65 years	1	(0.5)	Residency		
Insurance			Northeast ²	87	(41.8)
Uninsured	9	(4.3)	Southeast ³	90	(43.3)
University provided	16	(7.7)	Midwest ⁴	6	(2.9)
Government provided ¹	20	(10.6)	Northwest ⁵	1	(0.5)
Self-provided private	28	(13.5)	Southwest ⁶	5	(2.4)
Employer private	134	(64.4)	Did not specify	19	(9.1)

¹Includes Medicaid and Medicare

²Includes ME, VT, NY, NH, MA, RI, CT, NJ, PA, DE, MD, DC

³Includes VA, WV, KY, TN, NC, SC, GA, FL, MS, AL, Puerto Rico

⁴Includes OH, IN, MI, IL, WI, MN

⁵Includes WA, OR, ID, MT, AK, WY, CO

⁶Includes TX, NM, AZ, NV, UT, CA, HI

Table 2.2 Demographic comparison of aware versus unaware individuals

	<i>Aware Individuals</i>		<i>Unaware Individuals</i>		<i>Group Total</i>
	n = 155	(%)	n = 53	(%)	N
Race					
Asian	9	(90.0)	1	(10.0)	10
Black American	32	(59.3)	22	(40.7)	54
Hispanic	6	(85.7)	1	(14.3)	7
Non-Hispanic White	107	(79.3)	28	(20.7)	135
Highest Education					
Less than high school	1	(50.0)	1	(50.0)	2
High school/GED	14	(46.7)	16	(53.3)	30
Some college	32	(64.0)	18	(36.0)	50
Associate's degree	4	(80.0)	1	(20.0)	5
Bachelor's degree	64	(84.2)	12	(15.8)	76
Graduate degree	40	(88.9)	5	(11.1)	45
Age					
< 20 years	15	(53.6)	13	(46.4)	28
20 – 29 years	59	(68.6)	27	(31.4)	86
30 – 39 years	16	(94.1)	1	(5.9)	17
40 – 49 years	16	(76.2)	5	(23.8)	21
50 – 59 years	43	(86.0)	7	(14.0)	50
> 60 years	6	(100.0)	0	(0.0)	6
Income					
< \$10,000	9	(45.0)	11	(55.0)	20
\$10,000 - \$24,999	8	(72.7)	3	(27.3)	11
\$25,000 - \$49,999	8	(72.7)	3	(27.3)	11
\$50,000 - \$74,999	13	(56.5)	10	(43.5)	23
\$75,000 - \$99,999	14	(73.7)	5	(26.3)	19
\$100,000 - \$124,999	13	(92.9)	1	(7.1)	14
\$125,000 - \$149,999	13	(76.5)	4	(23.5)	17
\$150,000 - \$174,999	5	(71.4)	2	(28.6)	7
\$175,000 - \$199,999	4	(80.0)	1	(20.0)	5
> \$200,000	41	(85.4)	7	(14.6)	48
Prefer not to answer	27	(81.8)	6	(18.2)	33
Health Insurance					
Uninsured	6	(66.7)	3	(33.3)	9
University-provided	10	(62.5)	6	(37.5)	16
Government-provided ¹	8	(40.0)	12	(60.0)	20
Self-provided private	20	(71.4)	8	(38.6)	28
Employer-provided private	110	(82.1)	24	(17.9)	134

¹Includes Medicare and Medicaid

2.4.3 Interest in Genetic Testing for Hereditary Breast Cancer

The majority of participants (68.75%) indicated they would be interested in pursuing genetic testing for hereditary breast cancer. One-way ANOVAs showed statistical significance ($p < 0.05$) between reported average interest within multiple demographic factors, including marital status and number of children. Married women were statistically significantly less likely to report interest in testing than women who had never been married ($p < 0.01$). Women who reported having no children were statistically significantly more likely to report interest in genetic testing for hereditary breast cancer than those who reported having 2 or more children ($p < 0.01$). Race did not have a statistically significant association with interest in testing. However, a considerable trend towards significance was noted in that Black women tended to be more likely to report interest in testing than non-Hispanic white women ($p = 0.051$).

Age had a moderate but statistically significant relationship with interest in testing services ($r = -0.32$, $p = 0.01$), with younger women more frequently expressing interest in genetic testing for hereditary breast cancer. Highest level of education was found to have a weak but statistically significant correlation with interest ($r = -0.17$, $p = 0.01$), with women who reported obtaining higher levels of education less frequently reporting interest than other participants. Women's perceptions of their risk of developing breast cancer was not statistically significantly associated with interest in genetic testing.

Women who reported interest in genetic testing were statistically significantly more likely to perceive themselves to be at risk for a genetic mutation that would predispose them to breast cancer ($p < 0.01$). Interested participants were also more likely

to agree or strongly agree with the proposed benefits of genetic testing than those who were not interested ($p < 0.01$). Interested participants reported statistically significantly higher frequencies of day to day discrimination ($p < 0.01$). Prior awareness of testing services was not found to be statistically significantly associated with interest. There was no statistically significant difference between interested participants and uninterested participants concerning perceptions of the severity of a genetic predisposition to cancer, overall perceptions of barriers to genetic testing, perceptions of self-efficacy, and general knowledge of hereditary breast cancer. A comparison of the demographics of interested and uninterested participants can be found in Table 2.3.

2.4.4 Health beliefs

Table 2.4 represents 20 items that were derived from the HBM. The data was generated by the Likert scale rating format to determine individual perceptions of the susceptibility to and the severity of a genetic mutation that would increase their risk of breast cancer, as well as the benefits and barriers of preventative testing for *BRCA1/2* pathogenic variants. A high average is associated with agreement (strongly agree = 4) to a statement, whereas a low average is associated with disagreement (strongly disagree = 1). Items were compared among demographic groups, analyzed, and averaged.

In general, women reported high agreement to the proposed benefits of genetic testing and low agreement to the proposed barriers of genetic testing (Figures 2.1 & 2.2). The most strongly agreed with benefits included learning about their children's risk for breast cancer, informing action regarding dealing with cancer risk, and helping other family members decide whether to test. Women disagreed most strongly with the proposed barriers that their family would not be supportive, testing would not cause them

Table 2.3 Demographic comparison of interested versus uninterested individuals

	<i>Interested Individuals</i>		<i>Uninterested Individuals</i>		<i>Group Total</i>
	n = 143	(%)	n = 65	(%)	N
Race					
Asian	6	(60.0)	4	(40.)	10
Black American	45	(83.3)	9	(16.7)	54
Hispanic	5	(71.4)	2	(28.6)	7
Non-Hispanic White	86	(63.7)	49	(36.3)	135
Highest Education					
Less than high school	2	(100.0)	0	(0.0)	2
High school/GED	21	(70.0)	9	(30.0)	30
Some college	43	(86.0)	7	(14.0)	50
Associate's degree	4	(80.0)	1	(20.0)	5
Bachelor's degree	46	(60.5)	30	(39.5)	76
Graduate degree	27	(60.0)	18	(40.0)	45
Age					
< 20 years	20	(71.4)	8	(28.6)	28
20 – 29 years	73	(84.9)	13	(15.1)	86
30 – 39 years	13	(76.5)	4	(24.5)	17
40 – 49 years	11	(52.4)	10	(47.6)	21
50 – 59 years	23	(46.0)	27	(54.0)	50
> 60 years	3	(50.0)	3	(50.0)	6
Income					
< \$10,000	14	(70.0)	6	(30.0)	20
\$10,000 - \$24,999	8	(72.7)	3	(27.3)	11
\$25,000 - \$49,999	7	(63.7)	4	(36.4)	11
\$50,000 - \$74,999	18	(78.3)	5	(21.7)	23
\$75,000 - \$99,999	15	(78.9)	4	(21.1)	19
\$100,000 - \$124,999	9	(64.3)	5	(35.7)	14
\$125,000 - \$149,999	11	(64.7)	6	(35.3)	17
\$150,000 - \$174,999	6	(85.7)	1	(14.3)	7
\$175,000 - \$199,999	3	(60.0)	2	(40.0)	5
> \$200,000	26	(54.2)	22	(45.8)	48
Prefer not to answer	26	(78.8)	7	(21.2)	33
Health Insurance					
Uninsured	8	(88.9)	1	(11.1)	9
University-provided	11	(68.7)	5	(31.3)	16
Government-provided ¹	14	(70.0)	6	(30.0)	20
Self-provided private	19	(67.9)	9	(32.1)	28
Employer-provided private	90	(67.2)	44	(32.8)	134

¹Includes Medicare and Medicaid

Table 2.4 Health Belief Model and comparison of groups

	All Participants N = 208	Aware Individuals n = 155	Unaware Individuals n = 53	<i>p</i>	Interested Individuals n = 143	Uninterested Individuals n = 65	<i>p</i>
Perceptions of susceptibility	2.0944	2.13635	1.9519		2.2183	1.8047	<0.01
Perceptions of severity	2.8476	2.8538	2.8302		2.8718	2.7949	
Benefits							
Learn about children's risk for breast cancer	3.1990	3.1623	3.3077		3.2676	3.0469	0.02
Help other family members decided whether to test	3.0386	3.0260	3.0755		3.1469	2.7969	<0.01
Ease my mind, regardless of test result	2.5865	2.5290	2.7547		2.6993	2.3385	<0.01
Inform action regarding dealing with cancer risk	3.1463	3.1307	3.1923		3.2746	2.8571	<0.01
Help me reduce uncertainty about the future	2.7902	2.7647	2.8654		2.9149	2.5156	<0.01
Help me make important life decisions	2.5099	2.4901	2.5686		2.6170	2.2623	<0.01
Provide a sense of personal control	2.9565	2.9481	2.9811		3.0563	2.7385	<0.01
Barriers							
May not be able to cope with the result	2.1359	2.1176	2.1887		2.1408	2.1250	
Do not understand what will be done	1.9275	1.8701	2.0943		2.0350	1.6875	<0.01
Would not do anything different to manage cancer risk	1.8019	1.8247	1.7358		1.7203	1.9844	<0.01
Would have a negative impact on family	1.8641	1.8824	1.8113		1.8028	2.0000	0.03
Cannot afford the cost	2.4975	2.4342	2.6863		2.5929	2.2857	0.02
Family would not be supportive	1.5700	1.5649	1.5849		1.5664	1.5781	
Testing may lead to discrimination	1.9712	2.0129	1.8491		1.9371	2.0462	
Testing would not provide new information	1.8317	1.8710	1.7170		1.7622	1.9846	0.03

Statistically significant differences between groups appear in bold type, $p < 0.05$

