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Genetic Testing and the Power of the Provider: Women's Experiences with Cancer Genetic Testing

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Genetic Testing and the Power of the Provider:
Women’s Experiences with Cancer Genetic Testing

by

Dana Erin Ketcher

A dissertation submitted in partial fulfillment
of the requirements for the degree of
Doctor of Philosophy
Department of Anthropology
with a concentration in Medical Anthropology
College of Arts and Sciences
University of South Florida

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DEDICATION

Dedication is needed for anything that takes over 5 years to complete. I certainly dedicated myself to this endeavor, but it was also the dedication of multiple individuals over these many years that helped me accomplish this goal. First, my family, who provided inspiration, help, and sanity breaks when I desperately needed them. I love you all so much, thank you for always being there for me.

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ABSTRACT

Genetic testing has become ubiquitous in contemporary society, from determining ancestry to addressing health concerns. This dissertation focused on a qualitative, feminist approach to understand women’s experiences of genetic testing for hereditary cancer syndromes, as well as their perspectives of risk. A total of 33 participants agreed to a semistructured interview and drawing of their family tree (pedigree). Eleven (40.7%) participants had been diagnosed with breast cancer, and 16 (59.3%) participants with ovarian cancer. Thirty-one (93.9%) participants had genetic testing, and of those, 17 (54.8%) had genetic counseling. Participants voiced several reasons why they wanted to undergo genetic testing or how they planned to use genetic test results, including: to obtain information for their family members, for cancer treatment options, for surveillance of other cancers, and/or for more information about their disease. Family was the most frequently discussed theme, but surveillance and treatment discussions highlighted important areas for provider communication improvement. Participants were also asked about how their reaction to their genetic test results, which ultimately underscored the importance of the provider in pre-test genetic counseling. Women who received positive test results (n=5, 16.1%) with a non-genetics professional (NGP) reported largely negative reactions, while women who received positive results (6, 19.3%) with a genetic professional (GP) reported much more relaxed reactions to their test results. Eighteen (58.1%) participants received negative test results, and many reported “mixed feelings” about their results, regardless of the type of provider they saw. While provider type did not seem to affect the way women in this group viewed their test results, their reactions point to important questions about how risk and ideas of genetic determinism are discussed with patients. When asked about risk, participants described constant worry around their cancer risks, and the ways in which they attempted to control their perceived risks. This dissertation research provides an in-depth understanding of women’s experiences and aspirations for genetic testing, which can be used by providers to improve the process of genetic testing and counseling. Among the many findings, I
argue that male-dominated, biomedical hegemony can be seen in the patterns of care experienced and reported by the women in this study. This research contributes to the literature on women’s health, cancer, genetic testing, and biomedicine, and is meant to be applied to improve the outcomes of the many women who undergo genetic testing.
CHAPTER ONE:
INTRODUCTION

Introduction

Never before in our society has genetic testing been so omnipresent. Commercials for Father’s Day offer discounts for dad (and thus you partly, the biologically related gift giver) to find out his genetic ancestry. Celebrities write about their genetic testing experiences and consequent prophylactic surgery choices. Genetic testing is so easily available, you can pick up a genetic testing kit at your local pharmacy while you pick up some sunscreen. At first glance, this appears to be a democratization of genetic information, making health and ancestry information made easily available to the masses. Upon closer inspection, we start to see the cracks in the slick advertising – for instance, the persistence of health disparities to genetic services (Manrriquez et al. 2017) and the consequences of giving genetic information to private companies (Wood 2018). Incidentally, there is always more to the story when we look closer. In this case, we will examine genetic testing for hereditary cancer syndromes.

