Decision-Making among individuals at risk for Hereditary Breast and Ovarian Cancer Syndrome (HBOC)

by

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Dedication

This dissertation is dedicated to my mother, Judi, whose spirit will continue to live on and remind me daily of the joys in life and to persevere above all. To my father, Doug, for his unwavering love, support and guidance. To my future husband, Jamie, who always embodies selflessness and devotion as we go through life together. To my friends who’ve kept my spirits high with laughter and support throughout this journey.

Acknowledgement

This work would not have been possible without the continued support and guidance of my cherished family and friends. I am grateful to my dissertation committee members for their mentorship and friendship by helping me to grow personally and professionally. I am indebted to Meghan Underhill-Blazey at the Phyllis F. Cantor Center at Dana-Farber Cancer Institute for taking me under her wing to help cultivate my skills as a nurse researcher, a writer, and a contributor in our field. I am humbled to have been mentored so closely by Barbara J. Guthrie with her extensive experience in nursing academia and dedication to our field of nursing. Finally, I am grateful to the faculty and staff at Bouvé College of Health Sciences at Northeastern University for all the opportunities to collaborate and be mentored over the course of my academic tenure. This experience instilled in me the importance of interprofessional collaborative practice in advancing genomics within the continuum of hereditary risk and cancer care through nursing scholarship.

“If I have seen further than others, it is by standing upon the shoulder of giants.”

~ Isaac Newton
# Table of Contents

Copyright.................................................................................................................. ii

Dedication and Acknowledgment.................................................................................. iii

Table of Contents......................................................................................................... iv

Chapters

I. Introduction................................................................................................................. 6

II. Manuscript 1........................................................................................................... 24

III. Manuscript 2......................................................................................................... 65

IV. Manuscript 3......................................................................................................... 84

V. Summary and Conclusions.................................................................................... 127
I. Introduction
Cancer is a major public health problem worldwide, and is ranked as the second leading cause of death in the United States (American Cancer Society, 2019). An estimated 62,930 new cases of breast (female), 95,830 new cases of melanoma, 22,530 new cases of ovarian, and one in five new diagnoses in prostate cancer are expected (ACS, 2019). In 2010, treatment for breast cancer alone accounted for 13% ($16.5 billion) of all direct-medical spending on cancer in the United States (Farina et al., 2012). However, a percentage of the population are at risk for hereditary forms of cancer, which among the general population are made up of 20-25% of breast, 15% of ovarian, and 5-10% of all cancers diagnoses (National Cancer Institute, 2017).

Hereditary breast and ovarian cancer syndrome (HBOC) is a syndrome that is associated with multiple types of cancers such as breast, ovarian, pancreatic, prostate, and melanoma (National Comprehensive Cancer Network (2017), and an increased risk of early-onset, triple negative, and bilateral breast cancer, a second primary cancer, and cancer reoccurrence (Centers for Disease Control and Prevention, 2015). Well-known and commonly studied genes associated HBOC include the \textit{BRCA1} and \textit{BRCA2 (BRCA1/2)} genes. These genes are primarily responsible for suppressing the development of tumors. When \textit{BRCA1/2} genes are mutated, tumors can grow or be “over-expressed” because the ability to control the function of the gene becomes absent (Hall et al., 1990). In HBOC, deleterious \textit{BRCA1/2} mutations are important hereditary risk factors and are considered key determinants of HBOC risk.

In the general population, the prevalence of harboring a \textit{BRCA1/2} mutation is 1 in 400 (\textit{BRCA1}) to 800 (\textit{BRCA2}), and 1 in 40 among those of Ashkenazi Jewish descent (NCCN, 2017). In women, the risk of developing breast cancer with a \textit{BRCA1/2} mutation is increased from 13% in the general population by age 70 to 45% (\textit{BRCA2}) and 65% (\textit{BRCA1}) (NCI, 2017; NCCN, 2017). For ovarian cancer, women are at an increased risk of 17% (\textit{BRCA2}) and 39% (\textit{BRCA1}) as
compared to 1.3% in the general population (NCI, 2017; NCCN, 2017). In men, lifetime risk of breast cancer by age 80 is 8.9% (Evans et al., 2010) compared to less than 1% among the general population. A mutation in BRCA1/2 has been estimated to contribute to 5%-20% of diagnosed male breast cancers (ACS, 2019) with a reported higher prevalence of BRCA2 versus BRCA1 mutation (Fentiman, 2009).

**Genetic Counseling and Testing Guidelines**

The discovery of genetic testing has advanced our knowledge in how to screen and treat hereditary cancers. Since HBOC is a cancer syndrome that can be passed on to blood-related relatives, the National Comprehensive Cancer Network (2017) provides guidelines for individuals and families to determine eligibility for genetic counseling and testing. Genetic counseling and testing should be offered to individuals who meet high risk criteria for HBOC and often focuses on multi-gene panel including BRCA1/2 mutations and other HBOC-associated genes (NCCN, 2017). Since the development and application of multi-gene panel testing (i.e. looking at a large group of genes in DNA versus just one or two), NCCN recommends the inclusion of other altered genes known to be associated with HBOC and other HBOC-associated cancers: ATM, BRIP1, CDH1, CHEK2, MSH2, MLH1, MSH6, PMS2, EPCAM, NBM, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53 (NCCN, 2017; Tung et al., 2016) (Table 1).

Genetic counseling and testing have become an effective screening method and a gold standard recommendation for at-risk individuals as they benefit from advanced early detection, prevention recommendations, and targeted cancer treatments (Edwards et al., 2008; National Cancer Institute, 2017; Livraghi & Garber, 2015; Mahon, 2001; O'Neill et al., 2010; Hamilton, Williams, Bowers, & Calzone, 2009; Proulx et al., 2009; Zilliacus et al., 2012). Several studies have stated the importance of genetic testing in at-risk relatives (Dancyger, Smith, Jacobs,
Wallace, & Michie, 2010; Denayer, Boogaerts, Philippe, Legius, & Evers-Kiebooms, 2009; Edwards et al., 2008; Etchegary et al., 2009; Hamilton et al., 2009; Norris, Spelic, Snyder, & Tinley, 2009; Proulx et al., 2009; Sussner et al., 2015) However, other studies have outlined the wide range in variability of at risk relatives (upwards of 59%) (Fehniger, Lin, Beattie, Joseph, & Kaplan, 2013; Finlay et al., 2008; Halbert, Kessler, Stopfer, Domchek, & Wileyto, 2006; Ropka, Wenzel, Phillips, Siadaty, & Philbrick, 2006) and those with a current or previous diagnosis of breast or ovarian cancer (Dancyger et al., 2010; Patenaude et al., 2013; Proulx et al., 2009; Vadaparampil, McIntyre, & Quinn, 2010) who choose not to undergo genetic testing despite being aware of their increased risk status of either developing an HBOC-related cancer or a second cancer diagnosis.

Individuals with HBOC-associated mutations are gatekeepers of this information to their blood-related relatives, which may impact their future health-related decisions later in life. Individuals at-risk for HBOC who are considering undergoing genetic testing find themselves making this decision with the influence of their family, and this decision can be the catalyst to their at-risk family members for obtaining genetic testing themselves. The decision-making process to uncover cancer risk within a family context is complex and an integral part of the genetic counseling and testing process. Factors associated with genetic counseling and testing among individuals with increased HBOC risk has been studied in large, urban academic medical centers, but the current literature does not adequately assess the genetic testing decision-making process among individuals residing in rural areas with limited access to genetic testing medical care.

A novel component to this research work is that it focused on a rural non-academic medical center population. An integrative review was completed to uncover gaps in the literature
among individuals at risk for HBOC in the decision whether to undergo genetic testing. Outcomes revealed that genetic counseling and testing should be tailored to the individual and family’s perspectives, which will aid future health-related decisions when discussing screening and prevention options.

The dissertation explored the decision-making process in the utilization of genetic testing in a rural environment among individuals with HBOC. The findings have the potential to inform the scientific community how those at-risk for HBOC make informed decisions about genetic testing. The exploration of their decision-making process will provide avenues for further individualized interventions to be developed surrounding HBOC care. The objectives of this dissertation are accomplished through the submission and expected publications of the following three manuscripts, which are presented in subsequent chapters of this dissertation.

**Manuscript One**

Manuscript One is an integrative literature review to identify and understand the decisional needs associated with the utilization of genetic testing in families with HBOC risk. The objective of this review was to identify factors associated with the decision to undergo BRCA1/2 genetic testing for HBOC. The results of this review made it evident that further exploration of this topic is warranted and has the potential to generate new knowledge to advance the field. Outcomes from the study described the need for more theoretically based, primary studies focusing on decision-making in individuals with hereditary cancer risk, which provided rationale for the study reported in Manuscript Three. This manuscript is intended for submission to *Familial Cancer*. 
Manuscript Two

Manuscript Two compares a variety of mid-range health behavior theories within the context of individuals at risk for HBOC. It further explored the relationships of how concepts and frameworks are operationalized and used within this population. The purpose of this manuscript was to (a) describe the relevance and importance of the findings of studies that used a theoretical framework, and (b) demonstrate the need for more theoretically-based studies focusing on the decision-making process within this population. Findings from this article are linked to the rationale behind using the current study’s theory and methods within the same population. This paper along with Manuscript One prepared the author for a program of research in decision-making focused on individuals with hereditary cancer risk. This manuscript is intended for submission to Journal of Health and Social Behavior.

Manuscript Three

Manuscript Three describes an original study undertaken by the author. This study used a qualitative approach and grounded theory methods. Its purpose was to explore the decision-making process in utilizing genetic testing among individuals with HBOC residing in a rural environment. This manuscript is intended for submission to Journal of Genetic Counseling.

Philosophical assumptions. Since content and word count are limited in journal authorship guidelines, it is necessary to explain the philosophical assumptions that underpin the methodological approach to this dissertation. Findings from Manuscript One and Two inform the theory and method used in the current study, Manuscript Three. A social constructivist, interpretive framework was utilized with a pragmatic, naturalistic epistemology.

Social constructivism was established in the 1930’s by Lev Vygotsky, who was named the “father of social constructivism.” Vygotsky’s publication in 1962, Thought and Language,
discussed how individual consciousness was built from the outside through relations with others, and that social behavior and consciousness are one in the same; that our awareness with ourselves and awareness of others is the same position as others to us. In 1966, Berger & Luckmann published the book, *The Social Construction of Reality*, to describe work contributions that established social constructivism as a paradigm and addressed its potential in social theory.

Constructivism as described by Lincoln & Guba (1985) entails, “realities that are social constructions of the mind, and that there exists as many such constructions as there are individuals” (p. 43). Researchers who identify as Constructivists re-define issues of replicability, validity and reliability with concepts such as trustworthiness (i.e. credibility, transferability, dependability and confirmability) during data analysis because they believe in the inductive process of the construction of meaning (Lincoln & Guba, 1985). Charmaz (2014) defines using a constructivist approach as constructing research processes and products and acknowledging that the construction is influenced by the researcher’s perspectives, privileges, positions, interactions, and geographic locations. Using the method of grounded theory with Constructivism highlights more flexibility compared to the more traditional post-positivist grounded theories (Charmaz, 2014). This viewpoint questions the notion that the researcher is a neutral observer, and allows for the researcher to examine their own pre-conceived notions and how their beliefs may identify and shape the analysis. The constructivist approach acknowledges subjectivity and the researcher’s involvement in the construction and interpretation of the data (Charmaz, 2014). The suggestion that subjective experiences through social interactions shape the meaning, experience, and action of a person or group of people is highly relevant to genetics and genetic testing. The purpose of using Charmaz (2014) version of grounded theory is with the goal of developing a
theoretical understanding of the process of genetic testing decision-making among at-risk individuals of hereditary breast and ovarian cancer syndrome.

**Ontological positions.** Ontology poses the question, “what is real or what is reality?” In this study, the approach was taken that each person’s reality was specific to their experience. There are multiple constructed realities that are considered subjective (Lincoln & Guba, 1985). In Charmaz (2008) description of a social constructivist reality, she describes how it is not possible to separate the researcher from the data and the analysis. Therefore, for this dissertation, the social constructivist ontology proposed by Charmaz (2014) was adopted, in which “social reality is multiple, processual, and constructed” (p. 13). Taking this ontological approach allowed the research to make careful observations and provide a description of the participant’s ordinary conscious experience (i.e. hearing, seeing, feeling, acting) in the form of transcripts and memo-writing to construct their individual realities.

**Epistemological positions.** Epistemological statements pertain to reality needing to be interpreted to understand the underlying meanings. The epistemological foundation to this dissertation work is considered subjective, pragmatic, and relativistic in nature, which is just one way of knowing that is highlighted by Charmaz (2014). Using a constructivist epistemology and ontology were related by “placing priority on the phenomena of study and seeing both data and analysis as created from shared experiences and relationships with participants and other sources” (Charmaz, 2014, p. 239). Pragmatism is similar with a complementary approach to naturalism. The philosophy of pragmatism is to pay careful attention to the ‘why’s’ people believe a certain way, and emphasize the practical application of ideas by acting on them to test them within human experience (Gutek, 2014). Those who are pragmatists view the practical uses of the nature of knowledge, language, concepts, meanings, belief, and science. This closely
aligns with Interpretivism, where social constructs aid the individual’s development of their reality.

Naturalism was proposed by Gubrium & Holstein (1997) and defined as “…a way of knowing that locates meaningful reality in the immediate settings of people’s daily affairs (p. 7).” Those seeking to use naturalism to identify, construct and interpret individual’s realities use descriptions of people and their interactions within their native habits to further understand the ‘what’ and ‘how’ things mean to them (Gubrium & Holstein, 1997). Understanding individuals’ meanings in the ‘what’s and how’s’ of their social life is crucial in being able to construct their reality (p. 200). Gubrium & Holstein (1997) further conceive that naturalists use an interpretive practice in determining how human beings become to know their reality. Interpretivism is based on a naturalistic approach, and relies on both the researcher and the participant as important aspects in the generation of the construct. It helps to further aid the facilitation of understanding the individual’s meaning and interactions, the ‘what’s and how’s’, and the motivations and values of the social life of that participant.

**Relevance to the current study.** Until recently, most studies that explore decision-making among those with increased hereditary cancer risk have used a qualitative approach informed by theorists such as Glaser, Strauss & Corbin who outlined a more structured and rigorous approach to coding and structure. Glaser & Strauss (1967) outlined a generality and objectivity approach to data analysis in an ‘empirical world’ using a constant comparative method. In their book, *Discovery of Grounded Theory*, they outlined seven steps for data analysis that encouraged objectivity, structure and formatting to coding. Later, Strauss & Corbin (1998) further defined these methods to slightly add more flexible guidelines and procedures; however, still philosophically believed in structure and objectivity of the data. In 1992, Glaser discussed
his approach to grounded theory as an objectivist stance, and his work was more influenced within professional disciplines such as nursing and management than in social sciences (Charmaz, 2008).

A grounded theory approach that follows the methodologies of Glaser & Strauss (1967) and Strauss & Corbin (1998) is no longer compatible in a traditional healthcare model, and within the current context of those living with hereditary cancer risk. Grounded theory methodologies that include relativity and reflexivity are crucial so that researchers are able to construct and interpret the realities of those living with cancer risk, such as guidelines set forth by Charmaz (2014) rather than grounded theory methodologies that consists of objectivity and generality, as outlined by Glaser, Strauss & Corbin. As more discoveries are being made regarding our genome and its relationship to cancer, more emphasis should be placed on understanding the individuals’ perspective of living with hereditary cancer risk along with highlighting the importance for personalized, not precision medicine.

For this dissertation, the value of using a pragmatic, naturalistic approach within an interpretive framework outlined by Charmaz (2014) highlights (a) the contribution that reflexivity and relatively provide to the phenomena of interest, and (b) the need to acknowledge the ‘social reality as multiple, processual and constructed’ that exist among individuals at risk for hereditary cancers from a rural environment. There is a lack of literature that explores the decision-making process in the utilization of genetic testing in these diverse perspectives, specifically those residing in a rural environment. Outcomes from this study will provide more in-depth understanding of those living with hereditary risk within this context, and will inform and further science in the development of personalized interventions that will aid individuals decision-making process during this time.
Methods. The objective of this study (as described in Manuscript Three) was to (a) describe the process by which individuals with HBOC who reside in rural settings with limited access to care from large, academic medical centers make genetic testing decisions, and (b) understand the social context in the decision-making process related to HBOC genetic testing. Since documented literature related to populations living with HBOC in a rural environment is limited, following a methodology that supports the inductive, iterative approach to generate new theorizing categories was crucial. A benefit to utilizing grounded theory is the ability for researchers to use previous grounded theorists’ such as Glaser, Strauss & Corbin’s structured guidelines to construct and analyze their research content. However, due to the context of decision-making among individuals with HBOC, a more reflexive and interpretative use of grounded theory is needed as outlined by Charmaz (2014).

Grounded theory was a natural fit for studying the process of decision-making in the utilization of genetic testing among individuals with HBOC in a rural environment. Consistent with the philosophical assumptions of social constructivism and naturalism, Charmaz (2014) defines using a constructivist approach as constructing research processes and products, and acknowledging that the construction is influenced by the researcher’s perspectives, privileges, positions, interactions, and geographic locations. The suggestion that subjective experiences through social interactions shape the meaning, experience, and action of a person or group of people is highly relevant to genetics and genetic testing as often the individual of family experience is more salient to a person than the objective genetic information provided in a clinical setting. Therefore, a theory that is grounded in the voices of individuals who have experienced these processes may shed light on the broader issues surrounding decision-making
within the context of a cancer diagnosis, those at higher risk for hereditary cancers, access to care and socioeconomic issues, and directions for future research.

The following three chapters will describe, in manuscript format, three contributions that the author has made to the field of decision-making and cancer genomics over the course of her doctoral program. These contributions are similar in approach and methodology, and highlight the importance of exploring the individual realities of those living with increased cancer risk to further aid in the development of tailored interventions. In the final chapter, the author will synthesize the findings from these three manuscripts, summarizes the implications to cancer genomics, decision-making in individuals living with increased cancer risk, and proposes next steps in her intended program of research.
References


cancer. *Patient Education and Counseling, 72*(2), 276-282


Oxford University Press.


### Table 1

*NCCN Guidelines (2017) for Genes Associated with HBOC*

<table>
<thead>
<tr>
<th>Gene</th>
<th>Breast Cancer</th>
<th>Ovarian Cancer</th>
<th>High Penetration*</th>
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*(Tung et al., 2016)*
II. Manuscript 1
Factors associated with genetic testing for Hereditary Breast and Ovarian Cancer Syndrome (HBOC): An integrative review of the literature and implications for practice

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Compliance with Ethical Standards The authors declare that they have no conflict of interest. This article does not contain any studies with human participants or animals performed by any of the authors.
FACTORS ASSOCIATED WITH HBOC

Abstract

**Background:** Though at risk relatives of individuals with hereditary breast and ovarian cancer syndrome (HBOC) are recommended genetic testing, uptake is low presenting a missed opportunity to promote health and wellness in high risk families.