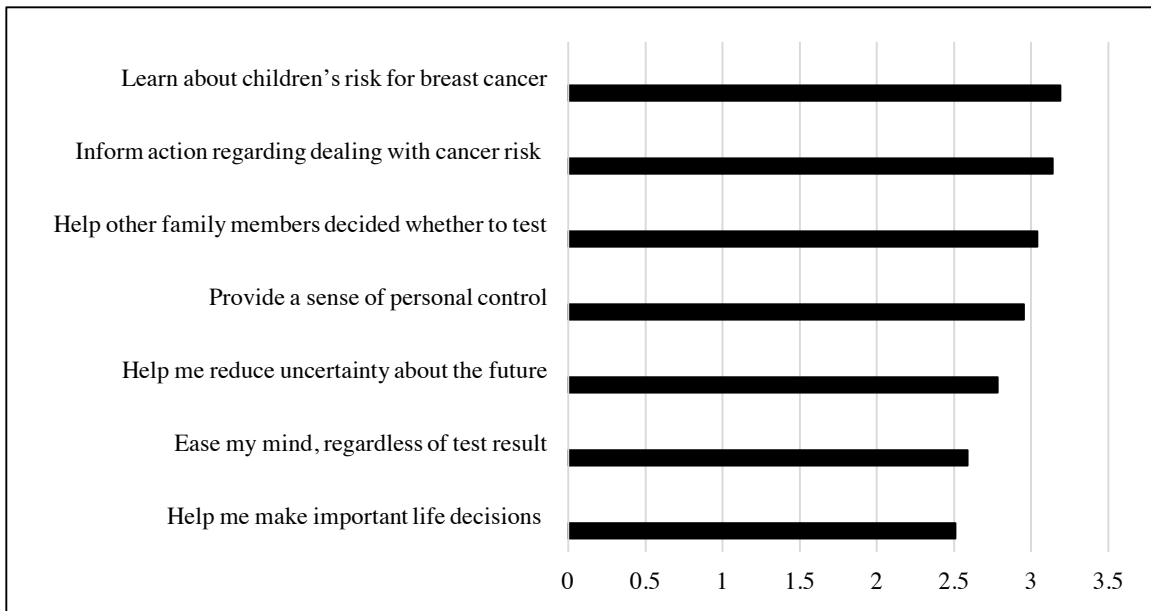


Figure 2.1 Agreement with benefits of genetic testing

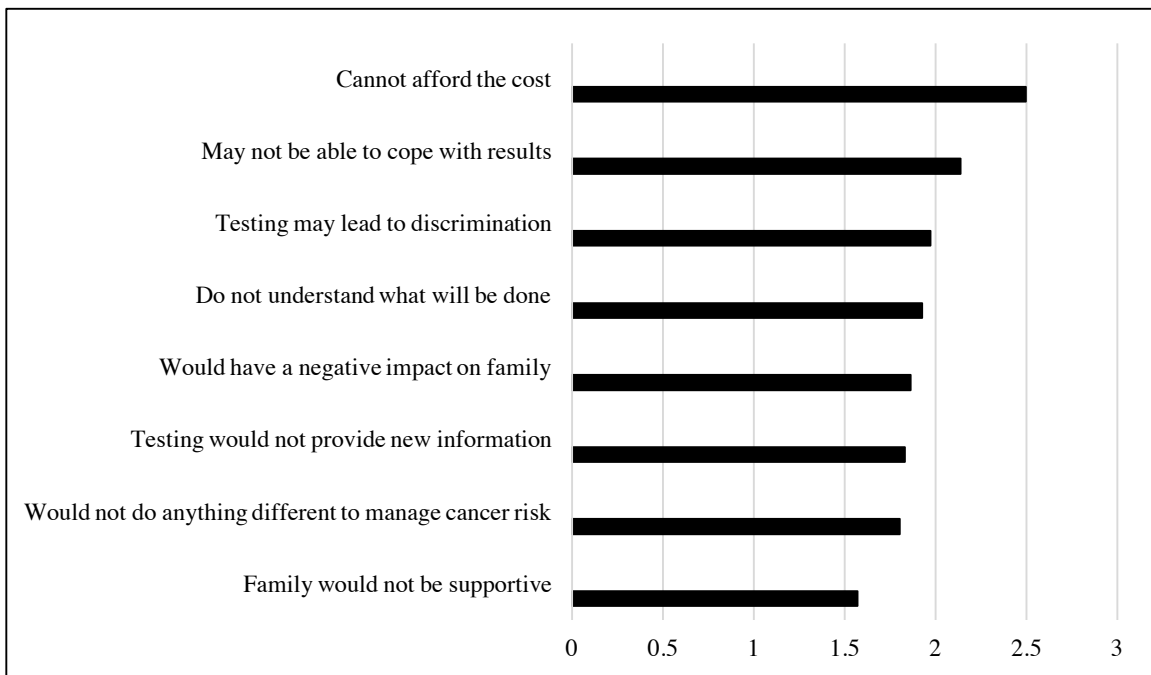


Figure 2.2 Agreement with barriers of genetic testing

to do anything different to manage their cancer risk, and testing would not provide new information. Women only agreed with one proposed barrier, that they would not be able to afford the cost of testing.

Interest strongly influenced women's perceptions of susceptibility, benefits, and barriers to genetic testing for hereditary cancer (Table 2.4). Women who were interested in testing were more likely to perceive themselves as susceptible to a genetic mutation that would predispose them to breast cancer ($p < 0.01$). Interested participants agreed more strongly with all proposed benefits to genetic testing ($p < 0.01$). Interested women statistically significantly agreed more with the statement "I am afraid to undergo genetic testing because I do not understand what will be done" ($p < 0.01$), and the statement "I cannot afford the cost of genetic testing" ($p = 0.02$). Women who were not interested in testing agreed statistically significantly more than interested women with a few barriers to genetic testing, including the statements, "Genetic testing will not help me because I would not do anything different to manage cancer risk" ($p < 0.01$), "Genetic testing will have a negative impact on my family" ($p = 0.03$), and "Genetic testing will not tell me anything new about my risk I do not already know" ($p = 0.03$). Interest had no statistically significant effect on women's reports of perceptions of the impact a hereditary breast cancer syndrome would have on their lives or women's reports of self-efficacy.

Age played a role in influencing women's perceptions of the benefits and barriers to genetic testing. Younger women were more likely to agree with the statement, "Genetic testing will help me make important life decisions" ($p = 0.02$). They more strongly agreed with a few barriers, including "I am afraid to undergo genetic testing

because I may not be able to cope with the result” ($p < 0.01$), “I am afraid to undergo genetic testing because I do not understand what will be done” ($p < 0.01$), and “I cannot afford the cost of genetic testing” ($p < 0.01$).

There were statistically significant differences in the benefits and barriers women most strongly agreed with between different racial groups (Figures 2.3 & 2.4). Black women were statistically significantly more likely ($p < 0.01$) to agree with the statement “Genetic testing will help me make important life decisions” than non-Hispanic white women. In terms of barriers, black women were more likely to agree with the statements, “I am afraid to undergo genetic testing because I may not be able to cope with the results” ($p = 0.02$), “I am afraid to undergo testing because I do not understand what will be done” ($p < 0.01$), and “I cannot afford the cost of genetic testing” ($p < 0.01$) than non-Hispanic white women. Asian women were statistically significantly more likely to agree with the statement “Genetic testing will not help me because I would not do anything different to manage my cancer risk” ($p = 0.01$) than Hispanic women.

Highest level of education influenced women’s perceptions of the benefits and barriers to genetic testing for hereditary cancer. Women who reported receiving less than a bachelor degree were more likely to agree with the statement, “Genetic testing will help me make important life decisions” ($p < 0.01$) than those that reported receiving a bachelor’s degree. Women who reported receiving less than a bachelor degree were also more likely to agree with the statements “I am afraid to undergo genetic testing because I may not be able to cope with the results” ($p = 0.03$), “I am afraid to undergo genetic testing because I do not understand what will be done” ($p < 0.01$), and “I cannot afford the cost of genetic testing” ($p = 0.01$).