This research study sought to understand the impacts that genetic information can have on individuals and families. More specifically, how are women understanding, using, and interacting with the world of cancer genetic testing for hereditary cancer syndromes? Before 2013, I knew nothing of genetic testing for cancer syndromes. That changed once I became a research assistant at a NCI-Designated Comprehensive Cancer Center1 in the U.S. Southeast in the summer of 2013, immediately before entering the PhD program at the University of South Florida. Between July 2013 and March 2017, I worked on a study which focused on hereditary cancer; this study assessed participants longitudinally through long questionnaires. These assessments were purely quantitative, capturing items like the frequency and type of

1 National Cancer Institute (NCI) Designated Comprehensive Cancer Centers are recognized for their scientific leadership and transdisciplinary research in basic, clinical, and/or population science.
cancer screenings, new cancer diagnoses, and prophylactic surgeries. Not only did I speak to the participants in this study on a regular basis, but I was able to work closely with genetic counselors and geneticists, attend Genetic Case Conferences, and observe genetic counseling appointments. And what a time to be a part of cancer genetic testing. As will be discussed further, 2013 was a pivotal year for cancer genetic testing options. Not only did I get a “peek behind the curtain” of how genetic counselors and other genetics practitioners dealt with specific patient cases, laboratories, and various testing options, but I witnessed in real time the impact of a Supreme Court ruling which changed the face of genetic testing options rapidly.

While genetic testing can include a wide range of options, with ancestry testing being the most easily available and most publicized, cancer genetic testing is typically only available to individuals who meet specific criteria. The U.S. Preventive Services Task Force (USPSTF) actually recommends against routine genetic counseling and/or BRCA\(^2\) testing for women who do not fit risk criteria because it was determined that “there is a moderate or high certainty that the service has no net benefit or that the harms outweigh the benefits” (Moyer 2014:282). But once women have been properly assessed and are determined to meet testing criteria, what happens during and after that encounter? There are numerous recommendations about what information should be covered in the genetic testing/counseling (GT/C) experience (Berliner et al. 2013, Berliner and Fay 2007, Moyer 2014), but what is actually covered, and what is remembered and implemented, are two different things. In extreme cases, this can have serious physical and emotional repercussions, as the story of one Oregon woman illustrates. Elisha Cooke-Moore was told by doctors that because of her positive genetic test results, she was at higher risk for ovarian and breast cancer. She subsequently underwent a prophylactic double mastectomy and hysterectomy. After being unhappy with the mastectomy results, she consulted another doctor who, upon review of her records, discovered that she had in fact not tested positive for any genetic mutations (Ducharme 2017).

Although this is an extreme example, it highlights several issues that are important when discussing the GT/C experience. First, the article states that Cooke-Moore’s doctors were the ones who misread the results. It never mentions a genetics professional or genetic counselor being present to order, explain, or

\(^2\) BRCA is an abbreviation for BReast CAncer gene. There are two genes under the BRCA umbrella: BRCA1 and BRCA2.
handle the test results. As I argue in this dissertation, the experience between seeing a genetic
counselor/genetic practitioner (genetic professionals, GPs) for GT/C is very different than the GT/C
experience of a woman who gets their testing through other medical practitioners (non-genetics professionals,
NGPs). Second, Cooke-Moore’s results actually showed a variant of uncertain significance (VUS) in the
MLH1 gene. With the advent of next-generation sequencing, genetic testing now can test more genes faster
and more cheaply than ever before (Blazer et al. 2015). While this may be considered a pro, there are cons to
consider as well – one being the increased chance for returning a VUS as a result. While most genetic
counselors tell their patients that a VUS will most likely be reclassified as a negative once more information is
gathered on the variant, Cook-Moore’s original doctors did not counsel that way. Third, this story highlights
the complete trust that many patients place in the medical system. Medicine and genetics can be highly
complicated subjects, and therefore patients may not question someone who is supposed to be an expert in
those fields. While this is typical of a biomedical system, I argue that significant differences exist between GPs
and NGPs in allowing a space for questions and understanding what testing means for an individual. Finally,
Cook-Moore’s story is also about the importance of family. Cook-Moore’s mother and grandmother had
cancer, which prompted her to seek out genetic testing in the first place. Because of the hereditary nature of
some breast and ovarian cancers, family is a strong component and motivation for genetic testing for almost
all of the women in this study as well.