**Purpose:** To understand HBOC genetic testing decision making.

**Methods:** EBSCOhost, CINAHL, PsychInfo, and Medline as well as reference lists from 2008 to 2018 were reviewed with relevant keywords. Two authors independently critiqued included articles.

**Findings:** 20 articles were included. The decision to undergo genetic testing was found to be influenced by social-ecological factors. Family relationships, communication among family members, personal experiences of a cancer diagnosis, cultural and gender influences, and access to care and insurance coverage were key factors in the utilization of genetic testing. There is a lack of data from diverse populations recruited outside of traditional academic medical settings.

**Discussion:** Future research should target social-ecological factors to improve the genetic testing process.

**Keywords:** Genomics, cancer risk, hereditary breast and ovarian cancer syndrome (HBOC), decision-making, genetic testing
FACTORS ASSOCIATED WITH HBOC

Introduction

Approximately 20-25% of breast, 15% of ovarian, and 5-10% of all cancers are due to a heritable cause.\(^1\) The etiology of hereditary breast and ovarian cancer is most commonly associated with tumor suppressor proteins \(BRCA1\) and \(BRCA2\) (referred to as \(BRCA1/2\) mutation).\(^2\) Other mutations in cancer susceptibility genes (i.e. \(ATM, CDH1, CHEK2, PALB2, PTEN, STK11, TP53\)) are also associated with HBOC.\(^1\) Among the general population, the prevalence of harboring a \(BRCA1/2\) mutation is 1 in 400 (\(BRCA1\)) to 800 (\(BRCA2\)),\(^3\) and 1 in 40 among those of Ashkenazi Jewish descent.\(^4\)

Nationwide standard guidelines exist for assessing who may be at high risk for HBOC and genetic counseling is considered standard of care.\(^4-6\) The hallmark presentation of hereditary breast cancer risk within a family is early age of cancer onset, bilateral cancer or the presence of associated cancers in multiple family generations. Individuals with risk factors that suggest a mutation in \(BRCA1/2\), have an increased predictive risk of developing cancer if they have inherited the mutation.\(^1\) First-degree relatives of family members with an identified deleterious \(BRCA1/2\) mutation have a 50% in inheriting this mutation, and risk also extends to more distant relatives.

In women, risk of breast cancer with a \(BRCA1/2\) mutation is increased from 13% in the general population by age 70 to 45% (\(BRCA2\)) and 65% (\(BRCA1\)).\(^1,4\) In ovarian cancer, women are at an increased risk of 17% (\(BRCA2\)) and 39% (\(BRCA1\)) as compared to 1.3% in the general population.\(^1,4\) In men, lifetime risk of breast cancer by age 80 is 8.9%\(^7\) compared to less than 1% among the general population.\(^8\) A mutation in \(BRCA1/2\) has been estimated to contribute to 5%-20% of diagnosed male breast cancers\(^9\) with a reported higher prevalence of \(BRCA2\) versus \(BRCA1\) mutation.\(^10\)
FACTORS ASSOCIATED WITH HBOC

Genetic counseling and testing is a gold standard medical recommendation for at high-risk populations to identify individuals who would benefit from advanced early detection, prevention recommendations, or targeted cancer treatments.\textsuperscript{1,11-14} Several studies have stated the importance of genetic testing in at-risk relatives,\textsuperscript{11,15-21} and among individuals diagnosed with HBOC because it has aided in the decision-making process for further detection and prevention strategies.\textsuperscript{18,20,22} Current studies have shown there are two categories of individuals who are aware of their high-risk status for developing an inherited form of cancer later in life, yet choose not to undergo genetic counseling and testing: those at high-risk who haven’t received a cancer diagnosis (from 13.5\% to upwards of 59\%)\textsuperscript{23-26,27,28} and those with a current or previous diagnosis of breast/ovarian cancer.\textsuperscript{15,20,29,30} Therefore, though there are clear guidelines as to the utility of genetic testing for HBOC, these guidelines clearly have not fully translated into an integrated clinical standard of care. Thus, the purpose of this integrative literature review was to identify and understand how individuals at risk for HBOC make \textit{BRCA1/2} genetic testing decisions to inform future intervention development to improve genetic testing decision making in this population. An in-depth exploration of the relevant gaps in knowledge, implications for future practice, and comparisons of themes are explained.

\textbf{Review Methods}

Searched articles included those that discussed genetic testing from either an individual diagnosed with a cancer associated with Hereditary Breast and Ovarian Cancer Syndrome (HBOC) or from a family member’s perspective with a known diagnosis associated with HBOC. Literature was identified using EBSCOhost, CINAHL, PsychInfo, and Medline through the following keyword searches: “genetic testing” “BRCA” “psychosocial factors” “genetic screening” “HBOC” “decision-making” in combination with the Boolean term ‘and.’ Keywords
FACTORS ASSOCIATED WITH HBOC

“famil*” “relativ*” were used in combination with the Boolean term ‘or.’ Eligible articles were published from 2008 to 2018 to capture the most recent literature, written in English, and NCCN guidelines\textsuperscript{31} were used to determine eligibility criteria. Included articles focused on the perspectives of either an at-risk individual of HBOC with no personal cancer history, an individual with a known HBOC-associated cancer, or an at-risk family member of a loved with a known HBOC associated-cancer. Articles related to conducting genetic mapping, outlining hereditary genes not associated with HBOC, studies testing technology interventions or validation measurement scales were excluded.

To enhance credibility, the co-authors of this literature review paper independently conducted a ‘quality check’ by selecting 10% literature identified at random and applying the inclusion and exclusion criteria. The results of the quality check were cross-referenced to the 20 included articles and it yielded 100% accuracy. The co-authors discussed exclusion for each article and any discrepancies were resolved during the quality check phase.

**Review Results of Characteristics of Included Studies**

Twenty research studies, eighteen of which are unique study populations, met inclusion criteria (Figure 1), including nine quantitative studies (Table 2), eight qualitative studies (Table 3), and three mixed-method studies (Table 4). Characteristics of included studies are detailed in Table 1. The most commonly cited factors identified in the studies reviewed were categorized as individual, familial, and health system context factors. The median age was 45 years old with one article including participants under the age of 18.\textsuperscript{19} Included studies discussed perspective from a variety of ethnic backgrounds. Seven articles did not report race/ethnicity, and perspectives from Asian and Hispanic background were either limited or not included. Most articles were from the female perspective with no articles pertaining to just the male perspective.
FACTORS ASSOCIATED WITH HBOC

Part of the criteria for this literature review was based on BRCA1/2 genetic testing, yet nine article didn’t state the BRCA1/2 status of their sample. Ten articles (50%) conducted their studies through academic/medical centers and/or large scale clinics whereas three articles (15%) were conducted by using cancer registries or databases. The remaining six articles (30%) were conducted through community-based strategies such as local flyers, web-based ads, support groups, local churches, and local non-profit organizations.

Decision Making about HBOC Genetic Testing: A Socioecological Decision-making Process

Factors identified as contributing to the genetic testing decision making process included personal and familial experiences with cancer, perceptions of screening, prevention and treatment, influence of culture and socioeconomic status, healthcare system factors, age and gender roles.

Individual factors contributing to genetic testing decisions

Demographic factors

Males and females approached genetic testing differently. Males were found to prioritize health in order to provide information to offspring, but often overlooked discovering risk status because males more often viewed breast cancer as a ‘woman’s issue.’ Females approached genetic testing focusing more on immediate health first then children’s risk second, and underwent genetic testing more often than males (p<0.0001). Generational differences were found to be factor in the decision whether to undergo BRCA1/2 genetic testing. Individuals who were considered ‘older’ (i.e. over 55 years old as defined by the cited study) felt undergoing genetic testing relied on the input from key male figures, and were considered significantly older than the rest of the sample population (p <0.0001). Younger women generally expressed more willingness to undergo BRCA1/2 genetic
counseling compared to older women. However, in another study conducted with young participants (ages of 15-25) believed they could not receive genetic testing until they were 25 years old, and needed a diagnosis of cancer first before they were eligible for genetic counseling and testing.

**Experiences with cancer in self or family**

Awareness of risk and being recommended to undergo genetic testing were associated with the decision to test. Having a familial cancer experience or a personal history of cancer contributed to the decision of whether to undergo BRCA1/2 genetic testing. Women who either experienced the loss of a mother at a young age from breast cancer, had a premenopausal diagnosis of breast cancer, or viewed that their relatives showed concern reported being more likely to undergo BRCA1/2 genetic testing. A common motivator among several articles in undergoing genetic testing was wanting to know more about their individual and familial cancer risk.

Participants with a personal history of cancer reported feeling either “pressured,” obligated, or responsible to undergo BRCA1/2 genetic testing because of the potential benefit of giving this information to their family members or knowing for themselves. Testing was considered a ‘duty to inform’ relatives and/or for a duty to contribute to the advancement of science research. Those with a personal history of cancer were significantly more likely undergo genetic testing for altruistic reasons (i.e. for their loved ones) compared to those with no personal cancer history. Data suggests that those with a family history of cancer were more likely to receive testing themselves (p < 0.001) when a family member with cancer had a positive BRCA1/2 result.
**FACTORS ASSOCIATED WITH HBOC**

**Informing health decisions:** Genetic testing results can inform cancer screening, prevention, and treatment. Women who never had a cancer diagnosis but were at high familial cancer risk felt that it was better to “know” than “not know” their genetic status; therefore, supported undergoing genetic testing as a way to provide reassurance and preparation for making potential preventative decisions. Several studies explained that participants wanted more risk-related knowledge to help guide their informed health-related decisions such as early detection and prevention options and felt genetic testing would fulfill this need. One study concluded that the availability of having close, follow-up appointments for screening and monitoring practices was associated with being more likely to agree to undergo BRCA1/2 genetic testing. Genetic testing was not always viewed as positive however, and some felt that knowing genetic risk status would not yield any cancer prevention benefit. In one study, treatment focused genetic testing (TFGT) aided participants in providing a sense of certainty in helping to make informed medical decisions. However, some participants felt overwhelmed and anxious in using TFGT, and wanted more time to process their genetic test results before making a surgical decision.

**Anticipatory psychosocial consequences.** Participants reported that genetic testing would help alleviate unwanted negative emotions such as worry, anxiety, uncertainty, and fear and cited this as a reason to undergo genetic testing. Additional reasons included increased feelings of empowerment, personal control, and that undergoing genetic testing felt ‘liberating.’ However, some participants felt negative emotions such as guilt, worry, fear, anxiety, and uncertainty over the possibility of a positive genetic test result and the impact on their family member’s lives as potential carriers, or it reduced the likelihood of getting tested.
FACTORS ASSOCIATED WITH HBOC

Interpersonal factors associated with genetic testing decision: the influence of familial interaction

The unique dynamics of a mother-daughter relationship were found to contribute to the decision whether or not to undergo BRCA1/2 genetic testing.\textsuperscript{11,15,17,20} Mothers with HBOC risk who had daughters were more likely to seek genetic testing due to feeling obligated to ‘do something’ and felt they were protecting their children.\textsuperscript{11,15,17,20} Daughters who learned their mothers were\textit{BRCA1}/2 positive were placed into three categories: wanted testing themselves, ambivalence, or felt obligated. Daughters in the first category used this information to lead their decision-making about getting genetic testing and undergoing screening practices themselves.\textsuperscript{19,34} Other daughters expressed ambivalence toward genetic testing and whether it was the right time in their life.\textsuperscript{34} In some cases, daughters viewed their mother’s request to undergo genetic testing as ‘nagging’ and felt obligated to fulfill their mother’s requests anyways.\textsuperscript{15} This group of daughters feared that if they chose to undergo\textit{BRCA1}/2 genetic testing, then receiving a positive result would be ‘devastating,’ could make them ‘paranoid,’ ‘pessimistic,’ and impact life plans such as child-bearing decisions.\textsuperscript{34} Lastly, individuals who had one or more children were significantly more likely to perceive benefits of genetic testing for altruistic reasons compared to those without children (p =0.013).\textsuperscript{38}

Family communication played a role in the decision to undergo\textit{BRCA1}/2 genetic testing; specifically, the impact of risk from a personal and familial perspective.\textsuperscript{17,20,32} Some participants underwent genetic testing and openly shared their genetic testing results with their family members so as to help them make informed health-related decisions.\textsuperscript{20,22,32,38,39} However, an increase in decisional conflict was seen among probands (i.e. primary person of interest for genetic screening and testing) when undergoing genetic testing themselves.\textsuperscript{32}
FACTORS ASSOCIATED WITH HBOC

Other family-based motivators to genetic testing included child-bearing decisions, future planning such as having children in the future,\textsuperscript{16,20,38} as well as having a partner that insisted.\textsuperscript{15,16} Katapodi (2013) conducted a quantitative study exploring individual and family characteristics associated with \textit{BRCA1/2} genetic testing and found that perceived utility (i.e. perceived benefits and barriers to genetic testing) (OR = 1.97, \( p = 0.009 \)), family hardiness (cohesion and ability to cope with adverse events) (OR =1.29, \( p = 0.04 \)), and perceived severity (OR = 0.52, \( p = 0.02 \)) were positively associated with genetic testing. Some participants were concerned about receiving a positive genetic test result, and explained that this information alluded to fear of passing on a mutation to their children,\textsuperscript{11} having to inform other family members of the results,\textsuperscript{15} and not wanting to be the ‘bearer’ of bad news within the family.\textsuperscript{17,22} Participants were concerned over potential stigma related to testing from how others would perceive them if they were positive for a mutation.\textsuperscript{11,39}

\textbf{Healthcare Systems Factors Associated with Genetic Testing}

\textbf{Role of healthcare providers.} Healthcare provider referral is associated with genetic testing. Discrepancies and inconsistencies in referral played a role in the decision to undergo genetic testing.\textsuperscript{35,36,40,42} In other cases, individuals were more likely to undergo \textit{BRCA1/2} genetic testing when their healthcare provider showed concern for their well-being.\textsuperscript{18} General medical mistrust was seen among several studies, and was associated with less engagement in genetic counseling and testing services.\textsuperscript{21,35,39}

\textbf{Cost of genetic testing.} Participants from several studies stated a potential barrier to receiving genetic testing was taking time off from work,\textsuperscript{34} and if the test would be expensive.\textsuperscript{21,30,34} One study found that those with a higher perceived risk of carrying a mutation, had relatives with breast or ovarian cancer, or were of Ashkenazi descent were more willing to
pay to receive BRCA1/2 genetic testing.35 Some participants felt that paying the full cost for the test was warranted while other participants were more willing to pay if at least part of the genetic test expenses were covered, otherwise they would be unable to afford the test 22. In one study, participants believed referring them to genetic counseling and testing was not necessary and only ordered to make money. 21

Some studies chose to remove the barrier of cost by offering counseling and testing at no charge.11,35 One study’s findings concluded that despite offering genetic counseling and testing services free of charge, 29% of participants were not aware of these services, 43% were aware but not referred by their provider, and 28% either received a referral and/or genetic counseling and testing.35 In Australia, the cost of the consultation with a genetics service is covered by a government subsidy through the national health system, and there is no cost to the patient for genetic testing. Therefore, participants did not have the barrier of cost associated with their decision whether to undergo BRCA1/2 genetic testing as seen in other studies.22

**Impact on insurance coverage.** Another factor found among these articles was the impact of insurance coverage on the decision whether to undergo BRCA1/2 genetic testing. Several studies expressed that participants stated that being tested for the BRCA1/2 mutation could jeopardize their health insurance coverage if they had a positive test result 11,30, and concerns about breach of privacy20,21 and whether their BRCA1/2 genetic test results would stay confidential.11 Some participants stated they did not have health insurance and could not afford the cost of the test themselves.30 Participants in one study stated that differential treatment by their healthcare provider, such as insurance discrimination and/or a negative interaction, impacted their decision whether to seek genetic counseling and testing.21
FACTORS ASSOCIATED WITH HBOC

Cultural influences

There were several cultural differences identified among at-risk Hispanic, African Americans, Asian and Latinas populations in the decision to utilize BRCA1/2 genetic testing. In one study, participants from three different ethnicities (Cubans, Mexican, and Puerto Ricans) were referred to genetic counseling and testing by their healthcare provider due to familial history, and all three groups did not follow through with genetic testing. Cubans chose not to undergo BRCA1/2 genetic testing due to financial hardships in paying for the test, Mexicans reported no follow-up discussions about getting genetic testing from their healthcare provider and chose not to pursue the conversation further, and Puerto Ricans reported fear and uncertainty about genetic testing and wanted more information about what to expect before proceeding forward. Cubans stated that the consideration of genetic testing came with the caveat to review the financial costs beforehand, Mexicans encouraged seeking guidance through prayer, and Puerto Ricans should consider and be aware of any risks involved.

African American participants discussed concerns about potential misuses of genetic information and had a general distrust of medicine and research. Asian participants discussed concerns over the stigma of having ‘bad genes,’ and reported being afraid that disclosing a positive result to prospective suitors and their families would impact their future. Latinas reported a more altruistic perspective and believed in prioritizing their family’s needs first over their own healthcare needs, which consisted of competing demands with their time. One study discussed how participants felt that there was a language barrier that may have impacted their understanding of medical terms and their perceived ability to obtain or receive a referral for genetic counseling and testing.
FACTORS ASSOCIATED WITH HBOC

Discussion

The purpose of this integrative review was to identify decisional-related factors associated with BRCA1/2 genetic testing in individuals at risk for HBOC. We identified several key points associated with genetic testing decision making. First, we learned that making the decision whether to undergo HBOC genetic testing is a complex, dynamic and interdependent decision, one that has family as a central focal point. The decision is multifaceted and is influenced by sociocultural, socioeconomic, and demographic backgrounds. This discussion aims to interpret these findings and discuss next steps.

Much of the literature was descriptive and not based in theory or theoretical frameworks. Theory is important to the advancement of science because it lays the foundation for a well-substantiated explanation with the potential to understand factors which predict outcomes and might improve with intervention. Many HBOC studies have been conducted through a descriptive lens and have evaluated clinically meaningful outcomes. More research studies are needed that are rooted in theory to enhance in-depth understanding of living with hereditary cancer risk so that tailored interventions can be developed and implemented.