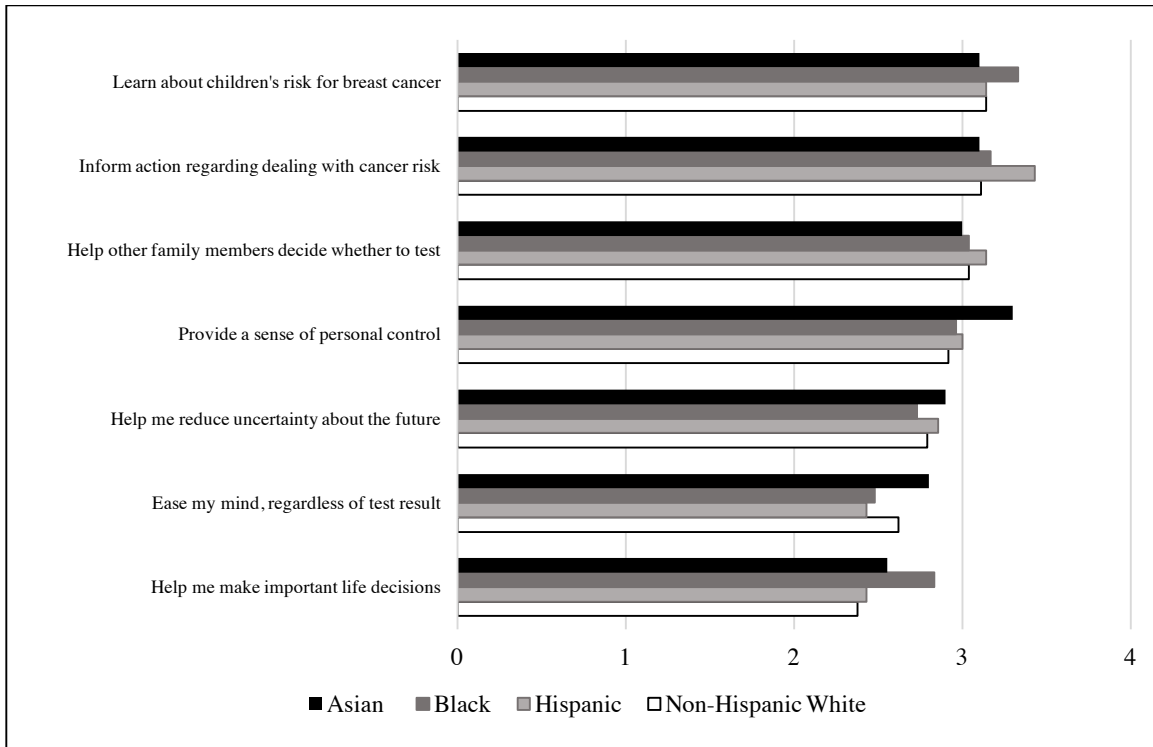


Figure 2.3 Race vs. agreement with benefits

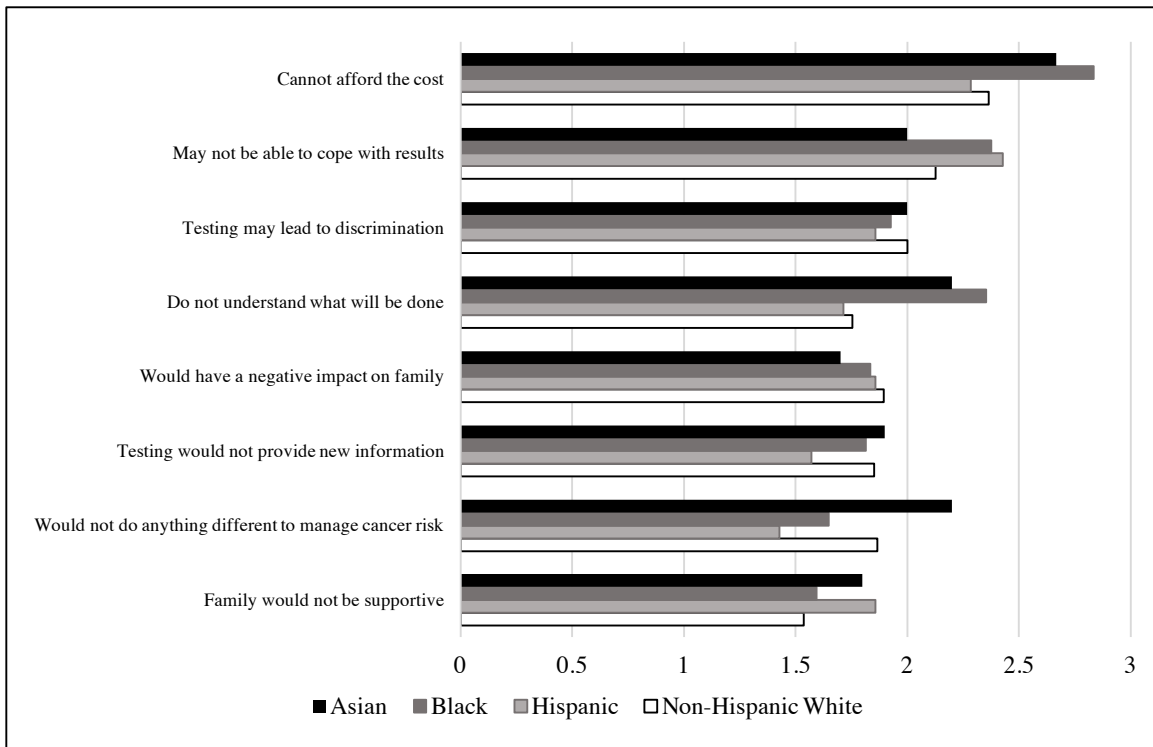


Figure 2.4 Race vs. agreement with barriers

Marital status was similarly associated with perceptions of benefits and barriers regarding genetic testing for hereditary breast cancer. Women who reported never being married were statistically significantly more likely to agree with the statements, “Genetic testing will help me make important life decisions” ($p < 0.01$), “I am afraid to undergo genetic testing because I may not be able to cope with the result” ($p < 0.01$), “I am afraid to undergo genetic testing because I do not understand what will be done” ($p < 0.01$), and “I cannot afford the cost for genetic testing” ($p < 0.01$) than women who reported that they were married.

Women who reported having no children were more likely to agree with the statements, “Genetic testing will help me make important life decisions” ($p < 0.01$). “I am afraid to undergo genetic testing because I may not be able to cope with the result” ($p < 0.01$), “I am afraid to undergo genetic testing because I do not understand what will be done” ($p < 0.01$), and “I cannot afford the cost for genetic testing” ($p < 0.01$) than women who reported having two or more children. Women who reported having one child trended to being less likely to agree with the statement, “Genetic testing to learn my risk will help other family members decide whether to undergo testing” ($p = 0.04$) than women who reported having no children or more than one child.

Participant’s reported insurance was only statistically significantly associated with increased agreement with barriers to genetic testing. Participants who reported utilizing government provided health insurance were more likely to agree with the statements “I am afraid to undergo genetic testing because I may not be able to cope with the results” ($p < 0.01$) than those that reported utilizing employer-provided health insurance. These participants were also more likely to agree with the statement, “I am afraid to undergo

genetic testing because I do not understand what will be done” ($p < 0.01$) than participants reporting utilization of university-provided health insurance or employer provided health insurance. Participants who reported that they were currently uninsured or utilizing university-provided health insurance were statistically significantly more likely to agree with the statement, “I cannot afford the cost of genetic testing” ($p < 0.01$). Participants who reported utilizing employer-provided or self-provided private insurance more strongly agreed with the statement, “Genetic testing will not tell me anything new about my risk I do not already know” ($p = 0.01$) than those who reported utilizing university-provided health insurance.

Annual household income was also only statistically significantly associated with increased agreement with barriers to genetic testing. Participants who reported annual household incomes of less than \$10,000 were statistically significantly more likely to agree or strongly agree with the statement “I am afraid to undergo genetic testing because I do not understand what will be done” ($p < 0.01$) than participants who reported annual household incomes of greater than \$200,000. Participants who reported annual household incomes of less than \$125,000 were statistically significantly more likely to agree with the statement, “I cannot afford the cost for genetic testing” ($p < 0.01$) than participants who reported annual incomes of greater than \$200,000.

2.4.5 Discrimination

We examined our hypothesis that interest in genetic counseling would be lower for those who have higher reports of perceived discrimination. Unexpectedly, participants who reported higher frequencies of daily discrimination also reported greater interest in genetic testing. To investigate our findings further, we examined the potential

mediating role of perceived susceptibility of a having inherited a genetic mutation that would predispose one to cancer between the relationship of perceived discrimination and interest in genetic counseling. Although current perspectives on mediation propose that the requirements outlined by Baron and Kenny (1986) are unnecessary (Shrout & Bolger, 2002), relations between our study variables did meet the expectations. Both the predictor, perceived discrimination, and the mediator, perceived susceptibility, were related to the outcome variable, genetic testing interest (Table 2.5). We present our findings in the conventional Baron and Kenny format for ease of interpretation.

Table 2.5 Correlations between perceived discrimination, perceived susceptibility, and interest in genetic testing

	1.	2.	3.
Perceived Susceptibility	-	.258**	.173*
Interest in Genetic Counseling		-	.244**
Perceived Discrimination			-

* Indicates $p < 0.05$

** Indicates $p < 0.01$

Using a multiple logistic regression, we regressed the criterion variable of interest in genetic testing on perceived discrimination in Step 1. In Step 2, we regressed interest in genetic testing on both the predictor of perceived discrimination and on the proposed mediator of perceived susceptibility and saw a reduction in the strength of perceived discrimination to predict interest in genetic counseling. The third regression equation provided an estimate of path c' , the relationship between perceived discrimination and interest in genetic counseling, controlling for perceived susceptibility. When path c' is zero, there is evidence of complete mediation. Path c' was found to be reduced, and no longer statistically significant, suggesting partial mediation. The $R^2_{adj} = .05$ for the final analysis indicated that almost 5% of the variability in interest in genetic counseling could

be accounted for by the mediation of perceived discrimination by perceived susceptibility.

We used the Sobel test to test the statistical significance of the mediating effect and to determine whether the change from path *c* to path *c'* of the standardized regression coefficients was statistically significant. The statistically significant decrease (Sobel tests = 2.24, $p = .03$) verified that perceived susceptibility partially mediated the relationship between perceived discrimination and interest in genetic testing. A graphic representation of this effect can be found in Figure 2.5

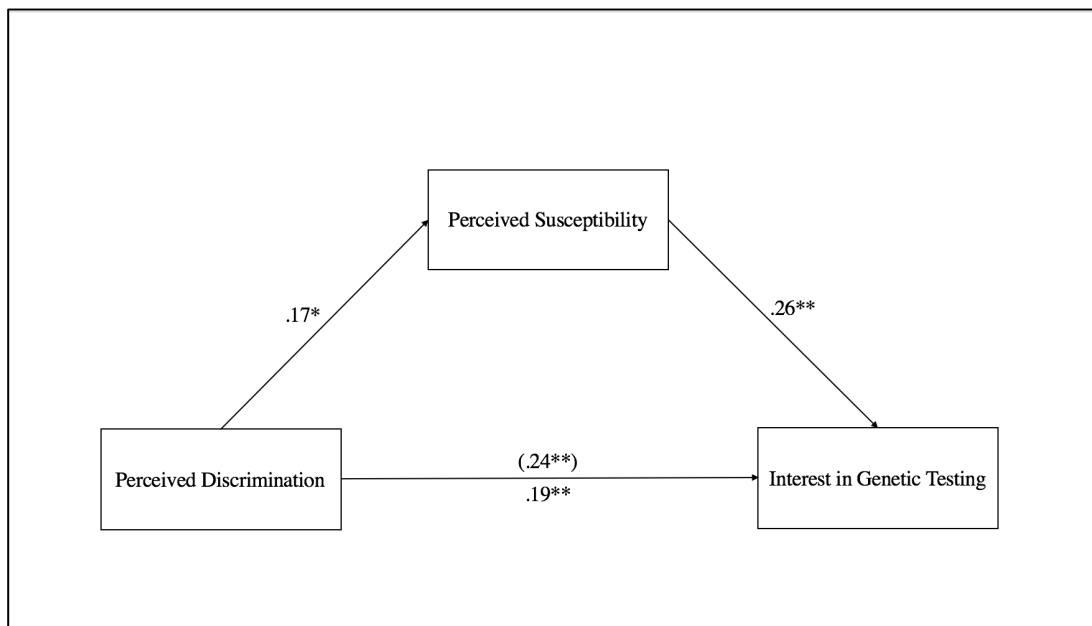


Figure 2.5 Mediating role of perceived susceptibility in the relationship between perceived discrimination and interest in genetic testing

2.4.6 BRCA1/2 Knowledge

In general, women in this study scored high on all measures of knowledge of *BRCA1/2*. For 5 of the 6 items, greater than or equal to 77% of women were able to correctly identify true or false statements. However, only 53% of participants were able

to correctly identify the statement, “The *BRCA* gene causes about one half of all breast cancers” as “False”.

2.5 Discussion

The purpose of this study was to assess women’s interest in and attitudes towards genetic testing for hereditary breast cancer across multiple demographic factors. The main goals of the study were to identify factors that predict women’s interest in testing and to clarify women’s perceptions of the utility of testing through the use of a Health Belief Model (HBM). Given the rapidly evolving attitudes towards and increasing accessibility to genetic testing, potential consumers’ thoughts regarding this service are especially pertinent.

Previous studies have shown that genetic testing services are primarily utilized by Caucasian women with private health insurance, and higher levels of education and income. Minorities are less likely to be aware of the genetic testing services available to them (Glenn, Chawla, & Bastani, 2012). Minority women have previously cited barriers to testing such as accessibility, language barriers, and distrust in the medical community (Glenn, Chawla, & Bastani, 2012; Thompson et al., 2003).

2.5.1 Awareness of Genetic Testing Services

The results of this study suggest the majority of women are aware of genetic testing services. Our study noted statistically significant differences in frequency of awareness between multiple demographic categories. Among our participants, black women, women who had lower levels of education, and women with lower annual incomes were all less aware of hereditary breast cancer genetic testing. These results

support the findings of previous studies, and indicate that further outreach in low socioeconomic communities by genetics professionals may be beneficial in reducing disparities among consumers (Komenaka et al., 2015; Mai et al., 2014).

2.5.2 Interest in Genetic Testing for Hereditary Breast Cancer

Over half of our participants expressed interest in utilizing genetic testing for hereditary breast cancer syndromes. There were statistically significant differences between the women who reported interest and those who did not. Some of these differences appear to stem from family unit demographics. Single women were more likely than married women to report interest, and women with no children were more likely to report interest in testing than women with 2 or more children. This observation adds support to previous findings suggesting women who perceive themselves to have stronger social supports are more likely to postpone genetic testing (Hesse-Biber & An, 2016).