Testing for BRCA has been commercially available since 1996. Since then, the subjects of genetic
testing, genetic counseling, and provider and patient knowledge has been explored to varying degrees. For
this study, the use of anthropological methods and theory helps shed new light on the ways in which cultural
values, biomedical hegemony, and provider power can construct patterns of care during GT/C. In all, 33
women were interviewed and their family trees (pedigrees) drawn. While women can and have been
quantitatively assessed on their GT/C experience in other studies, these results are usually determined
immediately after the GT/C encounter. More importantly, past research has focused on people who have
testing through some sort of GP – either a geneticist or genetic counselor – rather than other medical
providers such as OBGYNs or oncologists. Thus, this research adds to the literature of women’s perceptions
and recollections of the GT experience with NGPs as well as how they how women conceptualize cancer risk and the impact this perceived risk has on their lives.

Research objectives

What follows is a list of specific research objectives of this study, along with the methods utilized to address each of them:

(1) To document both the history of and current efforts of genetic counselors and other genetics professionals around cancer GT/C. This includes the rapidly changing landscape of GT/C due to next-generation sequencing. *This involved conducting archival research and current literature searches.*
   a. This effort was also informed by over three years of immersion as a research assistant at an NCI-designated Comprehensive Cancer Center, with attendance at Genetic Case Conferences to discuss genetic counseling cases, new testing techniques, and conversations with both genetic counselors and participants in research studies.

(2) To determine women’s views and experiences with genetic testing and counseling. *This involved conducting semi-structured interviews with 33 women determined to be at high genetic risk for cancer, in which three groups were posited to emerge:*
   a. Women who chose to undergo GT/C: *specifically, what did they find useful about GT/C? What was not useful? What did they remember about their experience?*
   b. Women who refuse GT/C: *specifically, why did they refuse? How do they conceptualize GT/C?*
   c. Women who were not given the option to undergo testing (was not offered, were not aware of testing as an option): *specifically, how do they conceptualize GT/C?*

(3) To determine women’s views of genetic testing and counseling, and current genetic testing options/availability. *This involved conducting 33 semi-structured interviews.*

(4) To determine how being labeled ‘high-risk’ for cancer affects women. *This was also assessed during semi-structured interviews with women.*
a. This has been a question identified as an important area of study by the U.S. Preventive Services Task Force since “[w]hat happens after patients are identified as high-risk in clinical settings is unknown” (U.S. Preventive Services Task Force 2013).

As with any research, objectives shifted and changed slightly based on the answers of participants. For instance, the posited groups (b and c) in objective (2) did not emerge. As such, the original research objectives have been slightly modified as follows. The first (1) objective is partly addressed in the Chapter 2 and Chapter 3 during the discussion of the literature review and researcher positionality. Before conducting interviews, I believed that newer genetic testing options such as panel testing (i.e., testing multiple genes simultaneously) through next-generation sequencing would be something participants would be aware of and have opinions on. However, during the course of interviews it became clear that while some participants had recently undergone panel testing, it became apparent that this was not necessarily something they knew or cared about. While I asked participants about newer genetic testing options, responses were scarce and did not provide much in the way for analysis.

The second (2) and third (3) research objectives turned out to be duplicates, as I was unable to find participants who refused GT/C or who were not given the option to undergo GT/C. As such, they have been condensed into one research objective which aims to broadly understand women’s views and experiences of GT/C and are dealt with extensively in Chapter 4. The fourth (4) research object was open-ended and exploratory, seeking to understand how women understand the label of “high-risk” due to their matching of criteria to undergo GT. Again, this objective is address in Chapter 4.