Most articles (85%) reported the sample’s personal and/or familial cancer history, which allowed for us to make comparisons about their genetic testing decision-making experiences. Lastly, we compared United States and international articles to identify local and global factors that affect undergoing BRCA1/2 genetic testing. This is important because hereditary risk is not just a domestic discussion, but encompasses all socioeconomic and cultural backgrounds. Understanding hereditary risk within different populations will aid in a more in-depth understanding of risk within each population so that tailored interventions can be
FACTORS ASSOCIATED WITH HBOC

developed and implemented with the goal to reduce global health disparities within hereditary risk genomics research.

The current sample literature was interpreted based on the categories of socioeconomic, sociocultural, familial, and perception of knowledge in HBOC hereditary risk. Individuals with HBOC risk identified several socioeconomic factors specific to the decision whether to utilize genetic testing. This included geographic location, gender, insurance coverage and medical costs associated with utilization of genetic testing. Sample demographics noted the lack of socioeconomic and geographic diversity within the included studies, which made interpretation of how geographic location such as rural versus urban and domestic versus challenging.

Familial influence was another main factor associated with decisions related to BRCA1/2 genetic testing. No articles in this review explored utilization of BRCA1/2 genetic testing from only the at-risk male perspective. Gender disparities exist within cancer genetic testing. Different age and gender roles within a family unit influenced how much or how little involvement certain family members had within that decision-making process. Women are three times more likely to receive cancer genetic testing than men.27 Similar studies have found females take a primary role in decisions surrounding planning, decisions about preventions, and communication of risk to their male relatives.27 A recent study found that men take a more active role in sharing information with other family members;43 however, men were not communicating with daughters or female relatives more often than male relatives, but are communicating with at-risk family members regardless of gender.24,30 Men with HBOC have similar concerns regarding cancer risk for their children and female family members but feel like they have been relegated to “second class status.”43 Current literature does not comprehensively examine the psychosocial, cognitive, and emotional perspectives and perceived level of preparedness in managing care in
FACTORS ASSOCIATED WITH HBOC

men with HBOC. It is important to understand gender differences in communicating HBOC risk as this review has demonstrated that in most cases, this is a complex decision that involves many different support systems. Similar findings of gender disparities and research gaps were seen by Bentley et al. (2017) where there still remains uneven progress in inclusion of males into research studies. Studies that are more inclusive of social and biographical factors will provide a more comprehensive patient perspective of their at-risk experience, which currently is not representative in the literature. Conducting more studies that explore these socioeconomic perspectives in greater detail and how these factors contribute to the decision-making process within HBOC genetic testing is vital in the advancement of science through tailored HBOC intervention-based decisional aids.

Another socioeconomic component highlighted in this review were decisional factors associated with relationship to insurance coverage and associated medical costs. The current cost of BRCA1/2 genetic testing without any insurance coverage depends on the company providing the services. One study examined the cost-effectiveness of germline BRCA1/2 testing in women with breast cancer, and results concluded that genetic testing was cost-effective, and was associated with the reduced risk of cancer and improved overall survival. Another study estimated effectiveness and lifetime costs from a payer perspective for women for hereditary breast cancer, which found that the cost-effectiveness ratio for the 7-gene test strategy compared to the BRCA1/2 test strategy was $42,067 per life-year gained or $69,920 per quality-adjusted life year gained for the 50-year old cohort. Now that multi-gene panel testing is considered the standard of care and proven to be cost-effective, more emphasis is needed to review insurance coverage policies and patient’s ability to receive the recommended genetic testing panel by their healthcare team.
FACTORS ASSOCIATED WITH HBOC

Sociocultural influenced decisions surrounding \textit{BRCA1/2} genetic testing. Findings from this review highlighted several relationships within and among different ethnic groups, such as Latino, Asian, and Black and factors associated within the context of culture and family in the genetic testing decision-making. Family involvement within the context of cultural beliefs was a key component that impacted 1) whether individuals at risk received medical advice and was referred for \textit{BRCA1/2} genetic testing, 2) how family members communicated risk to others, such as which family members were considered ‘gatekeepers’ of this risk information, and 3) their perspective of the genetic testing decision making process. Despite these results, comparison within each cultural group and comparison across cultural groups were conducted on a small scale, and did not capture other ethnic and cultural backgrounds from the individual and familial HBOC perspective. According to Hawley & Morris (2017), healthcare professionals need to offer more recommendations in a culturally competent manner that best involve patients early in the decision-making, and that this process is most meaningful to them. The role of cultural competency in decision-making should engage the patients to be more well-informed and share the role of decision-making with the loved ones and providers.\textsuperscript{47}

Lastly, participants perceived level of knowledge in understanding hereditary risk, inheritance patterns, and what medical recommendations were considered standards of care were all factors associated with \textit{BRCA1/2} genetic testing. Several articles pinpointed misunderstandings or communication challenges participants faced when making these genetic testing decisions. Other literature has identified discrepancies of information within the communication process such as mistrust of the medical system, fear of results, lack of referral by provider, or not wanting to discuss hereditary risk from various sources such as medical providers, family members, friends, and social media.\textsuperscript{48,49} This is consistent with our findings
FACTORS ASSOCIATED WITH HBOC

that there are varying degrees of understanding complex information within the genetic
counseling and testing process, and more individualized tailored and developmentally-sensitive
materials about genetic counseling and testing services are needed. Additionally, more studies
are needed to review the process of patient and provider initiated hereditary risk discussions
including the PCP referral process for those at risk for HBOC.

Practice & Policy Implications & Recommendations

Practice implications: Several barriers exist among family members considering BRCA1/2
genetic testing, ambivalence towards the medical community, lack of understanding in genetic
counseling and testing process and results, uncertainty, fear, and doubt in a positive result, and
potential strain on family relationships and communication difficulties. Health care providers
should be aware of the current NCCN guidelines that outlines an individual’s personal and
familial cancer risk, and provide referrals for genetic counseling and subsequently testing if they
meet the screening criteria. Healthcare providers should also have available resources such as
handouts of local centers that help patients with hereditary cancers and/or at-risk families
navigate through this process so they feel supported and connected to the community.

Policy implications: More studies are needed to explore cost-effective and individualized
measures of care. Current U.S. guidelines outline specific eligibility criteria in determining a
person’s cancer risk status, thus providing genetic testing to determine mutation status. However,
insurance companies currently have the right to deny treatment such as early mammograms,
genetic testing coverage, and certain reconstructive surgeries if they feel that they don’t meet
certain guidelines. More research is needed in this area to develop standardized, compliant
policies that accurately reflects the current state of patients living with hereditary cancers as well
as their at-risk family members.
FACTORS ASSOCIATED WITH HBOC

Research Implications: Based on these outcomes, more theoretically-based studies are needed focusing on the decision-making process within a family context. We also need to conduct more studies that explore the perspectives of other races, cultures, and geographic locations that contribute to the decision-making process in genetic testing. Additionally, the experience of males making HBOC related risk assessment decisions within a family should be further explored.

Summary

Based on these outcomes, this review had led to the conclusion that BRCA1/2 genetic testing should be tailored to the individual and family’s perspectives. Individuals with personal and/or familial history of cancer have an increased chance of developing cancer later in life, especially if there are noted hereditary factors. At risk individuals who learn of their cancer risk status through multi-gene panel testing can help tailor their future health-related decisions when discussing screening and prevention options. Continuing to explore this topic and how an individual makes these complex decisions has the potential to generate data needed to develop interventions and enhance clinical care to support decision-making strategies for at-risk individuals when deciding to undergo genetic testing. As multi-gene panel testing is now considered the standard of care, the impact of obtaining information on more moderate risk genes or obtaining variants on family decision making should be further explored.
References


FACTORS ASSOCIATED WITH HBOC

   January 9th, 2017

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    and attitudes about BRCA1/2 testing among women of African descent with family 

12. Livraghi L, Garber JE. PARP inhibitors in the management of breast cancer: current date 
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13. Mahon SM. Factors affecting genetic testing and decisions about prophylactic 
    surgery...commentary on Meijers-Heijboer EJ, Verhoog LC, Brekelmans CTM, Seynaeve 
    C, Tilanus-Linthorst MMA, Wagner A et al. (2000) Presymptomatic DNA testing and 
    prophylactic surgery in families with a BRCA1 or BRCA2 mutation. Lancet, 355, 2015- 
    Long-term satisfaction and psychological and social function following bilateral 
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FACTORS ASSOCIATED WITH HBOC


FACTORS ASSOCIATED WITH HBOC


FACTORS ASSOCIATED WITH HBOC


FACTORS ASSOCIATED WITH HBOC


Appendix

Figure 1. Study Selection Process

Keywords and index term searches in PsychInfo, PubMed, CINAHL, and EBSCOhost. Study reports for title & abstract screening: N= 831

Study reports excluded after title and abstract review. N=477

Study reports retrieved for full evaluation. N=354

Study reports excluded due to failure to meet inclusion criteria. N=334

Total studies included: N= 20

Table 1: Characteristics in Included Studies
### Characteristics in Included Studies

<table>
<thead>
<tr>
<th>Characteristics in Included Studies</th>
<th>Number of articles (N=20)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Study Design</strong></td>
<td></td>
</tr>
<tr>
<td>Qualitative</td>
<td>8 (40%)</td>
</tr>
<tr>
<td>Quantitative</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>Mixed Methods</td>
<td>3 (15%)</td>
</tr>
<tr>
<td><strong>Geographic Location</strong></td>
<td></td>
</tr>
<tr>
<td>United States</td>
<td>14 (70%)</td>
</tr>
<tr>
<td>International</td>
<td>5 (25%)</td>
</tr>
<tr>
<td>Domestic &amp; International collaboration</td>
<td>1 (5%)</td>
</tr>
<tr>
<td><strong>Sample Size (Range)</strong></td>
<td></td>
</tr>
<tr>
<td>Quantitative</td>
<td>50-1,574</td>
</tr>
<tr>
<td>Qualitative</td>
<td>17-54</td>
</tr>
<tr>
<td>Mixed Methods</td>
<td>40-120</td>
</tr>
<tr>
<td>Mean Age</td>
<td>45 years old</td>
</tr>
<tr>
<td><strong>Race/Ethnicity</strong></td>
<td></td>
</tr>
<tr>
<td>* Black</td>
<td>4 (20%)</td>
</tr>
<tr>
<td>Caucasian</td>
<td>6 (30%)</td>
</tr>
<tr>
<td>Hispanic</td>
<td>2 (10%)</td>
</tr>
<tr>
<td>Not Reported</td>
<td>7 (35%)</td>
</tr>
<tr>
<td>** Reported multiple races/ethnicities</td>
<td>1 (5%)</td>
</tr>
<tr>
<td><strong>Gender</strong></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>14 (70%)</td>
</tr>
<tr>
<td>Male</td>
<td>0</td>
</tr>
<tr>
<td>Male &amp; Female</td>
<td>6 (30%)</td>
</tr>
<tr>
<td><strong>BRCA Status</strong></td>
<td></td>
</tr>
<tr>
<td>***Results of BRCA1/2 genetic testing</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>Not conducted as eligibility requirement</td>
<td>1 (5%)</td>
</tr>
<tr>
<td>Not Reported</td>
<td>9 (45%)</td>
</tr>
<tr>
<td>Underwent GT but results not reported</td>
<td>1 (5%)</td>
</tr>
<tr>
<td><strong>Personal History of Cancer</strong></td>
<td></td>
</tr>
<tr>
<td>100% of sample Unaffected</td>
<td>1 (5%)</td>
</tr>
<tr>
<td>100% of sample Affected</td>
<td>3 (15%)</td>
</tr>
<tr>
<td>Mix of Unaffected &amp; Affected</td>
<td>13 (65%)</td>
</tr>
<tr>
<td>Not Reported</td>
<td>3 (15%)</td>
</tr>
</tbody>
</table>

* "Black" includes African, African American, West/Indian, and Caribbean descent.
** Several articles combined race and ethnicity together in results section.
*** Results of BRCA1/2 genetic test indicated by positive, negative, or inconclusive result.
### Table 2: Summary of Data Abstraction – Quantitative Literature

<table>
<thead>
<tr>
<th>Author (year,country)</th>
<th>Topic/Aim</th>
<th>Sample Characteristics</th>
<th>Theory &amp; Method</th>
<th>Main Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cragun et al. (2015, USA)</td>
<td>To evaluate the prevalence of and factors associated with referral to genetic counseling and access to genetic services.</td>
<td>Sample = 440 Female (≤50 years old) diagnosed with invasive breast cancer</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Mean age at cancer diagnosis: 42.1</td>
<td>No theory reported</td>
<td>49% (216/440) were not referred to GC services by their providers nor accessed genetic services.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Self-identified Black</td>
<td>Descriptive</td>
<td>Providers recommending GC counseling was associated with participants with college education or diagnosis &lt;45 years old or Triple Negative breast cancer diagnosis.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>36% (159/440) underwent genetic testing</td>
<td>Cross-sectional</td>
<td>Those with private insurance, household income &gt;$35,000 and diagnosed before age 45 were more likely to access genetic services.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>BRCA status not reported for those that underwent genetic testing</td>
<td></td>
<td>(36%) Black breast cancer survivors had genetic testing.</td>
</tr>
<tr>
<td>Denayer et al. (2009, Belgium)</td>
<td>To compare at risk individuals based on their gender, BRCA status, psychological profile, and motivation for predictive testing among adult offspring.</td>
<td>Sample =292 Female: 233 Male: 59 Subgroup: 180 different sibships= (87) BRCA1, (93) BRCA2</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Mean age:</td>
<td>No theory reported</td>
<td>Breast cancer related distress was significantly less for men than women.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Female: 36.9</td>
<td>Prospective</td>
<td>No difference between BRCA1 and BRCA2 families with IES – avoidance and intrusion subscale.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Male: 44</td>
<td>T1: baseline</td>
<td>‘Social support seeking’ widely used by men and women as a coping mechanism.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Age range:</td>
<td>T2: 1 year follow-up</td>
<td>Motivation: Early detection for breast cancer &amp; knowing children’s risk was significant.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Female: 18-74</td>
<td></td>
<td>BRCA testing more prevalent in females from BRCA2 families versus BRCA1 families.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Male: 18-77</td>
<td></td>
<td>Uptake of genetic testing higher in females (82%) than males (32%).</td>
</tr>
</tbody>
</table>
## FACTORS ASSOCIATED WITH HBOC

<table>
<thead>
<tr>
<th>Author (year,country)</th>
<th>Topic/Aim</th>
<th>Sample Characteristics</th>
<th>Theory &amp; Method</th>
<th>Main Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>From BRCA2 family:</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Female: 57%</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>• Male: 53%</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Personal history of cancer not reported</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Edwards et al. (2008, USA)</td>
<td>To explore the association between temporal orientation (i.e. past, present, future implications) and BRCA testing attitudes.</td>
<td>Sample = 140 Female (94) affected by personal history of breast cancer Mean age: 45.6 Age range: 22-79 African descent (74) Black/African American (58) Black/ West Indian/ Caribbean BRCA status not reported</td>
<td>No theory reported Descriptive Cross-sectional</td>
<td>Age was considered a significant factor on temporal orientation - older: present orientation - younger: future orientation Those with a personal history of cancer had a higher present-orientation compared to those without a personal history of breast cancer. Those with future orientation believed testing increased their personal control, and was positively associated with family related cons of testing (p =0.001) <strong>Perceived Pros</strong> Knowledge for other family members, help learn risk of children, motivation for surveillance, concerns about getting cancer reduced, increase sense of personal control, help make important life decisions. <strong>Perceived Cons</strong> Negative effect on family, passing gene on, guilt, stigma (singled out), ashamed, fear, hopeless/despair, emotionally difficult, wouldn’t stay confidential, jeopardize insurance coverage (11%).</td>
</tr>
<tr>
<td>Hurtado-de-Mendoza et al. (2015, USA)</td>
<td>To examine factors associated with HBOC knowledge, and assess the impact of knowledge on genetic screening</td>
<td>Sample=50 Female, age 21 or older Self-identified Black</td>
<td>No theory identified Descriptive Cross-sectional</td>
<td>48% (24/50) of survivors were referred to or used GCT services. 14% unaware of GCT</td>
</tr>
</tbody>
</table>
### Factors Associated with HBOC