Age had a statistically significant relationship with interest in testing. Consistent with previous findings, younger women were more likely to express interest in genetic testing for hereditary breast cancer than older women in our sample. As patient populations age, they have been found to have decreased interest in medical information seeking (Turk-Charles, Meyerowitz, & Gatz, 1997). Alternatively, younger women's increased interest in testing may be attributable to more exposure or education on genetic testing in their formal schooling, or an acknowledgement of the approaching midlife years and the increased risk of cancer that accompanies them (Sanderson et al., 2004; Bottorff et al., 2002).

Women who reported receiving lower levels of education were more likely to report interest in testing services than those who reported higher levels of education. Women who have higher levels of education may have more access and knowledge regarding genetic technology and may be more aware of the limitations and indications for genetic testing than those who have received lower levels of education (Sanderson et al., 2004).

Notably, no statistically significant relationship between prior awareness of testing services and interest in testing services was observed. Selected populations that reported less awareness of services, such as black women and women with lower annual incomes, did not have a statistically significant difference in their reported interest when compared to other participants. However, black women did trend towards significance in being more likely to report interest than non-Hispanic white women.

2.5.3 Health Belief Model

In this study, the Health Belief Model (HBM) provided a framework to better understand women's perceptions regarding the utility, benefits, barriers, and risks of genetic testing for hereditary breast cancer. The HBM was used to not only examine women's perceptions as a whole, but also women's perceptions within demographic categories in order to help clarify differing attitudes towards genetic testing for hereditary breast cancer.

Overall, participants expressed a positive attitude towards genetic testing for hereditary breast cancer syndromes. On average, women agreed with all of the proposed benefits to testing and disagreed with the majority of proposed barriers. Women agreed most strongly with the proposed benefits that genetic testing would help them learn about

their children's risk for breast cancer, help inform action regarding dealing with cancer risk, and help other family members make testing decisions. These benefits all confer alterations to the personal health management of the participant and their family members, and indicate that women are aware of and appreciate the clinical utility of this testing.

As a whole, women in this study only agreed with one proposed barrier to genetic testing for hereditary breast cancer syndromes. Participants were more likely to cite agreement with the statement that they would not be able to afford the cost of testing, suggesting that the perceived financial burden of genetic testing is a deterrent to women's decisions to pursue testing. Current billing procedures for genetic testing for hereditary cancer syndromes are complex and vary based on indication, insurance plan, and the laboratory chosen to complete the testing. The cost of genetic testing for hereditary cancer billed to insurance may vary from \$1,500 to \$5,000, but if the patient is determined to be an appropriate candidate for testing, out of pocket expenses are anticipated to be much lower at approximately \$100 (Invitae, 2017; Myriad Genetics, 2017). Currently, many laboratories offer financial assistance programs for underinsured patients determined to be appropriate candidates for testing that can reduce the out of pocket cost to less than \$100 or waive the out of pocket cost completely.(Invitae, 2017; Myriad Genetics, 2017). Education prior to the genetic counseling appointment regarding the average anticipated cost of testing may help encourage women who believe this service is not financially attainable to pursue genetic counseling when indicated.

Reported interest in testing was found to be statistically significantly associated with women's perceptions of their personal risk of having a genetic mutation that would

increase the risk of having breast cancer. Perceptions of severity were not significantly different between interested and uninterested women. Stronger agreement with all of the proposed benefits of genetic testing was statistically significantly associated with interest, suggesting that women who are interested in testing are also more likely to perceive there to be clinical and personal utility in testing than those who are not.

Women who did not report interest in genetic testing statistically significantly agreed more with three of the proposed barriers and risks. Uninterested women were more likely to believe that testing would not affect their decisions regarding the management of cancer risk and would not provide new information about their cancer risk. Both of these barriers relate to clinical utility and may indicate that women who are not interested in testing are less likely to perceive testing would provide personal health gain. The last barrier that uninterested women agreed statistically significantly more with was that testing would have a negative impact on their families. Given that the women in our sample who most frequently reported disinterest in testing were those who were married with more than one child, it is possible that this finding may be partially explained by appreciation for the significance a positive result would have on offspring.

There were statistically significant differences in our sample regarding attitudes and beliefs of racial groups towards genetic testing for hereditary breast cancer. Black women were statistically significantly less likely to be aware of testing services, indicating that they may be receiving less exposure and education on genetic testing services than other races. Interestingly, black women trended towards expressing significantly more interest in testing services than non-Hispanic white women. These findings may suggest that a contributory factor towards the lower frequencies of minority

women utilizing genetic testing services is lack of knowledge of and access to these services, as opposed to a lack of appreciation of the potential utility of these services.

Black women were statistically significantly more likely to agree with three barriers or risks to genetic testing than non-Hispanic white women. Black women were significantly more likely to agree that they were afraid to undergo genetic testing because they did not understand what would be done, and that they could not afford the cost of testing. Both of these barriers cite educational and accessibility concerns that may be aided by counseling and outreach.

Black women were also more likely to agree with the statement that they would not be able to cope with the results of genetic testing. Previous studies have revealed a complex relationship between fear of cancer and use of screening services. Women who perceive themselves to be mildly or severely at risk of cancer are less likely to utilize mammography than women who perceive themselves to be at moderate risk (Andersen et al., 2003). The underuse of mammography in black women has been hypothesized to be partially influenced by enhanced perceptions of breast cancer fatalism. In a qualitative study, African American women described coping strategies such as denial and repression to deal with fear of death due to breast cancer that caused them to avoid preventative care, such as breast cancer screening (Peek, Sayad, & Markwardt, 2008). Prior studies have also noted that women who report more intense emotional representations of hereditary cancer and difficulty understanding hereditary cancer are more likely to report difficulty coping with hereditary cancer testing (Voorwinden & Jaspers, 2015). It is possible a contributory factor to black women's underuse of genetic testing services is related to an underlying fear of breast cancer diagnosis and an

enhanced perspective of the probability of death due to breast cancer. Genetic counselors should be aware that these patients may benefit psychosocially as they adjust emotionally throughout the testing process from counseling addressing the effectiveness of treatment for breast cancer when medical care is proactively sought.

In our study, socioeconomic status was found to be associated with more agreement with perceived barriers to genetic testing. Education, health insurance coverage, and annual household income all statistically significantly influenced women's attitudes towards genetic testing for hereditary breast cancer. Women were more likely to agree that they would not be able to afford the cost of genetic testing if they had received less than a bachelor's degree, if they were currently uninsured or utilizing university-provided health insurance, or if they reported a household income of less than \$125,000. The financial burden of genetic testing is a significant concern for women in lower socioeconomic classes that may be aided by education on patient assistance programs and financial aid counseling. Women who reported receiving less than a bachelor degree, government-provided insurance, and a household income of less than \$10,000 were all more likely to agree that they were afraid to undergo genetic testing because they did not understand what would be done. Education in low socioeconomic communities should include an exploration of the agenda of a genetic counseling session and the subsequent testing process.

2.5.4 Discrimination

Previous studies consistently note the significance of historical discrimination and residual distrust in the medical community as a barrier to genetic testing in minority women (Kennedy, Mathis, & Woods, 2007; Glenn, Chawla, & Bastani, 2012). The

participants in our study did not statistically significantly differ in their reports of discrimination between racial groups or socioeconomic factors. Our results indicated a statistically significant relationship between reported interest in testing and increased frequency of daily events of discrimination. Further analysis revealed a confounding relationship between interest and discrimination that was partially explained by perceptions of susceptibility to a cancer predisposing mutation. Women who reported higher perceptions of susceptibility to a mutation not only significantly scored higher on the daily discrimination scale, but also reported significantly more interest in testing. This relationship indicates that women who experience more discrimination may perceive themselves to be more susceptible to a mutation that would increase their risk of breast cancer.

Previous studies have linked discrimination to many adverse physical and mental health outcomes (Pascoe & Richman, 2009). To our knowledge, this is the first study to show a relationship between discrimination and increased perceptions of susceptibility to a genetic syndrome. Discrimination has the potential to be a source of chronic stress that may accumulate to have significant effects on long-term health. Beyond the impact stress may have on health, distress and anxiety have been associated with overestimations of cancer risk (Tilburt, et al., 2011). Our findings indicate that those who experience discrimination may be at greater risk of anxiety inflating their perceptions of their risk of having a genetic mutation that would cause an increased risk of cancer.

2.5.5 BRCA1/2 Knowledge

Overall, the women in our sample scored high on the knowledge portion of the questionnaire. Of note, one item, “The *BRCA* gene causes about one half of all breast

cancers” (False), was answered incorrectly by almost half of the sample. As *BRCA* mutations account for approximately 5% of all breast cancer cases, women in this study had inflated perceptions of the frequency of *BRCA* related cancers. Women may benefit from education and counseling from their healthcare providers regarding the frequency of genetic cancers.

2.5.6 Practice Implications

Previous studies have noted the underuse of preventative genetic testing for hereditary breast cancer in minority women. This study revealed that black women tend to be less aware of genetic testing services, but equally as interested as their peers in receiving them, indicating that lack of knowledge and accessibility are key contributory factors to this underutilization as opposed to a lack of appreciation of the potential utility of genetic testing for hereditary breast cancer. Similarly, women of lower socioeconomic status, who reported lower household incomes, levels of education, and government provided health insurance, were more likely to be unaware of services than their peers but no less interested. Increasing exposure to genetic testing services in underserved populations is possible, but may require the aid of multiple health care providers.

Mobile mammography has been shown to be an effective method at increasing breast cancer screening in underserved communities and offering a channel of access to women who have barriers to the medical community (Chen et al., 2016; Brooks et al., 2013; Massin-Short et al., 2010). Given that our participants from these communities expressed equal interest in genetic testing, despite less awareness, our findings support the inclusion of a genetic counselor or genetics-trained health provider on a mobile

mammography unit to speak with interested or high-risk women as a strategy to allow for more education and exposure to genetic testing in these populations.

A novel relationship between increased experiences of discrimination and increased perceptions of susceptibility to a genetic mutation that would increase breast cancer risk was noted in our participants. Prior studies have indicated that women who feel more discrimination are less likely to utilize health services (Pascoe & Richman, 2009; Andersen et al., 2003). While it may be more difficult to reach women who feel more discrimination, they may benefit significantly from counseling and education regarding risk estimates and strategies to decrease the risk of cancer.

As stated previously, historically, minorities, women who have non-private health insurance, and women in lower socioeconomic classes are among the least likely to pursue genetic testing services. In our study, black women, women with government-provided health insurance, women with lower annual household incomes, and women with less formal education all consistently agreed statistically significantly more with one barrier to testing, “I am afraid to undergo genetic testing because I do not understand what will be done”. Unlike many of the potential barriers proposed in this study, this is a patient concern genetic counselors and other health professionals are equipped to directly address and impact.