**Organization of the dissertation**

Following this introduction chapter, Chapter 2 will review the literature and provide theoretical orientations. This research is situated at the nexus of a variety of subfields and disciplines; therefore, this chapter covers topics such as oncology care in the United States, a brief history and primer of genetic testing and counseling for cancer syndromes, and information on breast and ovarian cancer in the United States. I also discuss how feminist theory and critical medical anthropology are important within the context of this
research. Chapter 2 connects the microlevel, individual lived experience that this dissertation research addresses with a macrolevel understanding of healthcare and cultural attitudes. Chapter 3 discusses the methodology used to conduct the research, as well as the justifications for the use of these methods. Immersion through research work at an NCI-Designated Cancer Center, hereditary cancer conferences and local meetings, as well as semistructured interviews with 33 participants, provide the basis for the methodology and results. Chapter 3 also presents basic demographics about the participants in this study and descriptive statistics derived from participant pedigree data.

Chapter 4 presents the results of the semi-structured interviews which addresses research objectives (2) + (3) as well as (4). In this chapter, a thematic analysis of women’s motivations and aspirations for their genetic testing results is presented and focuses on themes of family, cancer treatment options/uses, medical surveillance, information/research, and Ashkenazi Jewish heritage. Through qualitative analysis, important conceptual distinctions between participants and the medical community were found. In addition, Chapter 4 covers women’s reactions to their genetic testing results and is divided into groups based on their genetic test results (positive or negative) and what kind of provider ordered testing (GP or NGP). While women who had a positive result and had testing through a GP reported ambivalent reactions, women who had a positive result and had testing through an NGP reported negative emotions and reactions. Women who received negative test results reported similar reactions to their results regardless of the provider they saw. However, the results discussed in Chapter 4 also suggest that negative results should be interrogated with women more than they traditionally are. Finally, Chapter 4 covers women’s conceptualizations of risk. Cancer risk presented a worry that almost all participants voiced. Participants discussed their ideas of how cancer risk always seemed to be looming and spoke about ways in which they hoped to control risk, which is very much informed by cultural notions of risk and individual responsibility.

Chapter 5 provides a discussion of the study findings, incorporating results from this study to the extant literature. Results from the semi-structured interviews add to the existing literature on the importance of genetic counseling prior to undergoing genetic testing. Additionally, this research adds to a previously understudied area: that of women and their testing experiences with NGPs. Implications for future research,
contributions to theory, as well as study limitations, are also discussed. Finally, Chapter 6 offers some conclusions and future recommendations. As an applied research project, I see application of this data to improve outcomes in two main areas: during the actual act of genetic counseling, and when NGPs order genetic testing. I argue that while genetic counseling is incredibly important both before and after undergoing genetic testing, changes can and should be made to ensure that the needs of people who undergo testing are being met fully.
CHAPTER TWO:
LITERATURE REVIEW & THEORETICAL ORIENTATION

Introduction

In this chapter, I lay the ground work for topics and theoretical orientations that I believe are important in understanding the context of this research. This includes a brief overview of the practice of oncology and genetic counseling, data on breast and ovarian cancer, and hereditary cancer and cancer-associated genes. This research is situated at the nexus of many fields and medical specialties outside of anthropology and relies on a variety of literature sub-specialties.

Two critical theoretical fields inform this research: feminist theory and critical medical anthropology (CMA). In their own specific ways, both of these theories help researchers interrogate the ways in which complex hierarchies are reproduced based on categories such as sex, class, race, and gender (Disch and Hawkesworth 2015). Theory helps us decide what data to collect, how to give meaning to data, prioritizes some data over others, and influences how we interpret our data (McGee and Warms 2017). Using these theoretical frameworks, I seek to interrogate the biomedical hegemonic paradigm, which is itself a folk model of illness and health, which is therefore subject to sociocultural forces that ultimately impact the kinds of care women receive (White 2005).