<table>
<thead>
<tr>
<th>Author (year,country)</th>
<th>Topic/Aim</th>
<th>Sample Characteristics</th>
<th>Theory &amp; Method</th>
<th>Main Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>counseling and testing (GCT) engagement in a sample of Black survivors at risk for HBOC.</td>
<td>98% had diagnosis of breast cancer Mean Age: 51.15 BRCA status not reported Recruited from community-based and hospital setting Setting and procedures of study reported in Sheppard, 2013.</td>
<td>Stress &amp; Coping Theory Decisional-Conflict Theory Family Systems in Genetic Illness Framework Correlational, cross-sectional cohort study Mailed questionnaires</td>
<td>38% had been referred to GCT despite of being aware about GCT. Lower age and lower stage was associated with higher knowledge. Higher knowledge and higher self-efficacy were significantly associated with GCT engagement. 56% of survivors reported not being at all or rarely worried about developing breast/ovarian cancer again. Perceived self-efficacy to engage in GCT was relatively high as well as perceived cancer-related stress, and medical mistrust.</td>
</tr>
<tr>
<td>Katapodi et al. (2011, USA)</td>
<td>To examine difference and correlations in appraisals of HBOC, psychological distress, family environment, and decisional conflict between women who pursued genetic testing and their at-risk relatives who did not.</td>
<td>Sample = 372 Female 200 probands and 172 relatives (86%) probands affected by cancer (21%) of relatives affected by cancer Mean age: • Probands: 51 • Relatives: 48 Age range: • Probands: 22-83 • Relatives: 18-81 95% Caucasian BRCA Status</td>
<td>Stress &amp; Coping Theory Decisional-Conflict Theory Family Systems in Genetic Illness Framework Correlational, cross-sectional cohort study Mailed questionnaires</td>
<td>Probands Greater knowledge of risk factors of gene inheritance, and greater decisional conflict regarding genetic testing, which correlated with lower perceived risk and better family communication. Perceived controllability was related to lower perceived risk. Relatives Had perceived controllability was related to lower distress in relatives. Higher decisional conflict was associated with less knowledge and less perceived severity.</td>
</tr>
<tr>
<td>Author (year,country)</td>
<td>Topic/Aim</td>
<td>Sample Characteristics</td>
<td>Theory &amp; Method</td>
<td>Main Findings</td>
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| Katapodi et al. (2013, USA) | 1) To examine whether individual and family characteristics have a direct effect on women's decisions to use genetic testing for hereditary breast cancer | *Probands: 25  
  • Known in other family members: 21  
  *Relatives: -  
  • Known in other family members: 23 | Stress & Coping Theory  
  Family Systems in Genetic Illness Framework  
  Decisional-Conflict Theory | Positive association with genetic testing were education (p=0.01), personal history of cancer (P<0.0001), perceived cause (p<0.0001), psychological distress (p=0.03), perceived utility of genetic testing (p<0.0001), and family hardiness (p=0.02). Negative association with genetic testing were perceived controllability (p<0.04) and perceived severity (p<0.0001). Those with a personal history of cancer was associated with genetic testing than those without a personal history of cancer (OR= 4.76, p = 0.009). Genetic testing was positively associated with higher family hardiness (p=0.04). There was a negative association between genetic testing and perceiving negative consequences of breast cancer. |
|                        | 2) To explore whether family characteristics moderate the relationships between individual characteristics and the decision to use genetic testing. | Sample= 372  
  Female  
  168 probands, and 168 relatives (86%) probands affected by cancer compared with (21%) of relatives affected by cancer (p <0.05)  
  52% first-degree relatives  
  Age range:  
  • Probands: 22-83  
  • Relatives: 18-81  
  95% Caucasian  
  BRCA Status  
  • Probands: 25  
  • Relatives: - | Descriptive  
  Cross-sectional, cohort study  
  Mailed questionnaires | |
| Lynch et al. (2009, USA) | To determine the effects of family information service (FIS) attendance on extended family members from HBOC families with BRCA1/2 | Sample= 1574  
  Male & Female  
  Extended family members (from 60 HBOC syndrome families where BRCA1 or BRCA2 had been identified)  
  2 cohorts: those that attended FIS and those | Retrospective  
  Descriptive  
  Educational materials | No significant difference in age among FIS attendees between those that underwent genetic testing and those that didn’t. But those that were not part of FIS were significantly older who underwent genetic testing (p<0.0001). In both cohorts, females were more likely to have undergone genetic testing than males (p <0.001). |
<table>
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<th>Main Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>Miron-Shatz et al. (2015, USA)</td>
<td>To examine predictors of the decision to undergo cancer related genetic testing exploring perceived risk, family risk of breast or ovarian cancer, and numeracy as predictors to test among women at high risk for breast cancer.</td>
<td>Mean Age: Female = 49.9-53.9 Male =49.6-55.3 Race/Ethnicity not reported BRCA status not reported Personal history of breast/ovarian cancer: N=133</td>
<td>No theory identified</td>
<td>70% (N=315) had not previously tested for BRCA1/2 genetic mutation. Those with a family history of breast or ovarian cancer or a family member who tested positive for the BRCA1/2 mutation, of Ashkenazi Jewish descent, and young age were more likely to receive genetic testing. Willingness to pay (for test) was significantly correlated with perceived risk of having a mutation, number of relatives with breast or ovarian cancer, and Ashkenazi descent.</td>
</tr>
<tr>
<td>Sheppard et al. (2013, USA)</td>
<td>To identify socio-cultural influences associated with BRCA1/2 genetic counseling and testing (GC/T) engagement in Black women.</td>
<td>Sample=100 Female Self-identified Black women N=50 unaffected with at least 1 first-degree relative</td>
<td>Framework proposed by Freeman and Chu</td>
<td>Greater GC/T engagement was associated with older age, greater educational attainment, higher household income, greater GC/T self-efficacy, and less medical mistrust (p &lt;0.10). Findings from this study confirm that Black women’s awareness of GC/T may not translate into actual GC/T engagement.</td>
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### Table 3: Summary of Data Abstraction – Qualitative Literature

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<tr>
<th>Author (year, country)</th>
<th>Topic/Aim</th>
<th>Sample Characteristics</th>
<th>Theory &amp; Method</th>
<th>Main Findings</th>
</tr>
</thead>
</table>
| Dancyger et al. (2010, England) | To investigate the attitudes and motivations of both index patients and their relatives towards genetic testing and the influence of family communication and relationships upon their motivations. | Sample= 30
10 index patients (F) with either breast/ovarian cancer and a positive *BRCA1/2* mutation with 2 of their relatives (5M & 15F) | No theory reported | 2 distinct groupings: either showed strong commitment to, and motivations for genetic testing (GT) or families uncertain about GT |
| | | *Index patient:* (6) BRCA1 (4) BRCA2 | Interpretative Phenomenological Analysis (IPA) | 3 subthemes to strong group: obligation to be tested; not fully thought through; testing for oneself |
| | | *Relative:* (11) Untested (3) BRCA positive (4) Negative (2) Pending results | Semi-structured interviews | |
| | | | | |
| | | | | |
### FACTORS ASSOCIATED WITH HBOC

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</table>
| Etchegary et al. (2009, Canada) | To present a framework of dimensions of responsibility as they relate to genetic risk for inherited cancer, specifically HBOC and HNPCC. | A total of 15 female & 5 male relatives (total of 10 families)  
Mean age: 43.6  
Age Range: 20-71  
Race/Ethnicity Not Reported | No theory reported  
IPA  
Semi-structured Interviews  
Thematic Clusters | 3 themes about the dimensions of genetic testing:  
1. To know about the self for self  
2. To know about the self for others.  
3. To know about the self to oblige others to know |
| Glenn et al. (2012, USA) | To assess awareness of genetic testing for breast cancer risk and identify influences on the decision-making process regarding counseling and testing among an ethnically-diverse sample of women. | Sample = 33  
Female  
Breast/ovarian cancer survivors or first-degree relatives of survivors  
Mean age: 51.9  
52% Asian  
24% African American | No theory reported  
Open-ended semi-structured interviews | Beliefs about Risk Factors  
- Half of sample cited stress as a risk factor, most were Asian and Latina  
**Awareness of Genetic Testing**  
- 11/32 women heard of genetic counseling and testing (GC/T) before study, majority who hadn’t heard were of ethnic background.  
- Awareness did not differ between breast cancer survivors and first-degree relatives. |
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</tr>
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<tbody>
<tr>
<td>Hamilton et al. (2009, USA &amp; Canada)</td>
<td>To explore the influences on decision-making in women who tested positive for the BRCA1/2 mutation</td>
<td>Sample = 44 Female 21 diagnosed with breast cancer, 23 no history of cancer 22 states (41 participants) &amp; Canada (3 participants) Age range: 18-40 43 White European 1 African American BRCA1: 30 BRCA2: 15 (one participant affected by both)</td>
<td>Theory of Genetic Vulnerability (Hamilton &amp; Bowers 2007) Grounded Theory Method Focus: on women who tested positive for the BRCA1/2 mutation Family Systems Genetic Illness Model</td>
<td>Identified 4 life trajectories: 1. Acutely Aware: Long-standing awareness of breast cancer in the family. 2. Loss of Mother to breast cancer at a young age. 3. Expression of concern by a health care provider. 4. Personal diagnosis of breast cancer (premenopausal)</td>
</tr>
<tr>
<td>Norris et al. (2009, USA)</td>
<td>(1) To develop an inter-generational perspective on families’ stories to highlight aspects</td>
<td>Sample = 17 Index patient = 5 mothers Participating relatives = 3 husbands, 6 daughters, 3 sons</td>
<td>No theory reported Qualitative design described by</td>
<td>10 themes identified: 1. Women are gatekeepers of HBOC risk information. 2. Women may spare others from anxiety/depression. 3. Men refrain from HBOC discussions</td>
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<tr>
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<td>of the family context as influences on communication of risk and decision making about testing and follow-up.</td>
<td>A total of 5 families</td>
<td>Sandelowski (2000)</td>
<td>4. Various approaches to sharing information</td>
<td></td>
</tr>
<tr>
<td>(2) To establish the feasibility of examining the influences of family experiences on members’ adaptations and responses to awareness of hereditary cancer risk.</td>
<td>Age range of child: 15-25</td>
<td>Naturalistic inquiry methods</td>
<td>5. Daughters follow mother’s lead in decision-making</td>
<td></td>
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<tr>
<td></td>
<td>Age range of adults not reported</td>
<td>Purposive sampling: open-ended; content analysis</td>
<td>6. Children learn cancer risk informally from parents</td>
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<td></td>
<td>100% Caucasian</td>
<td></td>
<td>7. Expectations of BRCA1/2 genetic testing.</td>
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<tr>
<td></td>
<td>BRCA status not reported</td>
<td></td>
<td>8. Promotion of early detection and prevention.</td>
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<tr>
<td></td>
<td>Personal history of cancer not reported</td>
<td></td>
<td>9. Unknowingly at risk</td>
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<td></td>
<td></td>
<td></td>
<td>10. Ongoing assessment and support are needed</td>
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<tr>
<td>Proulx et al. (2009, Canada)</td>
<td>To develop a detailed understanding of the experiences and decisions that motivate women with increased risk of hereditary breast cancer to participate in a Quebec experimental breast screening program.</td>
<td>Sample = 21 Female</td>
<td>No theory reported</td>
<td>- Women’s living with the experience of breast cancer find it an emotionally charged subject.</td>
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<tr>
<td></td>
<td>Age range: 24-67</td>
<td>Used framework by Rees et al. (2001)</td>
<td>- Genetic screening and testing was considered a family affair</td>
<td></td>
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<tr>
<td></td>
<td>Race/Ethnicity not reported</td>
<td>Purposive sampling</td>
<td>- The decision to undergo GT was largely based on resources to follow-up.</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Multi-centered Quebec experimental breast screening program</td>
<td>Semi-structured interviews</td>
<td>- Differences among the decision to undergo GT based on a personal history of cancer.</td>
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<td></td>
<td>7 had a previous diagnosis of breast cancer and decided to get genetic testing. 8 unaffected with cancer underwent testing and were carriers. 6 unaffected with cancer declined testing.</td>
<td></td>
<td>- Women unaffected with cancer emphasized they preferred knowing over not knowing their genetic status.</td>
<td></td>
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<td></td>
<td></td>
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<td>- Those that declined GT was to ‘protect themselves’ emotionally.</td>
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## FACTORS ASSOCIATED WITH HBOC

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<tbody>
<tr>
<td>Sussner et al. (2015, USA)</td>
<td>To examine beliefs and attitudes about BRCA genetic counseling for HBOC among diverse, at risk women (both affected and unaffected) who have not previously undergone counseling.</td>
<td>Sample = 54 Female, at least 18 years old Self-identified Latina women 60.5% had a personal history of breast/ovarian cancer 46.5% had a family history of breast/ovarian cancer 76.7% had children Mean Age: 49.9 12 focus groups + 30 in-depth interviews BRCA status not reported</td>
<td>Integrative Model of Behavior Prediction From HBM, SCT, and Theory of Reasoned Action Thematic content analysis</td>
<td>Themes 1) Illness Prevention 2) Personal &amp; Community Knowledge about BRCA Genetic counseling 3) Perceived Benefits and Barriers of BRCA Genetic Counseling and Cultural Influences on Genetic Counseling Participation 4) Influence of Previous Interactions with the Healthcare System Suggestions for Educational Tool about BRCA Genetic Counseling for HBOC</td>
</tr>
<tr>
<td>Zilliacus et al. (2012, Australia)</td>
<td>To explore the actual experience of women who were eligible for and had treatment-focused genetic testing (TFGT) during their treatment process, as well as the hypothetical views towards TFGT of women who had been diagnosed with breast cancer, but had not had TFGT</td>
<td>Sample = 26 (all female) Mean age: 42 2 groups Age range: 18-50 Group 1 14 had breast cancer, not had TFGT Group 2 12 had breast cancer, not had TFGT 2 BRCA positive, 12 inconclusive results Race/Ethnicity not reported</td>
<td>No theory reported Conceptual framework of Miles and Huberman Semi-structured telephone interviews</td>
<td>Women expressed positive attitudes towards TFGT and felt it was highly relevant to their surgical decision making. They did not feel that an offer of TFGT shortly after, or at the time of diagnosis, added undue psychological burden. Majority of women interviewed felt that TFGT should be incorporated into standard clinical care.</td>
</tr>
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</table>
## Table 4: Summary of Data Abstraction – Mixed Methods Literature

<table>
<thead>
<tr>
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<th>Sample Characteristics</th>
<th>Theory &amp; Methods</th>
<th>Main Findings</th>
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<tbody>
<tr>
<td>Garg et al. (2015, USA)</td>
<td>To conduct an exploratory, longitudinal study by testing perceived benefits of genetic testing among men and women at elevated risk for BRCA1/2 mutations over 3 key time points in genetic counseling and testing process to understand altruistic motivations to test.</td>
<td>Sample = 120 Female: 89.2% (70 families) Mean Age: 3 Categories ranged from 47.5 to 54.9 Age Range: 18-83 Race/Ethnicity not reported Ashkenazi Jewish descent 89.2% underwent BRCA genetic testing • 68.2% uninformative negative • 22.4% positive • 9.4% informative negative 47.5% had personal history of cancer</td>
<td>Hamilton’s Kin Selection Theory Narrative Analysis guided by principles from Labov Immersion/ Crystallization method Prospective T1: before genetic testing T2: One-two days after counseling T3: 6 months after receiving genetic testing results</td>
<td>6 Themes Personal Motivation: 1) Cancer prevention (64.3%) 2) Acquire knowledge (42%) 3) To alleviate anxiety (16.1%) 4) For future planning (5.4%) Altruistic motivation: 5) For family’s sake (25%) 6) For science and research (13.4%)</td>
</tr>
<tr>
<td>Patenaude et al. (2013, USA)</td>
<td>To determine what daughters of BRCA1/2 mutation carriers understand about their 50% cancer risk status and about risk reduction or management options, understand the extent and nature of cancer-related distress, and the effect of knowing their BRCA1/2 results.</td>
<td>Sample = 40 Female 53 mothers who tested positive for BRCA provided daughters information to contact Median Age: 21 Age Range: 18-24</td>
<td>Methods of Weiss (1994) Telephone interviews Demographic and family questionnaires Thematic Analysis</td>
<td>Almost all daughters were made aware of their mother’s BRCA1/2 results. Misconceptions in age and criteria to receive BRCA1/2 genetic counseling and testing. Significant gaps and lack in knowledge regarding cancer risk. Cancer-related distress was higher than general distress Cancer worries about results and the effect on future planning</td>
</tr>
</tbody>
</table>
FACTORS ASSOCIATED WITH HBOC

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<td>mother’s mutation status on their future plans.</td>
<td>100% White/Non-Hispanic or Latino</td>
<td>BRCA status not reported</td>
<td>Social Marketing</td>
<td>-There were a variety of different genetic testing intentions ranging from wanting genetic testing to thinking the information would be more detrimental.</td>
</tr>
<tr>
<td>Vadaparampil et al. (2010, USA)</td>
<td>To explore awareness of risk factors for hereditary breast and ovarian cancer, awareness, knowledge and concerns about genetic testing, and preference for how to have genetic testing recommended by a healthcare provider among at-risk Hispanic women.</td>
<td>Sample = 53 Female Mean Age: Not Reported Age Range: In sum of the 3 ethnic groups: &lt;34: N=13 35-44: N=17 45-50: N=12 51-65 N=11 Hispanic 32% Cuban 30% Mexican 37.7% Puerto Rican BRCA status not reported Personal history of breast cancer before age of 50: N=11 Personal history of ovarian cancer: N=2</td>
<td>Grounded Theory Strauss &amp; Corbin (1990) Cross-sectional Semi-structured interviews</td>
<td>Across the 3 groups: - Cited three primary risk factors: family history, lifestyle choices, and sporadic use of healthcare - Had limited or no knowledge of genetic testing (GT). - Had concerns about the impact of a positive test result of themselves and their families - Majority of women stated a healthcare professional did not suggest GT to them. Based on Ethnic background: 1. Cuban: All reported did not pursue GT for financial reasons. 2. Mexican: No follow-up by healthcare provider. 3. Puerto Rican: Had fear/uncertainty about GT &amp; would want more information.</td>
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</table>
FACTORS ASSOCIATED WITH HBOC
III. Manuscript 2
Understanding use of Mid-Range Health Behavior Theories & Models among Individuals at risk for Hereditary Breast and Ovarian Cancer Syndrome (HBOC)

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Abstract

**Background:** An integrative review of the literature was previously conducted to identify decisional factors related to whether individuals at-risk for hereditary breast and ovarian cancer syndrome (HBOC) utilize *BRCA1/2* genetic testing. Findings from this review indicated the need for more theoretical frameworks and models in this population. This has the potential to further understand of how individuals at risk for HBOC make genetic testing related-decisions in the context of mid-range theories and theoretical frameworks.

**Methods:** Five articles from the integrative review identified several mid-range theories and theoretical frameworks that explored individuals at-risk for HBOC and their decision whether to utilize *BRCA1/2* genetic testing.

**Results:** Two articles were quantitative, two were qualitative, and one article was a mixed-methods study. These five articles utilized six theories, three models, fifteen concepts, six constructs, and five methods. The sample population was similar across all five articles while the purpose of each study and the concepts and constructs explored varied.

**Conclusions:** What’s been highlighted in this theoretical review of the literature among individuals at risk for HBOC is there is no one theory that is a ‘one size fits all’ when discussing a population with increased hereditary cancer risk. Therefore, understanding the complexities of how mid-range theories and frameworks influence the decision-making process of individuals with HBOC during genetic testing is crucial in advancing our understanding of how decisions are made so that we can tailor individualized interventions such as decisional aids among this population.

**Keywords:** Theory, hereditary breast and ovarian cancer syndrome, HBOC, genetic testing
Introduction

The purpose of this paper is to provide an overview of mid-range theories and models used to understand Hereditary Breast and Ovarian Cancer Syndrome (HBOC) and to propose theoretical recommendations for future HBOC studies that include individuals living with hereditary cancer risk and their decision-making process related to genetic-testing. The integrated review included an overview of the most current literature (beginning with 2008-2018), a description of documented frameworks, and concludes with an in-depth description of the integrated framework used to guide decision-making related research and practice among individuals with hereditary cancer risk. The integrative review of the literature identified selected decision-making factors and was conducted by the listed authors that identified key decisional factors associated with the decision whether to utilize genetic testing. The findings have the potential to further understanding of how individuals at risk for HBOC make BRCA1/2 genetic testing related-decisions in the context of theoretical frameworks. Below is an in-depth exploration of the five included theories and frameworks, how they defined and operationalized their constructs, and their relationship to each other within this phenomena of interest.