Genetic counselors providing cancer-based genetic testing services should be aware that minority women and women with lower socioeconomic status may arrive to a genetics appointment with different concerns than other patients. Beyond contracting with the patient, specifically addressing the procedural and logistical aspects of the genetic testing process early in the appointment may aide in reducing perceived barriers

to these patients. Although unconventional, touching on the testing process and anticipated cost of testing prior to or in the beginning of the appointment may allow women less comfortable in a medical setting to better focus on the genetic information and education portions of the session later on if their concerns are adequately addressed from the start. Additionally, information and educational materials designed for women in underserved communities should specifically address the procedural and financial aspects of genetic testing.

2.6 Limitations and Future Studies

One of the goals of this study was to capture the perspectives of a diverse population. In order to do so, a number of self-selecting sampling methods were utilized, which created potential self-selection bias. Women who chose to participate in this study may have had stronger beliefs and attitudes towards genetic testing than women who did not.

While the sample collected was diverse in terms of education, income, age, and family unit characteristics, 85% of participants reported residing in the eastern region of the United States. Future studies may wish to explore differences in attitudes across the United States. Black women were well represented in the study, but the perspectives of women of other minority groups were less represented. These results are not generalizable to women of all minority backgrounds.

Information regarding HBOC and genetic testing was provided in a summary to the participants. It is not clear if participants fully understood the summary, as the item used to assess knowledge of *BRCA1/2* most frequently incorrectly answered by

participants was addressed in the summary. This may exemplify the complex nature of explaining genetic predispositions by written material alone. We did not determine how participants received information regarding genetic testing and HBOC. Learning which sources women use to learn about genetic testing and hereditary cancer risk may have provided revealing information.

While the HBM provided an abstract framework for exploring women's perceptions and attitudes towards genetic testing, further clarification through qualitative data would have aided in the interpretation of results. A significant observation that was noted in this study was women's agreement that the cost of testing was perceived as a barrier. Participants were not asked specifically what they believed the cost of testing to be. Future studies should explore women's concrete perceptions of the potential financial burden of genetic testing to form a more accurate picture of this significant barrier.

This study focused specifically on the breast cancer risks of HBOC. In reality, women with *BRCA1/2* mutations are at increased risk for multiple cancers, including ovarian and pancreatic cancer. It is possible that knowledge of these cancers risks may have an impact on participants' interest in and attitudes towards genetic testing for *BRCA1/2* that was not measured in this study.

A relationship between women's reports of discrimination, increased interest in testing, and increased perceptions of susceptibility to the inheritance of a genetic mutation that would increase breast cancer risk was noted. To our knowledge, this is the first study to demonstrate this relationship. It would be interesting to explore if this relationship is present in other adult-onset conditions that are moderated by both genetic

risk and environmental risk factors like stress and anxiety, such as Alzheimer's disease or heart disease.

The results of this study support intervention and outreach, potentially through mobile mammography or mobile health units, in underserved communities to provide education on genetic testing. Future studies should examine the effectiveness of such interventions in the appropriate utilization of genetic testing services in underserved communities, as well as the effectiveness of altering the genetic counseling agenda to address issues such as expected out of pocket costs and the testing procedure prior to the genetic counseling session through handout or phone call, or in the beginning of the genetic counseling session.

2.7 Summary

In recent years, genetic testing for hereditary cancer syndromes has not only become more accessible to the general population, but also received more media attention and exposure, increasing the general public's awareness of this medical service. Genetic testing offers many potential benefits, including earlier and more frequent screening to prevent advanced stages of cancer, and the provision of personalized risk information to family members. Given the rapidly evolving attitudes towards and increasing accessibility to genetic testing, potential consumers' thoughts regarding this service are especially pertinent.

Our findings indicate that the majority of women are aware of and interested in genetic testing for hereditary breast cancer syndromes, and perceive there to be clinical and personal utility in testing, across demographic categories.

We noted statistically significant differences in the concerns and barriers to genetic testing cited by black women and women of lower socioeconomic status when compared to other participants. We suggest that genetic counselors providing services to these women are especially cognizant of the potential concerns of cost and lack of awareness of what genetic testing entails expressed by these populations, and consider addressing these concerns prior to or earlier in the genetic counseling session to ensure these patients are able to focus on the education provided without the distraction of these potential barriers. Future studies are necessary to determine the effectiveness of this proposal.

We observed a relationship between participants reports of increased frequency of discrimination and increased interest in genetic testing that was partially modified by participant's perceptions of their susceptibility of having a genetic mutation that would increase their risk of cancer. These results suggest that women who experience more discrimination have inflated perceptions of their hereditary cancer risk. Future studies may wish to explore if this relationship is present with other adult-onset diseases with genetic risk, such as heart disease or Alzheimer's disease. Previous studies have shown that individuals who experience more discrimination are less likely to utilize medical services. Given that the women in our study who reported more discrimination cited higher concern about their susceptibility to a mutation as well as increased interest in testing, strategic outreach to provide counseling services on personalized risk estimates to this population may aide in decreasing cancer-related anxiety.

Our findings have shown that women from underserved communities and women who experience more discrimination are equally as interested in genetic testing as other

participants, but more likely to experience barriers related to education and access that make obtaining services more difficult. We recommend the inclusion of a genetics trained health professional on health units designed to reach underserved communities to aide in counseling on hereditary cancer risk and the process of obtaining genetic testing to women who qualify as high-risk. Future studies are necessary to determine the effectiveness of this proposed intervention in increasing the uptake of appropriate testing in these populations.

CHAPTER III. CONCLUSIONS

In this study, participants were generally aware of and interested in genetic testing services. We accept our hypothesis that participants of all backgrounds express similar levels of interest in genetic testing services. While women from traditionally underserved populations were less likely to have heard of genetic testing for hereditary breast cancer prior to this study, they reported similar levels of interest to other participants in this study.

Notably, there were no significant differences between populations' agreement with benefits and barriers regarding the clinical utility of testing, suggesting that even women who have less exposure to genetic technology are able to perceive the benefit and personal gain they might receive through predictive risk analysis.

As we expected, women from traditionally underserved populations were more likely to agree with proposed barriers regarding accessibility. Specifically, women from these populations agreed significantly more that they were concerned about cost and lack of awareness of what would be done in genetic testing.

We observed an unanticipated relationship between perceived susceptibility to a risk increasing genetic mutation and experiences of discrimination. Women who experience more discrimination may also have increased levels of stress and anxiety that inflate perceptions of hereditary cancer risk.

REFERENCES

- Allain, D.C., Friedman, S., & Senter, L. (2012). Consumer awareness and attitudes about insurance discrimination post enactment of the Genetic Information Nondiscrimination Act. *Familial Cancer, 11*(4), 637-644.
<http://doi.org/10.1007/s10689-012-9564-0>
- Andersen, M.R., Smith, R., Meischke, H., Bowen, D., & Urban, N. (2003). Breast cancer worry and mammography use by women with and without a family history in a population-based sample. *Cancer Epidemiology, Biomarkers & Prevention, 12*(4): 314 – 320. Retrieved from: <http://cebp.aacrjournals.org/content/12/4/314.long>
- Anderson, D.E. (1992). Familial versus sporadic breast cancer. *Cancer, 70*(S4), 1740-1746. [http://doi.org/10.1002/1097-0142\(19920915\)70:4+<1740::AID-CNCR2820701615>3.0.CO;2-1](http://doi.org/10.1002/1097-0142(19920915)70:4+<1740::AID-CNCR2820701615>3.0.CO;2-1)
- Apse, K.A., Biesecker, B.B., Giardiello, F.M., Fuller, B.P., & Berhardt, B.A. (2004). Perceptions of genetic discrimination among at-risk relatives of colorectal cancer patients. *Genetics in Medicine, 6*(6), 510-516.
<http://doi.org/10.1097/01.GIM.0000144013.96456.6C>
- Baron, R.M., & Kenny, D.A. (1986). The Moderator-Mediator Variable Distinction in Social Psychology Research: Conceptual, Strategic, and Statistical Considerations. *Journal of Personality and Social Psychology, 51*(6): 1173 – 1182. Retrieved from: <http://www.sesp.org/files/The%20Moderator-Baron.pdf>
- Becker, M.H., ed (1974). The Health Belief Model and Personal Health Behaviors. *Health Education Monographs, 2*(4), 324 – 473.
- Beran, T.M., Stanton, A.L., Kwan, L., Seldon, J., Bower, J.E., Vodermaier, A., & Ganz, P.A. (2008). The Trajectory of Psychological Impact in BRCA1/2 Genetic Testing: Does Time Heal? *Annals of Behavioral Medicine, 36*(2), 107-116.
<http://doi.org/10.1007/s12160-008-9060-9>

- Berg, W., Zhang, Z., Lehrer, D., Jong, R., Pisano, E.D., Barr, R.G., Bohm-Velez, M., Mahoney, M.C., Evans, W.P., Larsen, L.H., Morton, M.J., Mendelson, E.B., Farria, D.M., Cormack, J.B., Marques, H.S., Adams, A., Yeh, N.M., & Gabrielli, G. (2012). Detection of Breast Cancer With Addition of Annual Screening Ultrasound or a Single Screening MRI to Mammography in Women with Elevated Breast Cancer Risk. *JAMA: The Journal of the American Medical Association*, 307(13), 1394-1404. <http://doi.org/10.1001/jama.2012.388>
- Botkin, J.R., Smith, K.R., Croyle, R.T., Baty, B.J., Wylie, J.E., Dutson, D., Chan, A., Hamann, H.A., Lerman, C., McDonald, J., Venne, V., Ward, J.H., & Lyon, E. (2003). Genetic testing for a *BRCA1* mutation: Prophylactic surgery and screening behavior in women 2 years post testing. *American Journal of Medical Genetics*, 118A(3), 201-209. <http://doi.org/10.1002/ajmg.a.10102>
- Bottorff, J.L., Ratner, P.A., Balneaves, L.G., Richardson, C.G., McCullum, M., Hack, T., Chalmers, K., & Buxton, J. (2002). Women's Interest in Genetic Testing for Breast Cancer Risk: The Influence of Sociodemographics and Knowledge. *Cancer Epidemiology, Biomarkers & Prevention*, 11(1): 89-95. Retrieved from: <http://cebp.aacrjournals.org/content/11/1/89.long>
- Brooks, S.E., Hembree, T.M., Shleton, B.J., Beache, S.C., Aschbacher, G., Schervish, P.H., & Dignan, M.B. (2013). Mobile Mammography in Underserved Populations: Analysis of Outcomes of 3,923 Women. *Journal of Community Health*, 38(5): 900-906. <http://doi.org/10.1007/s10900-013-9696-7>
- Burke, W., Daly M., Garber, J., Botkin, J., Ellis-Kahn, M.-J., Lynch, P., McTiernan, A., Offit, K., Perlman, J., Petersen, G., Thomon, E., & Varricchio, C. (1997). Recommendations for follow-up care of individuals with an inherited predisposition to cancer. II. *BRCA1* and *BRCA2*. Cancer Genetics Studies Consortium. *JAMA: The Journal of the American Medical Association*, 277(12), 997-1003. <http://doi.org/10.1001/jama.1997.03540360065034>
- Chen, Y.R., Chang-Halpeny, C., Kumarasamy, N.A., Venegas, A., & Braddock, C.H. (2016). Perspectives of Mobile Versus Fixed Mammography in Santa Clara County, California: A Focus Group Study. *Cureus*, 8(2): e494. <http://doi.org/10.7759/cureus.494>
- Clain, E., Trosman, J.R, Douglas, M.P., Weldon, C.B., & Philips, K.A. (2015) Availability and payer coverage of *BRCA1/2* tests and gene panels. *Nature Biotechnology*, 33(9): 900-902. <http://doi.org/10.1038/nbt.3322>