Theoretical orientations

Feminist theory

This research broadly addresses women’s health in an oncology setting, specific to genetic testing and counseling for hereditary breast and ovarian cancers. While this research relies on women’s voices and experiences and is about cancers that either primarily or exclusively affect women, that in itself does not make this feminist research. Critical feminist research and theory reminds us that sex and gender are political
constructs by which there are ways of privileging some and disadvantaging others (Disch 2015:4). Davis and Craven (2016) explain that while power dynamics are important, feminist ethnographic work must also include a gender analysis which ultimately “takes into account all people in a field site/community/organization, and pays particular attention to gender by honing in on peoples’ statuses, the different ways in which (multiple) forms of privilege allow them to wield power or benefit from it, and the forces and processes that emerge from all of the above” (Davis and Craven 2016:9). An analysis of gender is particularly important in biomedicine, which in almost every way is and has been androcentric, reflected in who its practitioners are (Young et al. 2015) to the default human subject as a 70-kg male (Clayton 2015). In medical discourse, women have been portrayed as the sick or incomplete version of men, the constant (and opposite) Other to the strong, healthy male body (Lupton 2012).

Under biomedical hegemony, ways of knowing, especially what is considered authoritative knowledge, is used and privileged differently (Browner and Press 1996). One of the earliest examples of the privileging of scientific knowledge over experiential knowledge in medicine was the wresting of power away from midwives to male practitioners in the 19th century (Lupton 2012). A current example exists in the hierarchy of knowledge between patients and doctors today. Doctors and technology are the apex of the authoritative knowledge hierarchy, with patients and their embodied knowledge and experience at the bottom (Browner and Press 1996). Biomedicine tends to reflect and prioritize culturally valued “male” characteristics such as rationality and lack of emotion over culturally “female” characteristics of embodied knowledge and experience. Understanding how gendered diseases such as ovarian and breast cancer are dealt with under a biomedical paradigm that has a long history of devaluing the female makes a feminist theoretical approach absolutely necessary. Considering the relatively short history of attention to gender in health research and policy, which started in the early to mid-1980s (Read and Gorman 2010), research has found plenty of evidence of gender bias not only when treating diseases but among female medical professionals who recount discrimination and harassment (Risberg, Hamberg, and Johansson 2006). The privilege of the male over the female in biomedicine makes a gendered analysis and feminist theory critical.
A gendered analysis also forces us consider the biological diseases that are being studied; in this case, breast and ovarian cancer. While breast cancer does affect men on rare occasions, breast and ovarian cancer are generally seen as a woman’s disease, and thusly gendered, with ovarian cancer being the most exclusively gendered. Feminist health activism in the 1980s pushed to make breast cancer less stigmatized and successfully lobbied to increase federal funding (Grigg and Kirkland 2015). Breast cancer is now so destigmatized that it is a cause that even the (masculine) mainstream can get behind. For example, from 2009-2016, the National Football League (NFL) players wore pink during games in October to raise awareness and funds for breast cancer in conjunction with the American Cancer Society (Vrentas 2016). Ovarian cancer, on the other hand, is a disease that lacks effective screening mechanisms, where women struggle to get diagnosed, and has relatively high levels of mortality (Henderson, Webber, and Sawaya 2018). An analysis of gender not only assesses biomedical hegemony but the differences and similarities between these two gendered cancers.

Finally, in pursuing this work I follow three principles set forth by Hall and Stevens (1991:17) and of which I aim to incorporate into this work. While Hall and Stevens note that “feminisms” must exist because women’s experiences are multitude, they contend that there are three principles that feminist research can attend to: (1) valuing of women and a validation of women’s experiences, ideas, and needs; (2) recognition of the conditions that oppress women; and (3) a desire to bring about social change on behalf of oppressed people through political action (Leipert 2001).