Theories, Framework & Models utilized in HBOC Genetic Testing

Five articles that explored individuals at-risk for hereditary breast and ovarian cancer syndrome and their decision whether or not to undergo BRCA1/2 genetic counseling and testing included (6) theories, (3) models, (15) concepts, (6) constructs, and (5) methods. The sample population was similar across all five articles while the purpose of each study and the concepts and constructs explored were varied. Two articles were quantitative,1,2 two were qualitative,3,4 and one article was a mixed-methods study. 5
Stress & Coping Theory

The Stress & Coping Theory by Lazarus & Folkman (1984) highlighted the importance of stress as a dynamic process. This theory, originally referred to as the Transactional Theory of Stress and Coping (TTSC), presented stress as a product of a transaction between a person and his/her complex environment. The concept of stress was further developed into different categories: event, situation, cue, and condition. This was refined from Lazarus (1966) book titled, *Psychological Stress and Coping Process*, which discussed the integration of previous research on stress and combined additional concepts of health and coping together. This shifted the focus to the center of the individual’s appraisal of stress and its relationship to the stress experience.

The article by Katapodi et al. (2011; 2013) explained the first concept of primary appraisal or ‘illness appraisal of HBOC’ as the initial response to learning about a person’s hereditary cancer risk, and the associated level of threat given this information. The second concept, referred to as the ‘secondary appraisal,’ which included two constructs (psychological distress and family characteristics) followed this primary appraisal stage. The secondary appraisal stage examined the level of psychological distress caused by the primary appraisal, and the level of threat and availability of coping resources. These concepts were operationalized through use of several measurement scales (knowledge of modes of gene inheritance, subscale of revised illness perception questionnaire, subscale of IPQ-R, Family Problem Solving and Communication index, Family Relationships inventory, a subscale of family environment scale, and decisional conflict scale) that were validated with high Cronbach alphas. The use of this theory proposed that the primary appraisals of a disease would determine whether the disease was viewed as a threat to one’s well-being. Secondly, the individual’s emotional response led to
the secondary appraisals of the ability to cope with associated problems and emotions. However, the authors did not feel this one theory would adequately cover their proposed research question, and chose to integrate two more theories to adapt a new integrated, more comprehensive theoretical framework.

**Family Systems in Genetic Illness Framework (FSGI)**

The FSGI is a comprehensive model that organizes complex interactions between genetics and other factors placing individuals at-risk for disease, significant family and social variables, and lastly professionals involved in care. This model includes the time prior to clinical diagnosis as well as potential influences of genetic information on family systems. Katapodi et al. (2011; 2013) integrated the FSGI due to the dynamics of the family environment in the context of genetic disease. The FSGI model incorporated family system variables, such as family relationships and communication, which were concepts explored by the authors.

**Decisional Conflict Theory**

This general descriptive theory of decision-making under stress proposed by Janis & Mann (1977) are particularly interested in the interplay of associated factors that influence how people make decisions when confronted with decisional conflict. More importantly, how individuals make decisions when in conflicted or a high-stakes environment where the outcome could pose personal and/or familial struggles. However, the concept of decision-making was not included within the Stress & Coping Theory and the FSGI model; therefore, Katapodi et al. (2011; 2013) integrated the Decisional Conflict Theory by Janis & Mann (1977). In the Katapodi et al. (2011; 2013) articles, this theory allowed for the examination of the individual and familial context of HBOC and its influence on decision-making about genetic testing. Thus, the integration of these two theories and one model allowed for the examination of family
characteristics that influenced decisions for BRCA1/2 genetic testing and interactions between individual and family characteristics, as well as compared women who pursued genetic testing for HBOC and their at-risk relatives who did not pursue testing.

**Hamilton’s Kin Selection Theory**

Hamilton’s Kin Selection Theory, sometimes referred to as ‘inclusive fitness theory,’ which predicts that altruistic action will be favored when the benefit to actor and relatedness outweighs the cost. In a mixed-methods article by Garg, Vogelgesany & Kelly (2015), altruistic motivations and perceived benefits of genetic testing among men and women with elevated risk for BRCA1/2 mutations who underwent genetic counseling and testing was explored. Garg et al. (2015) defined the concept of altruistic motivations to include undergoing genetic counseling and testing to provide information for family and to advance scientific research. Altruistic motivations were operationalized by a repeated measures design at three different time points, participants received a perceived benefits of genetic testing questionnaire, and was augmented with transcripts of genetic counseling sessions. Additionally, immersion/crystallization method was used for the qualitative methods to explore perceived benefits of genetic testing, personal motivation, and mutual benefit.

This theory fits within the framework of Garg et al. (2015) article because of its relatedness to the population of interest. This theory outlines that an act of altruism increases the reproductive fitness of individuals and their genes by benefiting their related kin. Participants in this study had already acted to decrease their risk of developing cancer again by undergoing genetic counseling and testing, and some participants having surgical removal of their breast/ovaries. Thus, their actions are more likely to have altruistic motivation for others such as their loved ones and for the advancement of scientific research.
The Integrative Model of Behavior and Prediction

The Integrative Model of Behavior and Prediction takes a reasoned action approach, and is aimed at changing beliefs about consequences, normative issues, and efficacy with respect to a particular behavior. This theory is targeted to those that are designing interventions aimed at increasing health behavior and the decisions faced during the development phase of an intervention. One is assuming that the underlying intention of one’s belief will ultimately change the behavior of the intention when utilizing this model.

The third article by Sussner et al. (2015) examined the beliefs and attitudes about BRCA genetic counseling and sociocultural factors and their influence on their beliefs and attitudes. The Integrative Model of Behavior Prediction by Fishbein & Yzer (2003) was utilized and included key aspects of the Health Belief Model, Social Cognitive Theory, and Theory of Reasoned Action. The purpose of using an integrated model was to recognize the beliefs and attitudes that underlie the intention to perform health behaviors. This author believed that these beliefs and attitudes were foundational to the background influences one has in life, including social and cultural factors. These concepts were operationalized by thematic content analysis in which 12 focus groups were completed.

This theory is relevant to the current article by Sussner et al. (2015) because participant’s beliefs and attitudes surrounding BRCA genetic counseling along with their sociocultural views and how they influenced their beliefs and attitudes were explored. However, what was lacking from using this singular theory was the individual’s perceptions of risk (Health Belief Model), how their social and cultural background influenced their perceived behavior towards risk (Social Cognitive Theory), and actions as related to influence within an individual’s belief and attitudes (Theory of Reasoned Action). Therefore, the author integrated these three additional
theories and models to create a new theoretical framework that fit the purpose of this study.

**Health Belief Model**

The Health Belief Model proposes that a person first believes they are at risk, and will be somehow be affected by this disease or disorder.\(^\text{10}\) Rosenstock (1974) highlights that this model occurs in three stages: (1) the participant either feels personally susceptible to it, (2) the occurrence of disease has at least moderate severity to some component in his or her life, or (3) taking particular action would be more beneficial by reducing susceptibility to condition if disease occurred that outweighs barriers such as cost, convenience, and pain. Thus, participants vary widely in their acceptance of perceived susceptibility and seriousness to a condition or disease.\(^\text{11}\) In relation to the Sussner et al. (2015) article, participants were at-risk through family history or already tested positive for the *BRCA* mutation, which increases the probability of developing certain types of cancer later in life.\(^\text{17}\) In using this model, one has the potential to believe that the benefits of recommended protective behaviors outweigh the costs.\(^\text{10}\) Therefore, individuals with positive *BRCA* mutation status are recommended a different risk management and reductions plan than the average population.\(^\text{17}\)

**Social Cognitive Theory**

Bandura (1986) defines Social Cognitive Theory as a theoretical perspective in which learning by observing others is the focus of the study. Several assumptions underpin this theory: people learn by observing others, learning is an internal process that doesn’t always lead to a change in behavior, goals help to motivate learners and directs behavior, and behavior eventually becomes self-regulated.\(^\text{12}\) Bandura explains the interaction between a person’s behavior, personal factors, and their environment, referred to as the Reciprocal Causation Model, makes equal contributions to behavior. In the article by Sussner et al. (2015), the authors are exploring the
social and cultural factors in participants at risk for HBOC. Therefore, this key aspect within the Social Cognitive Theory further theoretically frames how participants perceive influences within their background on their beliefs and attitudes surrounding BRCA genetic counseling.

Theory of Reasoned Action

Theory of Reasoned Action focuses on the intention for an individual to engage in a certain behavior, which is more of a predictor of whether or not this individual will engage in that behavior; thus, leading to predicted attitudes and subjective norms.\textsuperscript{13,14} If the intended behavior is seen as positive or having a beneficial outcome, the person is more intended to act or engage in that behavior. Sussner et al. (2015) integrated the Theory of Reasoned Action into their study. This cognitive theory offers a framework for understanding human behavior in specific contexts.\textsuperscript{13,14} Since the participants in the Sussner et al. (2015) study are being interviewed to learn more about their beliefs and attitudes about BRCA genetic counseling and sociocultural factors that influence these beliefs and attitudes, it makes sense to use a theoretical framework that also explores the underpinning of their actions within those factors.

Symbolic Interactionism (SI)

Symbolic Interactionism is a sociological perspective in which we give meaning to an act based on our social interactions. Through the seven principles of SI, social context for interaction is defined as ‘society’, and this develops as a result of interwoven patterns of interaction and action.\textsuperscript{15} Crooks (2011) defines Symbolic Interactionism from the perspective of women’s experiences, their own understanding of health-related issues, and the social interactions within which they gain meaning and insight about their situation. Hamilton, Williams, Skirton & Bowers (2009) identified life trajectories that influenced the decision in young women to have genetic testing for mutations in BRCA1/2 and subsequent risk reduction decisions after receiving
MID RANGE THEORIES IN HBOC

a positive mutation status. Women met inclusion criteria if they had received genetic testing for \textit{BRCA1/2} mutations, and were willing to be interviewed using a grounded theory approach to explore genetic testing decision-making that influenced their life trajectories.\textsuperscript{16} This type of theoretical approach fit this current study as it provided a basis for the researchers to explore the women’s perspectives of how society influenced their social interactions around their health.

\textbf{Discussion}

What’s been highlighted in this theoretical review of the literature among individuals at risk for HBOC is there is no one theory that is a ‘one size fits all’ when discussing a population with increased hereditary cancer risk. In order to capture the richness, and in-depth description within these participant’s experiences in living with cancer risk, these articles used multiple mid-range theories, conceptual models and frameworks that were either adapted or integrated together to fit their purposed study needs. A majority of the models and theories were used to construct health behavior interventions while aiming to understand the underpinnings to behavior in those at-risk for HBOC.

Several articles within the past ten years have highlighted mid-range theories and frameworks related to populations with increased cancer risk. These articles discussed the use of grounded theory and other similar models and methodologies to further explain individual’s perception of living with hereditary cancer risk. These articles highlighted an important field of work and also catapulted the way in which we define grounded theory within this population. Charmaz (2008) discusses her belief in relativity and reflexivity within researcher’s positions, practices, and research situation.\textsuperscript{18} In contrast to Glaser & Strauss, and Strauss & Corbin, the addition of these two concepts in research utilizing a population living with hereditary cancer risk, more in-depth and rich data can be generated fueling further development of interventions to aid in these unique realities.
MID RANGE THEORIES IN HBOC

Recommendations

Health behavior theories have strength but not particular to only this population. Using several middle-range theories and/or integration of multiple health behavior models helps to identify and understand the context of those living with hereditary cancer risk. Utilizing higher order theories are great, but only partially informs constructs within this population. Therefore, more studies are needed that review the rigor of our current mid-range theories in relation to this population of interest, and those integrated theories and models in a more diverse population.

Conclusion

These five articles explored the same population of at-risk women in BRCA genetic counseling and screening; however, they widely used mid-range theories and models. Three of the four studies used several mid-range theories to comprehensively capture their sample population from a theoretical framework. More studies are needed to replicate and review these integrated mid-range theory models among this population to depict the psychosocial and cultural experiences of those living with hereditary cancer risk.

References


## Table 1. Integrative Review Articles containing Theory or Theoretical Frameworks

<table>
<thead>
<tr>
<th>Author (Year, Country)</th>
<th>Topic/Aim</th>
<th>Sample Characteristics</th>
<th>Theory &amp; Method</th>
<th>Measurements</th>
<th>Main Findings</th>
</tr>
</thead>
</table>
| Katapodi et al. (2011, USA) | To examine difference and correlations in appraisals of HBOC, psychological distress, family environment, and decisional conflict between women who pursued genetic testing and their at-risk relatives who did not. | Sample = 372 Female  
200 probands and 172 relatives  
(86%) probands affected by cancer  
(21%) of relatives affected by cancer  
Mean age:  
● Probands: 51  
● Relatives: 48  
Age range:  
● Probands: 22-83  
● Relatives: 18-81  
95% Caucasian  
BRCA Status  
*Probands: 25  
● Known in other family members: 21  
*Relatives: -  
● Known in other family members: 23 | Stress & Coping Theory  
Decisional-Conflict Theory  
Family Systems in Genetic Illness Framework  
Correlational, cross-sectional cohort study  
Mailed questionnaires | Demographics: age, race/ethnicity, level of education, employed full or part-time, household income  
Medical History: had insurance coverage (yes/no), when did you receive BRCA testing, personal history of cancer (breast/ovarian/other), prior breast/ovarian surgeries, BRCA status  
Family History: known mutation in other family members  
Surveys: Illness Appraisals of HBOC  
● Perceived caused  
● Perceived controllability  
● Perceived severity (p <0.001)  
Perceived risk of breast (p =0.001)  
/ovarian cancer Psychological Distress | Probands  
Greater knowledge of risk factors of gene inheritance, and greater decisional conflict regarding genetic testing, which correlated with lower perceived risk and better family communication. Perceived controllability was related to lower perceived risk.  
Relatives  
Had perceived controllability was related to lower distress in relatives. Higher decisional conflict was associated with less knowledge and less perceived severity. |
MID RANGE THEORIES IN HBOC

| Katapodi et al. (2013, USA) | (1) To examine whether individual and family characteristics have a direct effect on women’s decisions to use genetic testing for hereditary breast cancer.
(2) To explore whether family characteristics moderate the relationships between individual characteristics and the decision to use genetic testing. | Sample= 372 Female 168 probands, and 168 relatives  
(86%) probands affected by cancer compared with (21%) of relatives affected by cancer (p <0.05)  
52% first-degree relatives  
Age range:  
- Probands: 22-83  
- Relatives: 18-81  
95% Caucasian  
BRCA Status  
- Probands: 25  
- Relatives: - | Stress & Coping Theory  
Family Systems in Genetic Illness Framework  
Decisional-Conflict Theory  
Descriptive  
Cross-sectional, cohort study  
Mailed questionnaires | Demographics: age, race/ethnicity, level of education, household income  
Medical History: had insurance coverage (yes/no), personal history of cancer (breast/ovarian/other), prior breast/ovarian surgeries, BRCA status  
Family History: not reported  
Surveys: Illness Appraisals of HBOC  
- Perceived caused  
- Perceived controllability  
- Perceived severity (p <0.001) | Positive association with genetic testing were education (p=0.01), personal history of cancer (P<0.0001), perceived cause (p<0.0001), psychological distress (p=0.03), perceived utility of genetic testing (p<0.0001), and family hardiness (p=0.02).  
Negative association with genetic testing were perceived controllability (p<0.04) and perceived severity (p<0.0001).  
Those with a personal history of cancer was associated with genetic testing than those without a personal history of cancer (OR= 4.76, p = 0.009). |
### MID RANGE THEORIES IN HBOC

<table>
<thead>
<tr>
<th>Study</th>
<th>Aim</th>
<th>Sample</th>
<th>Integrative Model of Behavior Prediction</th>
<th>Demographics</th>
<th>Themes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sussner et al. (2015, USA)</td>
<td>To examine beliefs and attitudes about BRCA genetic counseling for HBOC among diverse, at risk women (both affected and unaffected) who have not previously undergone counseling.</td>
<td>Sample = 54 Female, at least 18 years old&lt;br&gt;Self-identified Latina women&lt;br&gt;60.5% had a personal history of breast/ovarian cancer&lt;br&gt;46.5% had a family history of breast/ovarian cancer&lt;br&gt;76.7% had children&lt;br&gt;Mean Age: 49.9</td>
<td>Integrative Model of Behavior Prediction&lt;br&gt;Thematic content analysis</td>
<td>Demographics: age, employment, income, level of education, nativity/country of origin, interview language (preference of English or Spanish) &lt;br&gt;Medical History: personal and family history of breast/ovarian cancer, insurance status&lt;br&gt;Family History: marital status, number of children, parent’s nativity</td>
<td>1) Illness Prevention&lt;br&gt;2) Personal &amp; Community Knowledge about BRCA Genetic counseling&lt;br&gt;3) Perceived Benefits and Barriers of BRCA Genetic Counseling and Cultural Influences on Genetic Counseling Participation</td>
</tr>
</tbody>
</table>

- Genetic testing was positively associated with higher family hardness (p=0.04).
- There was a negative association between genetic testing and perceiving negative consequences of breast cancer.
<table>
<thead>
<tr>
<th>Study</th>
<th>Methodology</th>
<th>Sample</th>
<th>BRCA Status</th>
<th>Demographics</th>
<th>Medical History</th>
<th>Family History</th>
<th>Pre-Counseling</th>
<th>Perceived Benefits</th>
</tr>
</thead>
<tbody>
<tr>
<td>Garg et al. (2015, USA)</td>
<td>To conduct an exploratory, longitudinal study by testing perceived benefits of genetic testing among men and women at elevated risk for BRCA1/2 mutations over 3 key time points in genetic counseling and testing process to understand altruistic motivations to test.</td>
<td>120</td>
<td>BRCA status not reported</td>
<td>Sample = 120 Female: 89.2% (70 families) Mean Age: 3 Categories ranged from 47.5 to 54.9 Age Range: 18-83 Race/Ethnicity not reported</td>
<td>Hamilton’s Kin Selection Theory Narrative Analysis guided by principles from Labov Immersion/ Crystallization method Prospective T1: before genetic testing T2: One-two days after counseling T3: 6 months after receiving genetic testing results</td>
<td>Demographics: age, gender, level of education, household income Medical History: personal history of cancer, genetic test results Family History: marital status, used medical records to confirm personal and family history of cancer, number of children, and number of siblings</td>
<td>Pre-counseling questionnaire Perceived benefits of testing questionnaire (open-ended)</td>
<td></td>
</tr>
</tbody>
</table>

| 4) Influence of Previous Interactions with the Healthcare System Suggestions for Educational Tool about BRCA Genetic Counseling for HBOC | acculturation scale for Hispanics (BAS) | 47.5% had personal history of cancer | | | | | |

6 Themes
Personal Motivation:
1) Cancer prevention (64.3%)
2) Acquire knowledge (42%)
3) To alleviate anxiety (16.1%)
4) For future planning (5.4%)
Altruistic motivation:
5) For family’s sake (25%)
6) For science and research (13.4%)
| Hamilton et al. (2009, USA & Canada) | To explore the influences on decision-making in women who tested positive for the \textit{BRCA1/2} mutation | Sample= 44 Female  
21 diagnosed with breast cancer, 23 no history of cancer  
22 states (41 participants) & Canada (3 participants)  
Age range: 18-40  
43 White European  
1 African American  
BRCA1: 30  
BRCA2: 15 (one participant affected by both) | Theory of Genetic Vulnerability (Hamilton & Bowers 2007)  
Grounded Theory Method  
Focus: on women who tested positive for the \textit{BRCA1/2} mutation  
Family Systems Genetic Illness Model | Identified 4 life trajectories:  
2. Loss of Mother to breast cancer at a young age.  
3. Expression of concern by a health care provider.  
4. Personal diagnosis of breast cancer (premenopausal) | Hamilton et al. (2009, USA & Canada) |
IV. Manuscript 3

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SHIFTING OF SELF

Abstract

Background: Risk assessment through genetic counseling and testing (GC&T) are offered to individuals who meet criteria associated with hereditary breast and ovarian cancer syndrome (HBOC). Individuals who know of inherited cancer risk are often gatekeepers of inherited risk information to blood-related relatives. However, upwards of 59% offered GC&T may choose not to undergo genetic testing. Current literature highlights how at-risk individuals receiving care at large, academic medical centers in urban areas experience and make decisions around GC&T. A critical gap exists in understanding the decision-making process of individuals with risk for HBOC are offered genetic testing in rural settings.