- Culver, J., Burke, W., Yasui, Y., Durfy, S., & Press, N. (2001). Participation in Breast Cancer Genetic Counseling: The Influence of Educational Level, Ethnic Background, and Risk Perception. *Journal of Genetic Counseling*, 10(3), 215-231. <http://doi.org/10.1023/A:1016662524428>
- Donnelly, L.S., Watson, M., Moynihan, C., Bancroft, E., Evans, D.G., Eeles, R., Lavery, S. & Ormondroyd, E. (2013). Reproductive decision-making in young female carriers of a BRCA mutation. *Human Reproduction*, 28(4), 1006-1012. <http://doi.org/10.1093/humrep/des441>
- DeSantis, C., Ma, J., Bryan, L. & Jemal, A. (2013). Breast cancer statistics. *CA: A Cancer Journal for Clinicians*, 64(1), 52-62. <http://doi.org/10.3322/caac.21203>
- Durfy, S.J., Bowen, D.J., McTiernan, A., Porleder, J., & Burke, W. (1999). Attitudes and Interest in Genetic Testing for Breast and Ovarian Cancer Susceptibility in Diverse Groups of Women in Western Washington. *Cancer Epidemiology, Biomarkers & Prevention*, 8(4.2), 369-375. Retrieved from http://cebp.aacrjournals.org/content/8/suppl_1/369.full.pdf+html
- Erwin, C. (2008). Legal update: living with the Genetic Information Nondiscrimination Act. *Genetic Medicine*, 10(12): 869 – 874. <http://doi.org/10.1097/GIM.0b013w31818ca4e7>
- Evans, D.G.R., Barwell, J., Eccles, D.M., Collins, A., Izatt, L., Jacobs, C., Donaldson, A., Brady, A.F., Cuthbert, A., Harrison, R., Thomas, S., Howell, A. Miedybrodzka, Z., & Murray, A. (2014) The Angelina Jolie effect: how high celebrity profile can have a major impact on provision of cancer related services. *Breast Cancer Research*, 16(5), 442-448. <http://doi.org/10.1186/s13058-014-0442-6>
- Feldman, E.A. (2012). The Genetic Information Nondiscrimination Act (GINA): public policy and medical practice in the age of personalized medicine. *Journal of General Internal Medicine*, 27(6), 743-746. <http://doi.org/10.1007/s11606-012-1988-6>
- Forman, A.D., & Hall, M.J. (2009). Influence of Race/Ethnicity on Genetic Counseling and Testing for Hereditary Breast and Ovarian Cancer. *The Breast Journal*, 15(S1), S56-S62. <http://doi.org/10.1111/j.1524-4741.2009.00798.x>
- Gabai-Kapara, E., Lahad, A., Kaufman, B., Friedman, E., Segeve, S., Renbaum, P., Berri, R., Gal, M., Grinshpun-Cohen, J., Djemal, K., Mandell, J.B., Lee, M.K., Beller, U., Catane, R., King, M.C., & Levy-Lahad, E. (2014). Population-based screening for breast and ovarian cancer risk due to *BRCA1* and *BRCA2*. *Proceedings of the National Academy of Sciences of the United States of America*, 111(39): 14205 – 14210. <http://doi.org/10.1073/pnas.1415979111>

- Glenn, B.A., Chawla, N., Bastani, R. (2012). Barriers to Genetic Testing for Breast Cancer Risk Among Ethnic Minority Women: An Exploratory Study. *Ethnicity & Disease*, 22(3), 267-273. Retrieved from <http://www.ishib.org/ED/journal/22-03/ethn-22-03-267.pdf>
- Gooding, H.C., Organista, K., Burack, J., Biesecker, B.B. (2005). Genetic susceptibility testing from a stress and coping perspective. *Social Science & Medicine*, 62(8), 1880-1890. <http://doi.org/10.1016/j.socscimed.2005.08.041>
- Gurmenkin Levy, A., Shea, J., Williams, S.V., Quistber, A., & Armstrong, K. (2006). Measuring Perceptions of Breast Cancer Risk. *Cancer Epidemiology Biomarkers & Prevention*, 15(10), 1893-1898. <http://doi.org/10.1158/1055-9965.EPI-05-0482>
- Hall, M.J., Reid, J.E., Burbidge, L.A., Pruss, D., Deffenbaugh, A.M., Frye, C. Wenstrup, R.J., Ward., B.E., Scholl, T.A., & Noll, W.W. (2009). *BRCA1* and *BRCA2* mutations in women of different ethnicities undergoing testing for hereditary breast-ovarian Cancer. *Cancer*, 115(12), 2222-2233. <http://doi.org/10.1002/cncr.24200>
- Hartmann, L.C., Schaid, D.J., Woods, J.E., Crotty, T.P., Myers, J.L., Arnold, P.G., Petty, P.M., Sellers, T.A., Johnson, J.L., Donnell, S.K., Frost, M.H., & Jenkins, R.B. (1999). Efficacy of bilateral prophylactic mastectomy in women with a family history of breast cancer. *New England Journal Medicine*, 240(2):77-84. <http://doi.org/10.1056/NEJM199901143400201>
- Hesse-Biber, S., & An, C. (2016). Genetic Testing and Post-Testing Decision Making among *BRCA*-Positive Mutation Women: A Psychosocial Approach. *Journal of Genetic Counseling*. Advance online publication. <http://doi.org/10.1007/s10897-015-9929-2>
- Hoffman, S.J., & Tan, C. (2013). Following celebrities' medical advice: meta narrative analysis. *British Medical Journal*, 347(14), f7151-f7157. <http://doi.org/10.1136/bmj.f7151>
- Hutson, S.P. (2003). Attitudes and Psychological Impact of Genetic Testing, Genetic Counseling, and Breast Cancer Risk Assessment Among Women at Increased Risk. *Oncology Nursing Forum*, 30(2), 241-246. <http://doi.org/10.1188/03.ONF.241-246>
- Invitae. (2017). Billing Information. Retrieved from: <https://www.invitae.com/en/billing/>
- Jacobsen, P.B., Valdimarsdottir, H.B., Brown, K.L., Offit, K. (1997). Decision-Making About Genetic Testing Among Women at Familial Risk for Breast Cancer. *Psychosomatic Medicine*, 59(5), 459-466. <http://doi.org/10.1097/00006842-199709000-00001>
- Janz, N.K., & Becker, M.H. (1984). The Health Belief Model: A Decade Later. *Health Education & Behavior*, 11(1): 1 – 47. <http://doi.org/10.1177/109019818401100101>

- Kennedy, B.R., Mathis, C.C., & Woods, A.K. (2007). African Americans and Their Distrust of the Health Care System: Healthcare for Diverse Populations. *Journals of Cultural Diversity*, 14(2): 56 – 60.
- King, M.C., Wieand, S., Hale, K., Lee, M., Walsh, T., Owens, K., Tait, J., Ford, L., Dunn, B.K., Costantino, J., Wickerham, L., Wolmark, N., & Fisher, B. (2001). Tamoxifen and breast cancer incidence among women with inherited mutations in BRCA1 and BRCA2: National Surgical Adjuvant Breast and Bowel Project (NSABP-P1) Breast Cancer Prevention Trial. *JAMA: The Journal of the American Medical Association*, 286(18), 2251-2256. <http://doi.org/10.1001/jama.286.18.2251>
- Krieger, N., Smith, K., Naishadham, D., Hartman, C., Barbeau, E.M. (2005). Experiences of discrimination: validity and reliability of a self-report measure for population health research on racism and health. *Social Science & Medicine*, 61(7): 1576 – 1596. <http://doi.org/10.1016/j.socscimed.2005.03.006>
- Koils, C.E., Everett, J.N., Hoechstetter, L., Ricer, R.E., & Huelsman, K.M. (2003). Differences in physician referral practices and attitudes regarding hereditary breast cancer by clinical practice location. *Genetics in Medicine*, 5(5), 364-369. <http://doi.org/10.1097/01.GIM.0000086477.00766.C9>
- Koh, H.K., & Sebelius, K.G. (2010). Promoting Prevention through the Affordable Care Act. *New England Journal of Medicine*, 363(14), 1296-1299. <http://doi.org/10.1056/NEJMp1008560>
- Komenaka, I.K., Nodora, J.N., Madlensky, L., Winton, L.M., Heberer, M.A., Schwab, R.B., Weitzel, J.N., & Martinex, M.E. (2015). Participation of lower-income women in genetic cancer risk assessment and *BRCA1/2* testing: the experience of a safety-net institution. *Journal of Community Genetics*. Advance online publication. <http://doi.org/10.1007/s12687-015-0257-x>
- Kosenko, K.A., Binder, A.R., & Hurley, R. (2016). Celebrity Influence and Identification: A Test of the Angelina Effect. *Journal of Health Communication*, 21(3), 318-326. <http://doi.org/10.1080/10810730.2015.1064498>
- Lerman, C., Daly, M., Masny, A., & Balshem, A. (1994). Attitudes about genetic testing for breast-ovarian cancer susceptibility. *Journal of Clinical Oncology*, 12(4), 843-850. Retrieved from: <http://jco.ascopubs.org/>
- Lerman, C., Narod, S., Schulman, K., Hughes, C., Gomez-Caminero, A., Bonney, G., Gold, K., Trock, B., Main, D., Lynch, J., Fulmore, C., Snyder, C., Lemon, S.J., Conway, T., Tonin, P., Lenoir, G., & Lynch, H. (1996). *BRCA1* Testing in Families with Hereditary Breast-Ovarian Cancer: A Prospective Study of Patient Decision Making and Outcomes. *JAMA: The Journal of the American Medical Association*, 275(24), 1885-1892. <http://doi.org/10.1001/jama.1996.03530480027036>