Methods: A social constructivist, grounded theory approach was conducted within a rural area of the Northeast region of the United States. Participants were eligible if they were a) at least 21 years old, b) had been offered genetic testing, c) did not seek medical care at a large, academic urban medical center, and d) were able to speak and understand English. Purposive sampling and data analysis were completed using grounded theory methods as outlined by Charmaz (2014).

Results: Thirteen participants with an HBOC associated cancer diagnosis were included in the analysis. Interviews ranged in length from 60 to 90 minutes. Participants described the shifting of self during the decision-making process in utilizing genetic testing. Three theorizing categories were identified: waiting in limbo, wanting answers, and having expectations.

Conclusions: Findings of this study suggest individuals with risk for HBOC-associated cancers are motivated to receive testing for themselves and loved ones, but additional individual and familial supports are needed during the decision-making process. Shifting of self plays a critical role at all stages of this process, and should be a priority in promoting the adoption of novel theoretically based decisional aids tailored to the individual living with hereditary cancer risk. Research efforts should continue to help guide practice, policy, and research initiatives.
1. Introduction

Genetic counseling and testing are offered to eligible individuals who have personal or familial factors associated with hereditary breast and ovarian cancer syndrome (HBOC) (National Comprehensive Cancer Network, 2017). The genetic testing process provides individuals and blood-related family members with information about certain genes associated with HBOC, of which a gene with a positive result (i.e. a mutation is present) indicates that individuals has a higher probability of developing HBOC associated-cancers (i.e. breast, ovarian, melanoma, prostate, and pancreatic) later in life (NCCN, 2017). Knowledge of cancer risk status has the potential to impact future health-related decisions for the individual and family. Patients diagnosed with HBOC-associated cancers are most often the gatekeepers of genetic information, and extend knowledge of risk to at-risk blood-related relatives (Proulx et al., 2009; Zilliacus et al., 2012; Katapodi et al., 2011; Garg, Vogelgesany & Kelly, 2016; Glenn, Chawla & Bastani, 2012). Therefore, those considering undergoing genetic testing find themselves making genetic testing decision within a familial context, and it could be the catalyst to decisions if their at-risk relatives will obtain genetic testing themselves.

Current literature suggests that individuals with risk for HBOC often make decisions within a familial context, and uncovering hereditary cancer risk among family members is complex and an integral step within the genetic counseling and testing process. Studies have investigated factors associated with the decision to utilize genetic testing among individuals with HBOC at large urban academic medical centers. However, studies focused on the decision-making process in individuals with HBOC residing within a rural environment with limited access to genetics services has been limited. Therefore, the purpose of this study was to explore the decision-making process in the utilization of genetic testing in a rural environment among
individuals with risk for HBOC. Outcomes from this study will inform the scientific community how those with risk for HBOC make informed decisions about genetic testing through the exploration of their decision-making process.

**Specific Aims**

1. Describe the process by which individuals with HBOC who reside in rural settings with limited access to care from large, academic medical centers make genetic testing decisions.

2. Understand the social context of the HBOC genetic testing decision-making process.

**2. Methods**

The objective of this proposed study was to explore the decision-making process of individuals with HBOC in the utilization of genetic testing. Although literature exists that explores the relationship of relative’s decisions to undergo genetic testing, there is a paucity of literature from a theoretical perspective while utilizing a grounded theory approach within a rural environment. Advancements in science among screening, treatment, and prevention strategies in hereditary cancers and genomics is expanding rapidly. Individuals with HBOC and their blood-related family members who reside in rural settings are considered an underserved and potentially vulnerable population.

Institutional Review Board approval was obtained from Northeastern University. Participants were recruited from a non-profit collaborative program that provided screening, educational, and additional support services for individuals and their families living with cancer within a rural Northeast region of the United States. The principal investigator (lead author) was invited to speak at several gatherings to recruit and enroll potential participants. Participants were eligible if they (a) were at least 21 years old; (b) the potential participant and/or loved one
was offered genetic testing from a medical professional, and (c) could speak and understand English. Participants were considered ineligible if either the participant or a loved one was currently on end-of-life care, and if they have received medical care at a large, academic medical center related to genetic testing. At the time of the study, there were no affiliated medical centers in the immediate area of this rural Northeast region that were considered under the umbrella of an “academic” medical center as defined by the Association of American Medical Colleges (AAMC). Interested participants were screened for eligibility by the principal investigator (PI) and provided informed consent prior to data collection. Each participant received remuneration of $25.00 for their time in this study.

Data were collected via individual unstructured interviews and conducted in-person. The interview guide was developed in collaboration with authors in this study and reviewed after the first three interviews were completed to ensure understandability. Interviews began with the opening prompt, “Please tell me about your experience when either you or your loved one was offered genetic testing” (Table 2). Participants were encouraged to speak freely and prompted to discuss the decision-making process to either receive or decline genetic testing. At the end of each interview, a paper-based demographic questionnaire was collected that asked additional study-specific information. Interviews were audio-recorded, lasted on average 60 to 90 minutes, were manually processed to remove any identifiers, and professionally transcribed. Transcripts were loaded into NVivo 11.4.3 and verified against de-identified audio recordings by the principal investigator prior to analysis.

Consistent with grounded theory methods (Charmaz, 2014), data analysis began immediately following the first interview and continued concurrently throughout the recruitment stage. This approach allowed the study team to purposively recruit participants that were likely
to add information-rich context to further understand the phenomena of interest. The sample for this study was derived using theoretical sampling described by Charmaz (2014). Theoretical sampling involves, “starting with data, constructing tentative ideas about the data, and then examining these ideas through further empirical inquiry” (p. 199). Theoretical sampling as defined by Charmaz (2014) is strategic, specific, and systematic. It’s used to define, elaborate, re-fine theoretical categories, which helps to delineate and develop properties in each category. Categories are considered ‘saturated’ when gathering fresh data no longer leads to additional insights or reveals new properties for core theoretical categories (Charmaz, 2014). Participants were not interviewed more than once.

An inductive approach was taken during data analysis. The PI and study team assigned open coding labels to similar underlying concepts found within the data. These concepts were further refined using the Constant Comparative Method originally developed by Glaser & Strauss (1967). Over time as more interviews were completed and analyzed, commonalities among concepts were formed into categories through open and axial coding. Then, following the method outlined by Charmaz (2014), the study team went a step further and developed subcategories from the emergence of data, explained the links between each category, and learned about the experiences each category represented. This emergent process was iterative, and kept the study team actively engaged in the data, and emphasized the process of analysis and development of theoretical categories by going back and forth between inductive and deductive reasoning (Charmaz, 2008).

Throughout the data analysis, the PI kept detailed methodological and reflexive memos and diagrams, and used thick description to ensure a detailed account of the field experience(s) for each participant (Holloway, 1997). The PI and research team met regularly to debrief, review
findings, and discuss commonalities and differences in the interpretation of data, and emergent conceptual analyses. Credibility was enhanced by regularly de-briefing and analyst triangulation, which provided checks during the analysis to illuminate any blind spots in the interpretive analysis to stay consistent with the theory and method (Lincoln & Guba, 1985). Reflexivity was conducted as a requirement to maintain quality of the PI’s appraisal, evaluation, and reporting of the data (Gentles, Jack, Nicholas & McKibbon, 2014). The PI’s research team closely examined the detailed audit trail to ensure reliability of this study (Lincoln & Guba, 1985).

3. Results

3.1 Approach and Purposive Sampling

Seventeen individuals with an HBOC-associated cancer showed interest in this study. After the PI screened for eligibility, fourteen individuals were provided informed consent and interviewed. During the first and second phase of recruitment, participants were recruited from an advocacy and support group located in a rural Northeast region.

The median age at the time of initial diagnosis of cancer for women (n=9) was 42 (range 30-57), and for men was 51 (n=4) (range 44-60). The median age was 47 for women and 57 for men. After the first phase of recruitment and de-briefing with the team, nine (69%) of participants were included into analysis. It was determined that understanding the perspectives and experiences of individuals through a gendered context was important and was lacking within the first phase. Therefore, effort was made to recruit additional men with a diagnosis of a HBOC-associated cancer as well as those whose characteristics differed in terms of age, education, and household income from phase one participants. Phase two of recruitment yielded three men and one woman who were recruited from a different branch of the advocacy and support group. These four participants were older with a wider range of educational backgrounds and household
SHIFTING OF SELF

income than those in the first phase. One participant was excluded after team de-briefing for not meeting criteria for inclusion in the analysis. Collectively, thirteen participants ranged from 43 to 78 years old from the same rural area accessing similar cancer treatment and genetic testing facilities. Participants reported thirteen family members (six parents, seven siblings) were offered and received genetic testing of which five familial mutations were identified. Twenty different types of cancers were reported among their family members with breast cancer and melanoma being the highest, respectively. Participant and familial characteristics are detailed in Table 1.

3.2 Thematic Results

The core category identified during data analysis was shifting of self during the decision-making process in the utilization related to completed genetic counseling and testing. Participants described shifting of self when discussing thoughts, actions, and experiences related to genetic testing. Shifting of self can occur when intrapersonal and interpersonal behaviors are impacted by ‘life-changing’ experiences such as learning one’s cancer diagnosis or cancer risk status or that of a loved one. Specific to this study, shifting of self was evident during the decision-making process when participants discussed wanting autonomy in making health-related decisions, the choice to self-direct care for themselves and their loved ones, and the ability to be adaptive and self-advocate over their medical diagnosis and treatment plans. When participants experienced these moments, their vulnerabilities were exposed and displayed through a variety of behaviors that led to actions such as accepting or declining genetic testing, this led to a shift in self because now they identified themselves as someone with higher cancer risk; their sense of self in “who am I” was shifted regardless if they received genetic testing.
SHIFTING OF SELF

Those that received genetic testing results continued to have a shift in self as their level of knowledge and understanding of “who am I” expanded.

Learning one’s cancer risk status through genetic testing results, that of a loved one, and/or receiving a cancer diagnosis was a pivotal moment in time for all participants. Participants in this study encountered several situations where they reflected on their past or contemplated their future. Some described that their genetic testing information led to emotions such as fear, frustration and/or grief when discussing their mutation status with their family members and how it could impact their lives as well as how it potentially impacted having a cancer diagnosis. Other participants however described satisfaction and relief when discussing their decision-making process for genetic testing in similar situations with their loved ones.

“Even while you are vulnerable, feel vulnerable and scared, you become alive and joyful.” (Participant 2)

“It makes you more aware. It makes you think about things. Would I still do it? Yes. I would still do it, because I think it would be wrong if I didn’t.” (Participant 9)

Figure 1. discusses the relationship of the core category, shifting of self, to the three theorizing categories, subthemes, and perspectives and experiences of others within the participant’s life that influence their decisions.
SHIFTING OF SELF

For participants in this study, the vulnerabilities they felt within the core category of shifting of self was associated with the process of decision-making surrounding genetic testing, which represented a complex and dynamic intermingling of past, present and future health-related decisions. Many of the participants were being protective of their loved ones while simultaneously reflecting on the impact their actions will have or have had on themselves and their loved ones. In figure 1, the inner circle represents the theorizing categories and their subthemes as related to shifting of self. Note that these categories cross the inner and outer rings. The outer ring represents the perspectives and experiences of others within the participant’s life that were shown to influence their decisions. There are no arrows and all lines are solid to indicate that this process is fluid and intertwined in a variety of ways related to this decision. Below is an overview of these theorizing categories, and how these processes prompted participant’s decisions about genetic testing.

3.2.1. Waiting in Limbo

The concept of time was perceived by participants either as a linear, strategic step in the decision-making process where past, present, and future were three distinguished and separate time points or time was a fluid, circular process where participants reflected on the past, present, and future simultaneously. Some participants discussed time as the period between conversations with medical personnel and family as they waited for outcomes that would shape the decision they would make about genetic testing. For other participants, the process was more philosophical in which they synchronously reflected on previous health-related decisions and those outcomes and determined the pros/cons of forthcoming decisions at the individual and familial level.
**Waiting to clarify cost & insurance coverage.** Participants discussed how cost and/or insurance coverage influenced their decision-making process when offered genetic testing. Participants found themselves in two possible situations: they needed to determine if their insurance coverage would cover the test and if not, how much would be expected out pocket and could they afford this, or that this decision was already pre-determined by either a having a voucher from a support group or their insurance already stated it would be covered due to their cancer diagnosis. In each scenario, the process of time as well as their level of distress varied based on whether the cost and/or insurance would influence their decision. Some participants had to wait weeks to determine whether their genetic tests would be financially covered, which led to being more distressed about the forthcoming decision whereas others did not need to wait for this information.

“I remember them saying that if insurance doesn’t cover it, we will come back to you and tell you how much it will cost. And then you can decide whether or not you want to go forward. They didn’t come back with anything so my insurance covered it. Had they not covered it, I don’t know if I would have done it.” (Participant 1)

“At the time, we had fantastic insurance. The idea of cost never came up.” (Participant 3)

“I’m the type of person when she’s says, "I'm thinking about ...", go for it. Go do it. I'm not one to turn around and go, "Oh, how much money is it going to cost? When it comes to health issue I don't think money is an issue. If you got it, take care of yourself. Don't fool around. Don't throw your life away for a nickel or a dime. It isn't worth it.” (Participant 13)
Within context of cancer diagnosis and treatment. All participants had a diagnosis of an associated HBOC-related cancer (breast and prostate). In some cases, the timing of offering participants genetic testing occurred after meeting with an oncologist and having a plan in place for chemotherapy and radiation. Several participants felt that only during the counseling process, after being referred for testing to make cancer treatment decisions, did they then fully understand the medical consequences of having a positive genetic testing. The participant felt their current cancer treatment plans were ‘useless’ if their results came back positive for a mutation. Participants were overwhelmed with their cancer diagnoses and the multitude of discussions and decisions made within a short period of time, including having to now process genetic testing and what that test would mean to them, their cancer treatment, and to their individual and familial future.

“And genetic testing was a piece of that making a decision about whether to get radiation or getting the Lupron shot. I could have said no to both, I could have said nooooo, I’ll wait but I didn’t. I followed some advice…” (Participant 2)

“Yea, I mean…what I didn't ... I kind of was like “yeah, sure,” I mean I've had like 5000 tests, I have my shirt off every five seconds, somebody's poking me, somebody’s this, somebody's that, and I was like... ‘what the freak is one more test?’” (Participant 8)

Some found the offer to take blood samples for genetic testing during their cancer treatments helpful because this alternative method aided in the decision during a time where other medical decisions took priority. This was especially helpful for those who described making these decisions with ‘chemo brain,’ where remembering the details of this experience would have been more difficult for them.

“Yes, because I was already there. I already got the IV. They were already pulling blood
for the oncologist before chemo, so it was just like, a couple extra tubes is fine. It was convenient. It was helpful. It really was. Not another thing to remember” (Participant 9)

Upon several participant’s reflections, several wished the timing of their cancer treatment and genetic testing were not so intertwined. The timeframe between diagnosis of cancer, treatment plan, and conversations about genetic testing were perceived as quick, difficult to interpret and remember to process and share with their loved ones.

“I wish genetic testing went different.” (Participant 5)

“When she called and said, “it's a 'no',” I was just- (referring to genetic testing results) I'm gonna be okay. I can get back to the road I was on with this to begin with, because in waiting for that result, I kind of felt like I was in limbo. I'm on this road but now this road might change dramatically.” (Participant 8)

During their cancer diagnosis and treatment plan discussions, some participants needed no time to make the decision because it was a “no-brainer,” despite feelings of being unsure, uneasy and scared. Some felt there was little to no time offered to process what the decision to receive genetic testing would mean to them and their families. They received genetic testing immediately and therefore that time to process this decision was either absent or cut short as compared to those that had a previous discussion about it with their provider and family. In other cases, participants completed genetic testing first and then waited for their results, which provided time for reflection on how the outcome would affect them and their loved ones.

“No, it was when I was waiting. I was like, "Oh, crap." I didn't think about it when I went to go get it done.” (Participant 9)

In most cases, participants wanted time to emotionally cope with a potentially negative outcome first, and then share the information with loved ones if it was “bad.” Those who felt
they had sufficient amount of time to process their decision described more confidence and control in their immediate decision provided them with time to rationalize a negative outcome and determine the best route of communicating this information in a way that provided love, support, and understanding to their loved ones.

“I'm gonna be okay. I can get back to the road I was on with this to begin with, because in waiting for that result, I kind of felt like I was in limbo. I'm on this road but now this road might change dramatically and that's a whole other thing that I couldn't go to like I could before when I was deciding about the chemotherapy.” (Participant 8)

**Needing time to understand position of family members.** Participants valued input from their loved ones and would often state that once genetic testing information was communicated to them from a medical professional, several wanted time to discuss their options with family members, most commonly spouses and children. In some cases, this sharing of information had repercussions that led to regret and guilt by the participants.

“Because it both sent them (daughters) on a path of checking the internet to see what they could possibly have. I should have waited until it came back but I was trying to be open and make conversation.” (Participant 10)

In other situations, it motivated family members to seek out additional information and consider genetic testing for themselves. There was time in waiting to see their provider, and not knowing the outcome of their conversation and whether genetic testing would be considered a mutually agreed upon decision.