- Mai, P.L., Vadaparampil, S.T., Breen, N., McNeel, T.S., Wideroff, L., & Graubard, B.I. (2014). Awareness of Cancer Susceptibility Genetic Testing: The 2000, 2005, and 2010 National Health Interview Surveys. *American Journal of Preventive Medicine*, 46(5), 440-448. <http://doi.org/10.1016/j.amepre.2014.01.002>
- Manchanda, R., Legood, R., Burnell, M., McGuire, A., Raikou, M., Loggenberg, K., Wardle, J., Sanderson, S., Side, L., Balogun, N., Desai, R., Kumar, A., Dorkins, H., Wallis, Y., Chapman, C., Taylor, R., Jacobs, C., Tomlinson, I., Beller, U., Menon, U., & Jacobs, I. (2014). Cost-effectiveness of population screening for BRCA mutations in Ashkenazi jewish women compared with family history-based testing. *Journal of the National cancer Institute*, 107(1): dju380. <http://doi.org/10.1093/jnci/dju380>
- Massin-Short, S.B., Grullon, M.A., Judge, C.M., Ruderman, K.R., Grullon, M., & Lora, V. (2010). A Mobile Mammography Pilot Project to Increase Screening Among Latina Women of Low Socioeconomic Status. *Public Health Reports*, 125(5); 765 – 771. <http://doi.org/10.1177/003335491012500521>
- Moller, P., Hagen, A.I., Apold, J., Maehle, L., Clark, N., Fiane, B., Lovslett, K., Hovig, E., & Vabo, A. (2007). Genetic epidemiology of *BRCA* mutations – family history detects less than 50% of the mutation carriers. *European Journal of Cancer*, 43(11): 1713 – 1717. <http://doi.org/10.1016/j.ejca.2007.04.023>
- Myriad Genetics. (2017). Billing and Payment Options. Retrieved from: <https://new.myriadpro.com/documents-and-forms/billing-and-payment-options/>
- National Association of Community Health Centers, the Robert Graham Center. (2007). Access denied: a look at America's medically disenfranchised. *Washington: NACHC and the Robert Graham Center*. Retrieved from <http://www.graham-center.org/content/dam/rgc/documents/publications-reports/monographs-books/Access%20Denied.pdf>
- National Comprehensive Cancer Network. (2016). Genetic/Familial High-Risk Assessment: Breast and Ovarian (Version 2.2016). Retrieved from <http://www.nccn.org>
- National Cancer Institute. (2013). SEER Stat Fact Sheets: Female Breast Cancer. Retrieved from <http://seer.cancer.gov/statfacts/html/breast.html>.
- National Cancer Institute. (2014). A Snapshot of Breast Cancer. Retrieved from <http://www.cancer.gov/research/progress/snapshots/breast>
- National Cancer Institute. (2015). Risk Factors for Cancer. Retrieved from <http://www.cancer.gov/about-cancer/causes-prevention/risk>.

- Offit, K., Bradbury, A., Storm, C., Merz, J.F., Noonan, K.E., & Spence, R. (2013). Gene Pateints and Personalized Cancer Care: Impact of the *Myriad* Case on Clinical Oncology. *Journal of Clinical Oncology*, 31(21), 2742-2748.
<http://doi.org/10.1200/JCO.2013.49.7388>
- Pascoe, E.A., & Richman, L.S. (2009). Perceived Discrimination and Health: A Meta-Analytic Review. *Psychology Bulletin*, 135(4): 531-554.
<http://doi.org/10.1037/a0016059>
- Peek, M.E., Sayad, J.V., & Markwardt, R. (2008) Fear, Fatalism and Breast Cancer Screening in Low-Income African-American Women: The Role of Clinicians and Health Care System. *Journal of General Internal Medicine*, 23(11): 1847 – 853.
<http://doi.org/10.1007/s11606-008-0756-0>
- Pharoah, P.D.P., Antoniou, A., Bobrow, M., Zimmern, R.L., Easton, D.F., & Ponder, B.A.J. (2002). Polygenic susceptibility to breast cancer and implications for prevention. *Nature Genetics*, 33(1), 33-36. <http://doi.org/10.1038/ng853>
- Rebbeck, T.R., Friebel, T., Lynch, H.T., Neuhausen, S.L., van't Veer, L., Garber, J.E., Evans, G.R., Narod, S.A., Isaacs, C., Matloff, E., Daly, M.B., Olopade, O.I., & Weber, B.L. (2004). Bilateral Prophylactic Mastectomy Reduces Breast Cancer Risk in *BRCA1* and *BRCA2* Mutation Carriers: The PROSE Study Group. *Journal of Clinical Oncology*, 22(6), 1055-1062. <http://doi.org/10.1200/JCO.2004.04.188>
- Rosenbaum, S. (2011). The Patient Protection and Affordable Care Act: Implications for Public Health Policy and Practice. *Public Health Reports*, 126(1), 130-135.
Retrieved from <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3001814/>
- Rosenstock, I.M. (1974). Historical origins of the health belief model. *Health Education Monographs*, 2(4): 328 – 335. Retrieved from:
<http://journals.sagepub.com/doi/pdf/10.1177/109019817400200403>
- Rosenstock, I.M., Strecher, V.J., & Becker, M.H. (1988). Social learning theory and the Health Belief Model. *Health Education Quarterly*, 15(2): 175 – 183. Retrieved from:
https://www.iccwa.org.au/useruploads/files/soyf/2013_resources_videos/health_belief_model.pdf
- Sanderson, S.C., Wardle, J., Jarvis, M.J., & Humphries, S.E. (2004). Public interest in genetic testing for susceptibility to heart disease and cancer: a population-based survey in the UK. *Preventive Medicine*, 39(3): 458-464.
<http://doi.org/10.1016/j.ypmed.2004.04.051>
- Scheuer, L., Kauff, N., Robson, M., Kelly, B., Barakat, R., Satagopan, J., Ellis, N., Hensley, M. Boyd, J., Borgen, P., Norton, L., & Offit, K. (2002). Outcome of Preventative Surgery and Screening for Breast and Ovarian Cancer in *BRCA*

- Mutation Carriers. *Journal of Clinical Oncology*, 20(5), 1260-1268.
<http://doi.org/10.1200/JCO.20.5.1260>
- Schwarzer, R., & Jerusalem, M. (1995). Generalized Self-Efficacy scale. In J. Weiman, S. Wright, & M. Johnston, *Measures in health psychology: A user's portfolio. Causal and control beliefs* (33-37). Windsor, UK: NFER-NELSON.
- Shrout, P.E., & Bolger, N. (2002). Mediation in Experimental and Nonexperimental Studies: New Procedures and Recommendations. *Psychological Methods*, 7(4): 422 – 445. <http://doi.org/10.1037//1082-989X.7.4.422>
- Siegel, R.L., Miller, K.D., & Jemal, A. (2015). Cancer Statistics, 2015. *CA: A Cancer Journal for Clinicians*, 65(1), 5-29. <http://doi.org/10.3322/caac.21254>
- Strecher, V., & Rosenstock, I.M. (1997). The Health Belief Model. In Glanz, K., Lewis, F.M., & Rimer, B.K. (Eds.), *Health Behavior and Health Education: Theory, Research, and Practice*, 2nd end. San Francisco, CA: Jossey-Bass. 41-59.
- Susswein, L.R., Skrzynia, C., Lange, L.A., Booker, J.K., Graham, M.L., & Evans, J.P. (2008). Increased uptake of BRCA1/2 genetic testing among African American women with a recent diagnosis of breast cancer. *Journal of Clinical Oncology*, 26(1), 32-36. <http://doi.org/10.1200/JCO.2007.10.6377>
- Tambor, E.S., Rimer, B.K., & Strigo, T.S. (1997). Genetic testing for breast cancer susceptibility: Awareness and interest among women in the general population. *American Journal of Medical Genetics*, 68(1), 43-49.
[http://doi.org/10.1002/\(SICI\)1096-8628\(19970110\)68:1<43::AID-AJMG8>3.0.CO;2-Z](http://doi.org/10.1002/(SICI)1096-8628(19970110)68:1<43::AID-AJMG8>3.0.CO;2-Z)
- Thompson, H.S., Valdimarsdottir, H.B., Jandorf, L., & Redd, W. (2003). Perceived disadvantages and concerns about abuses of genetic testing for cancer risk: differences across African American, Latina, and Caucasian women. *Patient Education and Counseling*, 51(3), 217-227. [http://doi.org/10.1016/S0738-3991\(02\)00219-7](http://doi.org/10.1016/S0738-3991(02)00219-7)
- Tilburt, J.C., James, K.M., Sinicrope, P.S., Eton, D.T., Costello, B.A., Carey, J., Lane, M.A., Ehlers, S.L., Erwin, P.J., Nowakowski, K.E., & Murad, M.H. (2011). Factors Influencing Cancer Risk Perception in High Risk Populations: A Systematic Review. *Hereditary Cancer in Clinical Practice*, 9(1): 2.
<http://doi.org/10.1186/1897-4287-9-2>
- Turk-Charles, S., Meyerowitz, B.E., & Gatz, M. (1997). Age Differences in Information-Seeking among Cancer Patients. *The International Journal of Aging and Human Development*, 47(2): 85-98. <http://doi.org/10.2190/7CBT-12K3-GA8H-F68R>

- van den Broek, A.J., Schmitt, M.K., van't Veer, L.J., Tollenaar, R.A., & van Leeuwen, F.E. (2015). Worse breast cancer prognosis of BRCA1/BRCA2 mutation carriers: what's the evidence? A systematic review with meta-analysis. *PLoS One*, 10(3). <http://doi.org/10.1371/journal.pone.0120189>
- Voorwinden, J.S., & Jaspers, J.P.C. (2015). Prognostic Factors for Distress After Genetic Testing for Hereditary Cancer. *Journal of Genetic Counseling*, 43(1), 71-77. <http://doi.org/10.1016/j.jejca.2006.08.023>
- Walcott, F.L., Dunn, B.K., DeShields, M., & Baquet, C. (2014). The Affordable Care Act and Genetic Testing for Inheritable Cancer Syndromes: Impact on High-Risk Underserved Minorities. *Journal of Health Care for the Poor and Underserved*, 25(1), 46-62. <http://doi.org/10.1353/hpu.2014.0070>
- Wang, C., Gonzalez, R., Janz, N.K., Milliron, K.J., & Merajver, S.D. (2007). The role of cognitive appraisal and worry in BRCA1/2 testing decisions among a clinic population. *Psychology and Health*, 22(6), 719-736. <http://doi.org/10.1080/14768320600976216>
- Wang, S.M. (2016). Pros and Cons for Population Screening BRCA Mutation Carriers. *Clinics in Oncology*, 1(1161): 1-3. Retrieved from: http://www.clinicsinoncology.com/pdfs_folder/cio-v1-id1161.pdf
- Wevers, M.R., Aaronson, N.K., Verhoef, S., Bleiker, E.M.A., Hahn, D.E.E., Kuenen, M.A., van der Sanden-Melis, J., Brouwer, T., Hogervorst, F.B.L., van der Luijt, R.B., Valdimarsdottir, H.B., van Dalen, T., Theunissen, E.B.M., van Ooijen, B., de Roos, M.A., Borgstein, P.J., Vrouwenraets, B.C., Vriens, E., Bouma, W.H., Rijna, H., Vente, J.P., Witkamp, A.J., Rutgers, E.J.T., & Ausems, M.G.E.M. (2014). Impact of rapid genetic counselling and testing on the decision to undergo immediate or delayed prophylactic mastectomy in newly diagnosed breast cancer patients: findings from a randomised controlled trial. *British Journal of Cancer*, 110(4), 1081-1087. <http://doi.org/10.1038/bjc.2013.805>
- Williams-Jones, B. (2002). History of a Gene Patent: Tracing the Development and Application of Commercial BRCA Testing. *Health Law Journal*, 10, 121-144. Retrieved from <http://genethics.ca/personal/papers/HistoryPatent.pdf>
- Welsh, P.L., & King, M.C. (2001). BRCA1 and BRCA2 and the genetics of breast and ovarian cancer. *Human Molecular Genetics*, 10(7), 705-713. <http://doi.org/10.1093/hmg/10.7.705>
- Wilder-Smith, A., Liegey-Dougall, A., Posluszn, D.M., Somers, T.J., Rubinstein, W.S., & Baum, A. (2007). Psychological distress and quality of life associated with genetic testing for breast cancer risk. *Psycho-Oncology*, 17(8), 767-773. <http://doi.org/10.1002/pon.1291>

APPENDIX A. PARTICIPANT RESOURCES

LOCATE A GENETIC COUNSELOR

If at any point during this questionnaire you became concerned about your risk of having a genetic mutation that would increase your chance of having cancer, consider contacting a local genetic counselor by following the steps below:

1. Go to the National Society of Genetic Counselors website homepage at:
<http://www.nsgc.org/>
2. On the NSGC homepage, click the link titled, “Find a Genetic Counselor” in the bottom right hand of the corner of your screen



3. Enter your postal code and make sure to click “Cancer” under specialization. Then, click search!

Search for a Genetic Counselor:

Search Tips:

- The "First Name" and "Last Name" fields should only be used if you know the name of the genetic counselor you are looking to contact.
- If you do not return any results within your zip code, try searching just by state.
- Students interested in contacting a genetic counselor about the profession should check the "Student Contact Welcome" box.

Search For: ☒ Any criteria ☐ Exact criteria

State/Province:

OR

Near (Postal Code or City, State):

Within:

First Name:

Last Name:

Types of Specialization:

- ☐ Hematology
- ☐ Adult (Including Complex Disease)
- ☒ Cancer
- ☐ Cardiac-Aortic Aneurysm

Student Contact Welcome: ☐

Sees Patients: ☐

Telephone Counseling Service: ☐

Board Certified: ☐

APPENDIX B. INVITATION TO PARTICIPANTS

University of South Carolina School of Medicine USC Genetic Counseling Program

Dear Potential Participant,

You are invited to participate in a graduate research study focused on women's perceptions of the purpose of genetic testing for hereditary breast cancer syndromes. I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. My research investigates factors that influence women's interest in genetic testing. This research will involve the completion of this questionnaire, which will collect information about you and your viewpoints regarding genetic testing and cancer. Your responses will help genetic counselors and health providers better understand women's viewpoints regarding genetic testing services and provide these services to a broader range of women.

All responses gathered from the surveys will be kept anonymous and confidential. The results of this study might be published or presented at academic meetings; however, participants will not be identified.

Your participation in this research is voluntary. By completing the questionnaire, you are consenting that you have read and understand this information. At any time, you may withdraw from this study by not completing the questionnaire.

If you have any questions regarding this research, you may contact either myself or my faculty advisor, Crystal Hill-Chapman, Ph.D., using the contact information below.

Taylor Apostolico, BS
(610) 955 9615
taylor.apostolico@gmail.com

Crystal-Hill Chapman, PhD
(843) 661 1721
chillchapman@fmarion.edu

If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803) 777-7095.

Thank you for your time and consideration to participate in this survey!

APPENDIX C. LETTER OF SUPPORT
University of South Carolina School of Medicine
USC Genetic Counseling Program

To the University of South Carolina IRB:

I am familiar with Taylor Apostolico's research project entitled "Assessing Women's Attitudes Towards Genetic Testing for Hereditary Breast Cancer". I understand [Organization Name] involvement to be permitting interested clients and community members to be surveyed and distributing questionnaires.

I understand that this research will be carried out following sound ethical principles and that participant involvement in this research study is strictly voluntary and provides confidentiality of research data, as described in the protocol.

Therefore, as a representative of [Organization Name] I agree that Taylor Apostolico's research project may be conducted at our agency.

Sincerely,

Organization Representative

Date

APPENDIX D. BACKGROUND INFORMATION



HEREDITARY BREAST CANCER

A Brief Introduction

What is Hereditary Breast Cancer?

Breast cancer is a cancer that develops from the breast tissue. It affects 1 in 8 women in the United States. Typically, this happens by chance. In a small percentage of breast cancer cases, it is caused by a genetic mutation in one of a few genes. When one of these genes has a mutation, it stops working leading to an increased risk of cancer. This form of breast cancer is called hereditary breast cancer.

Two of the more common genes that cause hereditary breast cancer are the BRCA1 and BRCA2 genes. Women who have mutations in these genes have a higher risk of developing a few cancers, including breast and ovarian cancer. Around 1-3% of people in the United States have a mutation in one of these genes. It is possible to test a person's blood for the presence of mutations that may cause a person to have an increased chance of getting breast cancer. For the purpose of this study, this process of obtaining genetic information related to a person's risk of having breast cancer will be called 'genetic testing for hereditary breast cancer'

What is a Genetic Counselor?

Genetic counselors are trained health care workers that help individuals understand the many implications of genetic contributions to disease. Genetic counselors use family and personal histories to determine an individuals' risk of having a genetic mutation, like one in a BRCA gene, that would increase the individual's risk of having cancer.

APPENDIX E. QUESTIONNAIRE

1. Had you heard of genetic testing for hereditary breast cancer before beginning this survey?
☐ Yes ☐ No
2. Have you personally had genetic testing for hereditary breast cancer before?
☐ Yes ☐ No
3. Is genetic testing for hereditary breast cancer something you would be interested in pursuing?
☐ Yes ☐ No
4. What do you think your chance is of developing breast cancer in your lifetime? Please choose a number between 0 (no chance of breast cancer) and 100 (definitely will get breast cancer)
5. How strongly do you agree or disagree with the following statements?

It is likely that I carry a gene mutation that increases my risk for breast cancer
☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

The chances that a gene mutation runs in my family are great
☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree
6. How strongly do you agree or disagree with the following statements?

If I found out that I carried a gene mutation, it would greatly disrupt my life
☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Finding out I carried a gene mutation would be very difficult for me
☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

If I found out I carried a gene mutation, I would worry much more about developing breast cancer
☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

7. How strongly do you agree or disagree with the following statements?

I am afraid to undergo genetic testing because I may not be able to cope with the result

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will not tell me anything about my risk I do not already know

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing to learn about my risk will give me a sense of personal control

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will lead to unfair treatment of some people – that is, discrimination

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will help me reduce uncertainty about the future

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will help me decide on the best course of action to take to deal with my cancer risk

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

My family will not be supportive if I undergo genetic testing

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will help me learn about my children's risk for breast cancer

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

I cannot afford the cost for genetic testing

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will ease my mind, regardless of the test result

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

I am afraid to undergo genetic testing because I do not understand what will be done

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing to learn my risk will help other family members decide whether to undergo testing

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will not help me because I would not do anything different to manage my cancer risk

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will have a negative impact on my family

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Genetic testing will help me make important life decisions (such as getting married, having children)

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

8. How strongly do you agree or disagree with the following statements?

I can solve most problems if I invest the necessary effort

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

I am confident that I could deal efficiently with unexpected events

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

If I am in trouble, I can usually think of a solution

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

If someone opposes me, I can find the means and ways to get what I want

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

When I am confronted with a problem, I can usually find several solutions

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

It is easy for me to stick to my aims and accomplish my goals

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

I can usually handle whatever comes my way

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

I can always manage to solve difficult problems if I try hard enough

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

Thanks to my resourcefulness, I know how to handle unforeseen situations

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

I can remain calm when facing difficulties because I can rely on my coping abilities

☐ Strongly Disagree ☐ Disagree ☐ Agree ☐ Strongly Agree

9. In your day-to-day life, how often do the following things happen to you?

You are treated with less courtesy or respect

☐ Never ☐ Rarely ☐ Occasionally ☐ Frequently

You receive poorer service than other people

☐ Never ☐ Rarely ☐ Occasionally ☐ Frequently

People act as if they think you are not smart

☐ Never

☐ Rarely

☐ Occasionally

☐ Frequently

People act as if they are afraid of you

☐ Never

☐ Rarely

☐ Occasionally

☐ Frequently

You are threatened or harassed

☐ Never

☐ Rarely

☐ Occasionally

☐ Frequently

10. Please answer the following questions as either True or False

A father can pass down an altered BRCA gene to his daughters

☐ True

☐ False

All women who have an altered BRCA gene will get cancer

☐ True

☐ False

A woman who has a sister with an altered BRCA gene has a 50% chance of having an altered BRCA gene herself

☐ True

☐ False

A woman who doesn't have an altered BRCA gene can still get cancer

☐ True

☐ False

There are many different genes that cause cancer

☐ True

☐ False

The BRCA gene causes about one half of all breast cancers

☐ True

☐ False

11a. Have you ever been diagnosed with cancer?

☐ Yes

☐ No

11b. If so, what type of cancer were you diagnosed with?

11c. If so, at what age(s) were you diagnosed with cancer?

12a. Have you ever had genetic testing for any condition?

☐ Yes

☐ No

12b. If so, what was the reason you had genetic testing?

13a. Has anyone in your family been diagnosed with breast cancer at any time in their life?

☐ Yes ☐ No

13b. If so, how many relatives have had breast cancer?

13c. If so, which relative(s) have had breast cancer? Please check all that apply.

<input type="checkbox"/> Mother	<input type="checkbox"/> Aunt (on mother's side)
<input type="checkbox"/> Sister	<input type="checkbox"/> Aunt (on father's side)
<input type="checkbox"/> Daughter	<input type="checkbox"/> Cousin (on mother's side)
<input type="checkbox"/> Grandmother (on mother's side)	<input type="checkbox"/> Cousin (on father's side)
<input type="checkbox"/> Grandmother (on father's side)	<input type="checkbox"/> Other (please specify)_____

14. Does your family have Ashkenazi Jewish ancestry?

☐ Yes ☐ No

15. What race do you most identify with?

<input type="checkbox"/> Alaskan Native	<input type="checkbox"/> Native Hawaiian
<input type="checkbox"/> American Indian	<input type="checkbox"/> Non-Hispanic White
<input type="checkbox"/> Asian	<input type="checkbox"/> Pacific Islander
<input type="checkbox"/> Black American	<input type="checkbox"/> Other (please specify)_____
<input type="checkbox"/> Hispanic	

16. What is the highest level of school you have completed or the highest degree you have received?

☐ Less than high school degree
☐ High school degree or equivalent (e.g., GED)
☐ Some college but no degree
☐ Associate degree
☐ Bachelor degree
☐ Graduate degree

17. What type of health insurance are you currently covered by?

☐ Currently uninsured
☐ Student Health Insurance (provided by University)
☐ Medicare
☐ Medicaid
☐ Employer provided private insurance
☐ Self-provided private insurance

18. Which of the following best describes your current relationship status?

☐ Married
☐ Widowed
☐ Divorced
☐ Separated
☐ In a domestic partnership or civil union
☐ Single, but cohabiting with a significant other
☐ Single, never married

19. How much total combined money did all members of your household earn last year?

☐ \$0 to \$9,999

☐ \$10,000 to \$24,000

☐ \$25,000 to \$49,999

☐ \$50,000 to \$74,999

☐ \$75,000 to \$99,999

☐ \$100,000 to \$124,999

☐ \$125,000 to \$149,999

☐ \$150,000 to \$174,999

☐ \$175,000 to \$199,999

☐ \$200,000 and up

☐ Prefer not to answer

20. What is your zip code? _____

21. How many children do you have? _____

22. What is your current age? _____