“Then it was like, “You should really do this.” So, I think she had gotten in at the end of the year previous. And then my sister and I probably were doing it around the same point in time.” (Participant 4)
Decision was already made. When discussing genetic testing with providers and loved ones, several participants either felt the control to make their own decision was taken away because the decision was already made for them, or they trusted that their faith in the process or the Divine would provide the right decision and would prevail. The loss of control felt was demonstrated in three ways. In the first case, participants felt overwhelmed with a sense of responsibility and obligation towards their loved ones. Participants believed they had no choice but to receive genetic testing for the sole purpose of sharing this information in hopes of prevention on behalf of their loves ones. Part of this decision-making process was determining if they could live with the guilt associated with not doing genetic testing.

“You don't anticipate how scary that was because everybody said, "we don't think that's an issue for you." Nobody gave me any information about it. When I sat down in front of the television with this doctor, she said, “you'll get one of three results back: you'll get a 'yes', you'll get a 'no' or you'll get a 'we're not sure' ... She went through, in somewhat graphic detail about what 'yes' means ... double mastectomy and everything...Yeah, no, there was no participation in that part of it. It just was what it was...If it was a 'yes', I was gonna have to go back and do over again” (Participant 8)

In other cases, participants were offered genetic testing after their cancer diagnosis and treatment discussions felt the decision to receive genetic testing was already made for them by their medical providers. Some trusted their providers and would oblige their recommendation to receive genetic testing regardless if they personally felt unsure about that decision. If results revealed a positive mutation, decisions for additional screenings and prophylactic treatment were already determined based on current guidelines and standards of practice. They felt it would be irresponsible to not go through with best practice recommendations, which are set to decrease
cancer risk or re-occurrence for themselves and their loves ones. Although several participants described a feeling of not being in control in their decision-making process, others placed their faith in the medical team to provide the necessary support.

“Putting a lot of faith in terms of genetic testing into the genetic testing people, so yea...like I don’t know the deep science but there was nothing too unusual about my results so I didn’t really worry about it.” (Participant 2)

Lastly, participants that displayed their faith in their decision-making through conversations about their religion expressed that their path in life was already pre-determined and the choices they make were not actually their choice, but their Divinity paving their already destined path.

“But I am a woman of faith and I put that in my Lord’s hands at the very beginning of this process and that’s what got me through it and that’s the only thing that got me through it. Because I on my own, I gave up a long time ago. I’m not that strong.” (Participant 4)

Genetic testing was viewed as part of this pre-determined path and that communication through reading the bible, prayer, and attending church services would continue to connect them with their Divine, who was in control of their destined outcomes.

“Like I said, I’m a woman of faith. I know my expiration date is already been written down, but I’m definitely not going to push it and I don’t want to suffer until it happens. And there’s a reason for everything.” (Participant 4)

Shifting of self was evident throughout this theorizing category through rich descriptions of different time points during their decision-making process in utilizing genetic testing services. As they gained more understanding of their cancer risk status and the implications to their
decision, *shifting of self* occurred in understanding who they were as a person and how their relations and actions affected those they loved.

### 3.2.2. Wanting Answers

During the decision-making process, participants were wanting answers as to how a potential mutation may affect their loved ones, more information regarding the genetic counseling and testing process, and ways they could prevent harm to themselves and loved ones in the future. Being protective towards their loved ones in which they were concurrently reflecting on their past as well as looking towards the future for explanations in related situations that had occurred or they fear may occur in the future.

*What if I did something wrong.* Respondents viewed genetic testing as a way to answer questions about whether situations (i.e. biological or environmental) from their past could have caused their mutation for either personal closure or to share this information with their loved ones. Although several participants weren’t sure how they would have shared the news if it was ‘bad,’ all thirteen participants wanted to share this information in hopes that their loved ones would seek out the appropriate medical attention for preventative care to avoid having to make similar cancer-related decisions in their future. The main goal in sharing this information was to ‘protect’ loved ones, even when they weren’t sure how this information would be received and how it would influence their future health-related decisions.

“Yea. I was just so curious. I wanted to know why I got cancer. Was it just a genetic thing? If it was, I can understand that. But if it’s something environmental that I’m doing, I want to know that too... And the whole thing with my family. You know my sister, should she be concerned? Should her daughter be concerned? That sorta thing.” (Participant 1)
When participants reflected on this aspect within their decision-making, there was a sense of guilt or sadness at the possibility of being the cause of their love child’s cancer or placing their loved ones at a greater cancer risk if they had a positive mutation. Those that received negative results felt a sense of relief when given this news, which provided closure or justification to wanting answers as part of their decision-making process. Parents with children who did not have a diagnosis of cancer and vise versa had similar questions of looking back at the ‘what-if’ s’ of life, and how past decisions could be impacting their present situation. This led to having to make similar decisions regarding getting genetic testing in order provide a justification to their thoughts and feelings.

“Yeah. If my mother had done it, like if mine came back positive and my mother didn't do it, but what if my mother had done it and hers came back positive? Then would I still have gotten cancer? Could I have somehow done something different?” (Participant 9)

Wanting to educate themselves. Most participants were informed about genetic testing through their primary care provider or oncologist during conversations about their cancer diagnosis and treatment. The part of the conversation dedicated to discussing genetic testing were perceived as quick, vague, and sometimes left the participant to seek out additional information to decipher and digest on their own.

“He just came right out of the blue and said, "You know, I think we need to do some genetic testing on you." He didn't really give me a, you know, this is why, this is not why. And, I just figured it wouldn’t hurt...I have daughters, so I was kind of concerned that I know there’s a direct connection between prostate and breast cancer because I did research on it.” (Participant 10)
SHIFTING OF SELF

To learn more about the genetic testing process and to help with their decision, participants searched a variety of sources and were ultimately left to teach themselves and their loved ones if they chose to include them in the conversations. Some participants had a ‘thirst’ for knowledge because the process of deciding meant the more information they had, the more confident they were in their decision.

“... you really, really need to have the information so you can make informed decisions.” (Participant 6)

In other cases, providers and or loved ones recommended genetic testing and it was the participant that based on previous readings on the internet decided that either a particular genetic testing panel that was offered or genetic testing in general was not a good fit for them at that time.

“It didn't fit, because I read all this stuff about the BRCA information...but I agreed to do it basically to get people off my back so they wouldn't bother my sister.” (Participant 7)

Advocating for oneself. Several participants who advocated for themselves and displayed assertiveness with their providers also self-identified as confident by using similar words or phrases like speaking up, advocating for their health, and asking for referrals. Their advocacy towards their own health was a key piece in their decision-making process to receive genetic testing.

“So, I approached my oncologist saying I'm interested in this, can I ... I think it would be worth it to be tested. They just kind of blew me off for a while, blew me off. Then finally said, okay, well, we will go ahead and refer you to the genetic counselor.” (Participant 7)
Likewise, participants advocated for themselves with their family members when they decided not to receive genetic testing at that time.

“My sister came back negative, so I figured what are the odds of me getting it? It’s 50% really. I didn’t know it was those types of odds (at the time). I thought it was a lot lower. I figured I’d take my chances.” (Participant 14)

Several participants highlighted that genetic testing was never discussed as an option during their conversations with their medical teams about their cancer diagnosis and treatment plan. They learned about genetic testing through their loved ones when they received a cancer diagnosis and subsequently were offered genetic testing by their medical teams.

“I’m an advocate of you know, but no one’s ever approached me. I’d be more than happy to do one of those BRCA 1 BRCA 2 type of things, you know? Yeah, it's interesting.” (Participant 13)

Several participants in this study communicated risk to family after educating themselves and learning more about the meaning of having cancer risk. As cancer risk status became more apparent, advocating for oneself and loved ones developed the context of ‘who am I’ more deeply, and began to create a new understanding of how to protect themselves and their loves ones using additional screening and prevention methods.

3.2.3 Having Expectations

Conversations between medical providers and participants formed the initial expectation that genetic testing was another standardized test within the context of their cancer diagnoses and treatments. The implications associated with genetic testing within this context formulated participants views on their level of individual and familial cancer risk. Due to this understanding, some participants wanted to ‘get it over with’, or that there was no question in going through
with genetic testing immediately due to wanting to protect their loved ones. As time progressed and participants learned more information about the implications of cancer risk from genetic counselors, they were either satisfied or disappointed by their initial decision-making process as it currently related to their medical care. In some cases, participants described a whirlwind of unforeseen thoughts and emotions because they felt unprepared to share potentially ‘negative’ news with loved ones as well as make additional decisions regarding their medical care. In other situations, they felt the opposite and understood the implications of genetic testing, which provided a level of satisfaction to their medical care.

**Satisfaction in care.** Participants who felt that their initial conversations with their medical team about genetic testing was sufficient in explaining the implications and what to expect, were more satisfied with their decision-making process than those who didn’t receive the same experience. Participants that felt satisfied believed their provider had their best interests at heart and felt appreciative when communication was honest and straight forward. This included situations when providers were honest about not being informed about their mutation status and referring them to the appropriate resources. Several participants were offered additional panel testing by their providers when science became more advanced. The consensus from participants in this study was that genetic counselors were viewed as the first medical professionals to share in-depth knowledge of the implications of cancer risk, and what a positive result may mean for them as well as how it related to their loved ones.

“No, it was informative. It was informative. I try not to get too crazy about a lot of things.” (Participant 13)
"When I went in for the consultation, she explained the whole thing and she did a great job with it. It was very practical. She didn't get so turned around in the medical terminology. I really understood what she was saying." (Participant 8)

Some participants felt more positive about genetic counseling and testing if they perceived that the information received fit both the individual and family values. Although some participants were motivated to have additional panel testing done, others were surprised that additional testing would mean re-living this decision-making process again and wondering whether genetic testing was the correct decision for them at that time. This experience led to contemplating the pros/cons of going through this experience again for themselves and their loved ones and whether that decision was ‘worth it.’

“I ended up being negative for that (single site genetic test)... I was interested in learning about that (more comprehensive panel), and so I researched that. So, I approached my oncologist saying I'm interested in this, I think it would be worth it to be tested.”

(Participant 7)

Likewise, participants had conversations with their loved ones about the information shared by members of their medical team. Family was viewed as a type of support system when discussing whether receiving genetic testing would be necessary and ‘worth it.’ Although families within this study population commonly said the decision was up to them to make, they still valued input from family. Participants were concerned about how their family would react to hereditary cancer risk information, and how that information would be perceived and used within their own healthcare decisions. Satisfaction in sharing this information was met when families received this information and were supportive of their decisions. Similarly, participants found comfort within a variety of support systems, which aided their decision-making process. Natural
supports (i.e. family, friends, and community) was primarily seen when communicating whether to undergo genetic testing, but also the embodiment of spiritual, the Divine, and close relationship with Mother Nature were of equal importance in support of making this decision.

“You have to make the decision to make yourself comfortable.” (Participant 9)

Disappointment in care. Some participants felt that the genetic testing information from their medical team lacked the appropriate level of discussion about the implications associated with genetic testing, and instead learned this information during genetic counseling sessions described disappointment, anger, and frustration about the situation and sometimes about the care given by their medical team. In some circumstances where they trusted their medical team during the decision-making, some still felt unsatisfied and misled thereby compromising their level of trust.

“Because everybody (oncology team) had been telling me that I’d get the ‘no’, and here this person (genetic counselor) is telling me what the ‘yes’ could mean…a couple of times I kind of checked out cause it was just too much… I think I was … I was definitely more anxious about that test result than I was about the biopsy.” (Participant 8)

Within family members, participants sometimes felt that there was backlash such as excessive worrying, ‘nagging,’ and frustration when they shared their thoughts about whether to receive genetic testing. They wanted the input of their loved ones before finalizing this decision, but felt in hindsight that maybe sharing this information was more detrimental than helpful.

“Every so often when we ever talked about cancer, or somebody else in the family got it. It's like, "Oh you really need to get yourself (genetic) tested." "Okay, yeah (sarcastically).” (Participant 14).
SHIFTING OF SELF

_No one cares_. Several instances led some participants to have the perception that certain medical professionals did not care about their well-being due to the actions that took place either during their cancer diagnosis, treatment, or genetic counseling sessions. They were disappointed that their provider and/or the genetic testing experience didn’t meet their expectations. For example, in one scenario a male participant with prostate cancer was referred for genetic testing and was provided with a breast cancer booklet and counseling session because “that is all they had.” (Participant 10)

In another context, they felt that medical professionals “did not care” because they felt their concerns were not being acknowledged, and that they felt on their own, alone trying to make sense of their cancer diagnosis in the context of now possibly having a genetic mutation that may also increase cancer risk for their loved ones.

“I thought it was kind of a waste of time. I mean, I just felt pushed out. They didn’t care. I’ll go back to the same thing. It’s not breast cancer. They don’t care. Do some research before you go in. Have questions all ready for them because I don’t think they’re always going to have your interest in mind. I guess that’s the best way of putting it. I think you need to be more prepared.” (Participant 10)

Several participants perceived some of the actions taken by medical professionals such as referrals to specialists and sharing of patient documentation to be a different level of care that didn’t meet their expectations. Some were referred to out of state facilities or the option of Telehealth to compensate for a lack of access to certain information and treatments regarding genetic testing. This led to feeling frustrated, angry, and feeling alone in solving their concerns surrounding their medical care, which influenced their decision-making process about genetic testing. On one scenario, a participant described her local hospital using the notes of a specialty
SHIFTING OF SELF

hospital and “piggy-backing care” because they “didn’t really know much about my mutation.” (Participant 7)

Several instances participants described how different medical professionals were unsure exactly what genetic testing should be offered, and left that decision to them. They had to lean on loved ones, independent research, and how they felt about the decision of which test to receive. They felt alone in this decision and weren’t sure if they were making the right one.

_They gave me the choice. "Do you want to do just this one that targets this? Do you want to do a larger panel that targets a couple other things? Did you want to do a larger panel, which we're not sure what all that other stuff is yet, and it could come back as a false positive. We're not sure, but do you want to do that one?" (Participant 9)_

Each participant had a different experience when it came to how genetic testing was offered, and the genetic counseling session they received. Depending on how the initial genetic testing conversations went with either their families or medical teams helped to shape the experience they would later have receiving genetic counseling. Each scenario remained the same in which the pros and cons of their decision would be weighed against the implications of the test and how that decision would impact their future. Participants reflection of this experience led to identifying that part of their decision-making process that either made them feel satisfied or dissatisfied with their decision. Their level of satisfaction in this decision led to a shift in their sense of self and continued to provide a deeper understanding of “who am I” in the context of learning of their cancer diagnosis, potentially a positive mutation that increases cancer risk and the impact of this information on their loved ones. Additionally, this shift of self in “who am I” created a ‘bigger picture’ scenario that described how their decision-making process will influence their future health-related decisions for themselves and their loved ones, and how their
medical team could contribute to future approaches in hereditary cancer risk screening, prevention strategies, and treatment options.

4. Discussion

Living with hereditary cancer risk means understanding the implications of a positive mutation and how to navigate available resources to maintain current standards of practice for screening and preventative options. However, many in this study received a cancer diagnosis first and only learned about their hereditary cancer risk status afterwards through being offered genetic testing. A diagnosis of an HBOC-associated cancer means that this type of cancer can be more common among blood-related relatives, and genetic testing helps to identify the probability of cancer risk due to a genetic mutation. These experiences are complex and add to the intricacies providers face in referring those at risk to the appropriate genetic services and treatment plans on an individualized basis.

Findings in this study suggest that the decision-making process to utilize genetic testing is multi-faceted and deeply intertwined in decisions associated with a HBOC-associated diagnosis. Learning of one’s risk through either genetic testing and/or a cancer diagnosis has led participants in this study to re-define who they are now that they are living within this context. At times, they either felt a sense of control or at a loss when deciding about utilizing genetic testing in the context of a HBOC-associated cancer diagnosis. Shifting of self occurred when their behaviors and actions were shaped by various experiences with loved ones, health professionals, and the surrounding community regarding their hereditary cancer risk status.

Burkitt (1994) discussed that people are products of social relations and practices that embodies specific actions to occur within these relations. Within our lives, our self has the capacity to be molded through interactions by learning and training opportunities. As life
continues to influence our relations and actions, we begin to develop new conceptions of self both consciously and subconsciously. This shift of the self can be linked to a change in self-concept and identity. Self-concept has been described as an ongoing behavior that is mediating and helps to regulate self-relevant actions and experiences in response to challenges from the social environment (Markus & Wurk, 1987).

Current literature suggests that a variety of modifiable (i.e. psychosocial) and non-modifiable (i.e. sex, age, culture) impact the decision to undergo HBOC related genetic testing (Hamilton et al., 2015; Cragun et al., 2015). Similarly, individual and familial vulnerabilities in the context of genetic testing decision-making has been characterized previously (Hamilton et al., 2007). Loss of self is depicted in the advanced illness literature, where it is described as a process where illness increasingly impacts every aspect of life (Charmaz, 1993 as cited in Nanton et al., 2016).

Thus, the current study adds to the existing knowledge by shedding light on the three theorizing categories, which provide a new context to exploring shifting of self within this population residing in a rural environment. What’s crucial to understand from this study is how the decision-making process is related to an individual’s ongoing perception of themselves within the context of a hereditary cancer diagnosis. Once an individual has a shifting of self experience, this new perspective of how they perceive themselves within the context of hereditary cancer risk may shift how they now view themselves within their family and societal roles as well as how they view prevention and health overall. These three theorizing categories encapsulated the core category of shifting of self, which demonstrated the importance that the individual’s perception of themselves before, during, and after the genetic testing decision-
SHIFTING OF SELF

making process continuously evolves and shifts over time. This can influence the perspective of the past, present, and future-related decisions.

The first theorizing category, *waiting in limbo*, is related to self in that participants during this time period do not feel in control because they are dealing with multiple possible paths for the self at any given minute. They feel dis-oriented because they do not truly know which direction their future will go, and most use this time to reflect on their previous and current decisions. Time becomes an importance aspect to their decision-making process in utilizing genetic testing. Time was considered multi-faceted in that in some cases, they felt that the decision had already been made to utilize genetic testing (i.e. based on religious beliefs), or in other cases they needed longer periods of time to make this decision because it captured the nuances of the individualized experience in the decision-making process. Participants needing time to process this decision led to feelings of confidence that their decision felt right for themselves and their loved ones. When a participant was not confident in their decision, feelings in loss of control were illustrated through emotions such as regret, guilt, and distress. Thus, time was considered an important theorizing category that underpins a vital component within the decision-making process.

These findings are consistent with prior studies where time was affiliated with the awareness of living with genetic testing results (Hamilton et al., 2009) or the allotted time to make the decision to utilize genetic testing varied (Ziliacus et al., 2012; Dancyger et al., 2010); however, time has not been further developed as a philosophical underpinning to this decision. This current study illustrates the challenges that participants encounter during the decision-making process where current standard of care practices are generalized and standardized; therefore, are not suitable to an individual’s philosophical beliefs on how time influences their
decision. There is a need to develop and evaluate methods by which to support how one perceives time as an important component in the decision-making process.

In the second theorizing category, wanting answers, each participant’s sense of self has personal standards of morals and values, needs, and desired directions. They advocated for and educated themselves, and asked questions to satisfy their loss of control feeling and similarly assisting in not knowing what direction the future holds due to multiple paths. Asking questions can be empowering to one’s self as it adds stability and assurance during a time where as more information is gathered, the path can easily and quickly shift. Participants wanted to make well-informed decisions related to genetic testing and were willing to advocate for themselves when needed. According to Peshkin (2015), individuals need to be given the opportunity to “…make informed decisions about genetic testing and management based on their associated potential benefits, limitations, and risk as well as their own values, preferences, and goals” (p. 45).

One study found that individuals with HBOC and their families during genetic testing still live with unmet needs in emotional and informational support (O’Neill et al., 2018). Similarly, participants in this study felt their genetic testing conversations with providers and sometimes loved ones lacked the breadth and depth of information needed to feel confident that genetic testing would provide results that would satisfy the answers they needed for justification or closure. Wanting answers to prevent harm to themselves and their loved ones has previously been studied in the broader hereditary cancer risk literature, specifically communication patterns in sharing risk information (Katapodi et al., 2013; Patenaude et al., 2013; Garg et al., 2015), seeking answers out of responsibility/obligation (Dancyger et al., 2010; Etchegary et al., 2009), and perceived level of knowledge as related to understanding factors associated with genetic testing (Garg et al., 2015; Glenn et al., 2012; Hurtado-de Mendoza et al., 2017). This current
SHifting of Self

Study provides a lens through which to understand the potential ramifications of unmet emotional and informational needs in this population.

In the third theorizing category, having expectations, resonates with the core category of shifting of self in the sense that participants had feelings of being disoriented and vulnerable due to miscommunications and absence of knowledge in the current standards of practice with their medical teams. Genetic testing was offered within the context of a cancer diagnosis and treated as common practice. Genetic testing was considered by providers and participants alike as another standardized test as part of the HBOC treatment plan. However, the implications of offering genetic testing during the cancer diagnosis and treatment phase was found by participants in this study to not match their expectations and needs when they received genetic counseling and testing results. This unique perspective at the health systems level demonstrates some notable gaps and limitations in communicating genetic risk information and guidelines for treatment. Prior studies have highlighted the lack of provider referrals to the appropriate cancer genetic services of eligible patients (Jones et al., 2016; Vadaparampil et al., 2010; Sheppard et al., 2013; Cragun et al., 2015) as well as gaps in the actual referral process (Hurtado de-Mendoza et al., 2018). Similarly, previous studies have demonstrated miscommunication and misunderstandings of genetic risk information among providers and patients and their families leading to the growing problem of patients having limited health literacy surrounding genetic information, especially among vulnerable populations within the United States (Joseph et al., 2017).

Genetic testing should be treated differently than just another standardized test during conversations surrounding cancer diagnoses and treatment options. These findings demonstrate the individualized experience in the decision-making process for genetic testing. Given the
potential ramifications miscommunications with patient’s medical teams can have on patients understanding their hereditary cancer risk, future research is needed to better understand how we can assist providers and other clinicians in communicating this sensitive information in a culturally and health literacy appropriate level.

**Nursing implications.** A novel component to this study was that it focused on a rural, non-academic medical center population. Individuals in this population described this process as complex, dynamic, and was intertwined with individual and familial needs within the context of their cancer diagnosis. These decisions about genetic testing were considered part of an iterative process with a variety of unique approaches to genetic testing decision-making of which each decision made was unique to the individual.

Overall, the process of understanding the shifting of self in the decision-making process in utilizing genetic testing described by participants in this study is notable in the values that parallels decision-making tools, such as the Ottawa Decision Support Framework (ODSF). The ODSF is an evidence-based, practice, mid-range theory for guiding patients making health or social decisions (O’Connor, 2006). The ODSF provides an implementation toolkit where decisional aids provide individuals help in clarifying values, and making explicit the decision that needs to be made and designed to complement counseling from a health practitioner. According to the Patient-Centered Outcomes Research Institute (2018), “research shows patients are using decisional aids to gain knowledge and are more confident in their healthcare choices. However, barriers remain to implementing shared decision making approaches in clinical practice settings.” Shared decision-making is patient-centered where clinicians and patients work together to make decisions that balances risks and expected outcomes with patient preferences and values. The findings of the current study complement the use of decisional aids by informing
SHIFTING OF SELF

us that individuals at risk for hereditary cancers making decisions regarding genetic testing frame their vulnerability in unique ways that aren’t explicitly identified yet through current decisional implementation toolkits. As the findings of this study suggest, individuals need more assistance in understanding their *shifting of self* during the decision-making process regarding genetic testing.

Therefore, further research is needed to understand individuals unique and complex vulnerabilities during this process where the *shifting of self* is continually changing and ongoing. We need to focus more attention on understanding how individuals ask questions about risk and make well-informed decisions within the context of hereditary cancer risk so tailored implementation decision-making toolkits can be developed at an earlier stage. In addition, the decision-making process has limited theoretical frameworks that identify *shifting of self* as a key construct, and knowing one’s vulnerabilities when making decisions and asking risk-related questions to decrease their vulnerabilities should be explored. This is essential to helping individuals understand how to take care of themselves, to lessen the burden they face with making complex, but well-informed decisions they are confident with over time, and ensure that as clinicians we understand and support these decisions. Lastly, findings from this study will inform the next steps to advancing the science in this area through practice, policy, and research implications.

**Practice implications.** We need to develop and communicate genetic risk information at an appropriate health literacy level within the community to maximize patient’s understanding of the implications of genetic testing, and focus more attention on how to increase provider referrals for eligible patients to the appropriate genetic cancer services. We need to explore ways to assist providers in navigating how to help patients with hereditary cancer risk make well-informed
decisions regarding genetic testing, especially within the rural context that is culturally and geographically appropriate.

**Research implications.** More studies are needed to further develop and evaluate methods that support individuals in making decisions within a timeframe that allows them to see their decision as a quality-based, valid decision that meets individual and familial needs. Future studies should be theoretically based surrounding genetic testing decision-making in the context of hereditary cancer risk within a rural environment.

**Policy implications.** More efforts are needed on building up rural communities to include mandating educational materials that align with up-to-date national guidelines and standards. Policies should be created that address limitations to current genetic risk information within a rural environment and how to effectively implement these strategies. Practice, research, and policy implications and strengths are further outlined in Table 3.

**Limitations.** The study sample was comprised mostly of Caucasian participants who represented both genders, and a variety of age and income groups all with a HBOC-associated cancer diagnosis. Those recruited through purposive sampling are more apt to volunteer to participate, thus increasing potential selection bias. While three main theorizing categories were identified with a core category, further studies are needed to develop a theory based from these findings as defined by the method of grounded theory. Similarly, we do not know how or if other ethnic and socioeconomic backgrounds will experience similar findings in the utilization of genetic testing. More research should be done that explores the decision-making process in the utilization of genetic testing in individuals at risk but without an HBOC-associated cancer diagnosis. Lastly, our findings cannot be generalized beyond this population, which resided in a rural Northeast region with limited access to large, academic medical centers.
5. Conclusion

The findings of this study suggest that individuals with HBOC-associated cancers are motivated to receive testing for themselves and their loved ones, and that additional individual and familial supports are needed during the decision-making process. *Shifting of self* plays a critical role at all stages of the decision-making process, and should be a priority in promoting the adoption of novel theoretically based decisional aids tailored specifically to the individual living with hereditary cancer risk.

**Funding Acknowledgements**

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SHIFTING OF SELF

References


### Table 1.

<table>
<thead>
<tr>
<th>Participant Characteristics</th>
<th>N (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>N</strong></td>
<td>13</td>
</tr>
<tr>
<td>Gender</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>4 (31)</td>
</tr>
<tr>
<td>Female</td>
<td>9 (69)</td>
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<tr>
<td>Biological Children</td>
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<tr>
<td>Yes</td>
<td>7 (54)</td>
</tr>
<tr>
<td>No</td>
<td>6 (46)</td>
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<tr>
<td>Ethnicity/Race</td>
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<tr>
<td>Caucasian</td>
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<tr>
<td>Native American</td>
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<tr>
<td>Cancer Diagnosis</td>
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<tr>
<td>Breast</td>
<td>10 (77)</td>
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<tr>
<td>Prostate</td>
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<tr>
<td>Mutation Status</td>
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<td>Negative</td>
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<tr>
<td>Positive</td>
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<tr>
<td>Variant</td>
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<tr>
<td>N/A</td>
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<tr>
<td>Marital Status</td>
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<td>Never Married</td>
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<td>Divorced/Separated</td>
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<td>Highest Grade Level</td>
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<tr>
<td>Some College/Associates</td>
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<tr>
<td>Not Employed/Retired</td>
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<td>Insurance Coverage</td>
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<td>HMO/PPO</td>
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<td>Medicaid/Medicare</td>
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<tr>
<td>Military</td>
<td>3 (23)</td>
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</table>
### Table 2: Interview Guide for Grounded Theory Study

<table>
<thead>
<tr>
<th>Question Type</th>
<th>Interviewer Statement</th>
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<tbody>
<tr>
<td>Opening question</td>
<td>Please tell me about your experience when either you or your loved one was offered genetic testing.</td>
</tr>
<tr>
<td>Possible prompt</td>
<td>Please tell me about how you decided to either receive or decline genetic testing.</td>
</tr>
<tr>
<td>Possible prompt</td>
<td>Please tell me about how your loved one decided to either receive or decline genetic testing.</td>
</tr>
<tr>
<td>Possible prompt</td>
<td>Could you please describe the events that led up to your decision whether or not to receive genetic testing?</td>
</tr>
<tr>
<td>Possible prompt</td>
<td>What was going on in your life when this decision was made?</td>
</tr>
<tr>
<td>Possible prompt</td>
<td>How would you describe how you viewed genetic testing before this experience?</td>
</tr>
<tr>
<td>Possible prompt</td>
<td>What contributed to your decision whether or not to get genetic testing?</td>
</tr>
</tbody>
</table>

### Table 3. Practice, Research, and Policy Implications

<table>
<thead>
<tr>
<th>Implications</th>
<th>Strategies</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Practice</strong></td>
<td></td>
</tr>
</tbody>
</table>
| Individualized, tailored information | ➢ Explore how to incorporate decisional aids sooner in the decision-making process  
➢ Explore how to increase health literacy within rural communities surrounding hereditary genetic risk  
➢ Explore ways to navigate providers in the genetic testing referral process |
| **Research** |            |
| Descriptive | ➢ Explore diverse ethnic and socioeconomic backgrounds within context of genetic testing decision-making  
➢ Further explore the relationship of theorizing categories to develop theme |
| Assessment | ➢ Develop more theoretically based studies surrounding genetic testing decision-making process in context of hereditary cancer risk  
➢ Assess whether well-informed, quality-based decisions are being made within hereditary cancer risk patients in a rural environment |
| Intervention | ➢ Develop educational materials at appropriate health literacy level  
➢ Improve knowledge of hereditary cancer risk and recommendations among rural population |
| **Policy** |            |
| | ➢ Create policy to address limitations to current genetic risk information and national guidelines in rural environment  
➢ Increase education of providers in referral process  
➢ Mandating educational materials specific to rural communities that align to national standards |
V. Summary and Conclusions
The preceding manuscripts described three scholarship investigations undertaken by the author during her doctoral program. The combined outcomes of these three investigations contributes a unique perspective to the decision-making in the field of hereditary cancer risk. Manuscript One systematically reviewed and identified factors associated with utilizing \textit{BRCA}1/2 genetic testing to better understanding hereditary breast and ovarian cancer syndrome (HBOC) genetic testing decision-making. Its conclusion highlighted the need for future research to target social-ecological factors associated with decision-making in genetic testing from a theoretical perspective in individuals with HBOC. In turn, this conclusion served as a crucial piece for the justification in conducting the study described in Manuscript Three, which utilized social constructivism with a grounded theory approach within a rural environment.

Manuscript Two compared the use of several health behavior mid-range theories and frameworks commonly used among HBOC literature. Further explored the relationships of how concepts and frameworks were operationalized and used in the context of individuals at risk for HBOC seeking to undergo \textit{BRCA}1/2 genetic testing. Current HBOC literature lacks theoretically-based studies focusing on the decision-making process with a family context. Conclusions from this review highlighted that there is no one theory that is a “one size fits all” within the context decision-making and hereditary cancer risk. This is congruent with findings from the study described in Manuscript Three in which the decision-making process was considered complex and intertwined with many aspects of an individual’s life when making genetic testing decision. Results of Manuscript Two suggest a need to further understand the complexities of how mid-range theories and frameworks influence the decision-making process of individuals with HBOC during genetic testing, and this is crucial in adopting novel therapies so that we may tailor individualized interventions among this population.
Findings in Manuscript Three suggest that individuals with HBOC may experience a shifting of self during the decision-making process in utilizing genetic testing. Conclusions highlighted in Manuscript Three suggest individuals with HBOC may perceive a process by which they experience and live through a multitude of emotions, thoughts, and feelings that are intertwined with past, present, and future health-related decisions simultaneously, especially in those with a HBOC-associated cancer diagnosis at the same time. Individuals with HBOC are also exposed to a variety of internal and external cognitive, sociodemographic, and emotional factors that may influence their decision-making process. The shifting of self can be influenced by these factors and interactions with their loved ones, healthcare professionals, and surrounding community. Interventions that aim to improve the decision-making process in utilizing genetic testing among individuals with HBOC need to consider more tailored, individualized novel ways to incorporate more effective communication strategies about cancer risk and treatment options. Likewise, practice guidelines and institutional policies ought to incorporate provisions to address health literacy, the referral process, and mandating educational materials specific to rural communities that align to national standards.

The findings described in Manuscript Three suggest several priorities for future research. First, there is still a need for research that describes the experience of individuals at-risk for HBOC under-represented racial, ethnic, and socioeconomic groups. Although the author sought to recruit individuals in a rural environment in Manuscript Three, which was a novel component to this study, the sample was majority White and had a HBOC-associated cancer diagnoses. This was due to several unforeseen barriers such as several diverse ethnic and religious ‘micro-communities’ did not access cancer genetic services where the author conducted recruitment and data collection as well as low health literacy and poverty levels were identified. An example of
this includes several participants in the study described in Manuscript Three did not have email, access to electronics such as WIFI or personal transportation.

Replication of the current study in a region of the United States with more racially and ethnically diverse population is warranted; however, more efforts are needed now that these barriers have been identified to continue the exploration of the decision-making process in genetic testing among other rural populations with limited access to specialty care. Although familial and individual risk is a factor to determine eligibility for screening and testing, the need for this work is especially pressing given that individuals with hereditary cancer risk crosses a multitude of ethnicities, socioeconomic statuses, and age groups. Additionally, there are persistent racial, ethnic, and socioeconomic disparities in cancer (National Cancer Institute, 2017) and in hereditary cancer risk (Cragun et al., 2017). Similarly, previous studies have identified the need for more culturally sensitive efforts be tailored to health literacy levels, which may aid in increased genetic testing uptake (Salloum et al., 2018; Williams et al., 2018).

A second priority for future research entails the examination of the relationship between communication of providers and patients in identifying those at risk who meet eligibility for genetic screening and testing and referral to the appropriate cancer genetic services. Participants in the study described in Manuscript Three emphasized how their expectations of genetic testing did not meet the implications of the test once they received genetic counseling services. Participants also described situations where communication of genetic risk information from their providers led to misunderstandings, which led to implications in their decision-making for their future and their loved ones. The National Cancer Institute (2015) outlines the importance in good communication in cancer care, and the quality of information and support patients’ need in making well-informed decisions. This finding is consistent with decisional aids designed for
primary care providers, where user-centered tools are integrated into clinical workflow to assist providers in communicating risk to patients (Silverman et al., 2018). Further examination of similar decisional aids in specialty care surrounding genetic testing are needed. Patients diagnosed with an HBOC-associated diagnosis may not have a PCP or utilize primary care services, so it’s crucial we tailor decisional aids to include specialty genetic services from the beginning of the decision-making process so patients understand their risk and its implications for their loved ones. This examination may include the adoption new policies addressing mandating educational materials specific to rural communities that align to national standards, and addressing the noted limitations and barriers currently in genetic risk information. Exploration of these relationships will shed light on potential interventions to improve provider-patient communication among those with increased hereditary risk in a rural setting, which may improve the quality of care in early detection, prevention, and treatment strategies.

A final priority for research entails the development and evaluation of interventions that improve the experiences of individuals at risk for hereditary cancers in the utilization of genetic testing in rural settings. These interventions may include decisional, which are known to increase patient’s knowledge, participant decision-making, and decrease uptake rates for some elective procedures, which will allow patients to become more informed (Stacey et. al., 2018). Findings from the current study described in Manuscript Three discuss how use of decisional aids would complement individuals decision-making process regarding genetic testing. Yet understanding how shifting of self within the context of decision-making in genetic testing isn’t explicitly identified yet through current decisional implementation toolkits. Therefore, future studies are needed to further explore this core category during the decisional-making process in the utilization of genetic testing.
In conclusion, the body of work represented by this dissertation highlights the importance of research in acknowledging the need for more studies in exploring the decision-making process among those with increased hereditary risk in utilizing genetic testing. The experience is highly individualized and tailored to the area of which specialty care is being offered. A body of research focused only on identifying factors associated with whether to utilize genetic testing or not is not comprehensive enough to fully understand the process in which these complex, interwoven life altering decisions are made and the implications individuals with increased hereditary cancer risk encounter within a given timeframe. The study described in Manuscript Three represents the initial effort to explore these processes in decision-making within the context of a HBOC-associated cancer diagnosis in the utilization of genetic testing. The use of theory to drive the study described in Manuscript Three was to help explain, predict, and understand this phenomenon of interest in a rural environment, as this was lacking from the current literature. Further research using more theoretically-based studies is necessary to more fully understand how providers and patients can improve their decision-making processes, communication, and overall experience with genetic testing outcomes. As more research, clinical guidelines and standards are published within the field of genomics, nurses are more crucial than ever to disseminate and advocate for equity in deliver of genetic risk information, treatment options, and outcomes. The author of this dissertation is therefore committed to a lifelong program of research that aims to improve health and prevent illness by developing interventions that promote self-management behaviors that will enhance quality of life for families living with hereditary cancer risk.
References