**Critical Medical Anthropology (CMA)**

Critical medical anthropology (CMA) initially took cues from the viewpoint of political-economy and attempts to address various topics such as the social origins of disease under economic systems, health policy, medical pluralism, and the critique of biomedical ideology, practice, and structure (Singer 1989:1196). Various approaches can be made when deploying CMA, and I integrate a few approaches to accomplish my research objectives. First, a critique and understanding of biomedicine is essential for this research, for that is the

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3 In the United States there will be an estimated 2,550 new cases of male breast cancer in 2018, with an estimated 480 deaths (American Cancer Society 2018b).
paradigm this research is conducted under. In critiquing biomedicine, my aim is to engage “with issues of power, legitimacy, and discrimination, in revealing what risks to health are embedded in social structures” (Panter-Brick and Eggerman 2018:234). We can see the results of power, social structures and individuals in the idea of struggling. In this data, struggling was ubiquitous – for diagnosis, for treatment, to be heard by practitioners, to stay healthy, to understand genetic test results. Others have also identified that a “key component of health is struggle” (Baer, Singer, and Johnsen 1986:95). These struggles, while individual, must be understood within systems. Taking a “people-centered approach” to incorporate the lived experiences of women undergoing genetic testing is another facet of CMA (Panter-Brick and Eggerman 2018). This research reflects a strength of anthropological research: to capture the microlevel, of women’s cancer and GT/C lived realities, and situate that experience within larger macrolevel medical and cultural structures (Closser and Finley 2016).

While CMA has its roots in political economic approaches, I use CMA as a way to investigate and “connect the social group…to the larger regional, national and…human society…to the configuration of social relationships that contribute to the patterning of human behavior, belief, attitude, and emotion” (Singer 2004:24). This vertical connection from microlevel patient experiences and provider interactions to macrolevel structures that ultimately shape these experiences and beliefs is what I wish to elucidate. Further, I take seriously the critique from Nancy Schepker-Hughes (1990) that rather than be complicit in perpetuating, or being subservient to biomedical hegemony, critical medical anthropologists should “play the court jester, that small, sometimes mocking, sometimes ironic, but always mischievous voice from the sidelines” (Schepker-Hughes 1990:195). I believe that it is the job of anthropology to question the motives and practices of those in power. While Schepker-Hughes’ analogy may seem light-hearted, I take the suggestion seriously. CMA requires that I interrogate systems of power, which in this case is biomedical hegemony. Additionally, consideration was given to a critique by Estroff (1988) that biomedical hegemony can sometimes be written of as a bounded system, in that it is separate from culture and individuals. In this case, I see the connection of the individual/social group to macrolevel forces as a two-way street, in that individuals, both patients and providers, show agency in how they resist or conform to larger cultural ideals of biomedical thought.
Cancer and Healthcare in the United States

All of the participants in this study received cancer treatment and/or genetic testing in the United States. As such, a basic understanding of the healthcare system, and context for oncology care, is needed.

The United States Healthcare System

The United States, unlike all other high-income countries, does not have universal health care. A tangled web of public and private insurance coverage, which is market-based and for-profit, has resulted in the highest health spending per capita in the world, with middling to low health outcomes (Castañeda and Mulligan 2017:3-4, OECD 2018). By 2019, the Centers for Medicare and Medicaid Services projected that US healthcare spending will reach $4.3 trillion and make up 19.3% of the national GDP (Schnipper et al. 2015). Cancer care is one of the fastest growing areas of healthcare, increasing in cost from $125 billion in 2010 to $158 billion in 2020 (Schnipper et al. 2015). The Affordable Care Act (ACA), passed in 2010, has attempted to address some of these issues and has led to a decline in the uninsured population by 43%, with improvements in access to care and financial security (Obama 2016). Additionally, the ACA also had provisions for genetic testing for BRCA in its preventive health services benefits (Walcott et al. 2014).

Unfortunately, healthcare in the United States is typically centered around interventions at the level of the individual, which may not have broader impacts at the population level (Long 1998). In this sense, healthcare in the United States may be thought of more as “sick-care”, which addresses the individual when they are sick rather than prioritizing prevention activities to keep them healthy.

After 1900, the United States shifted from a largely medical pluralistic society to one that gave priority economically, politically, and socially to germ theory and “scientific medicine” (biomedicine) (Baer 2004). While medical pluralism still exists (such as osteopathic medicine, chiropractic, New Age, folk medicine), biomedicine ranks first in prestige in relation to these other medical systems (Baer 2004).
Oncology and Cancer Care in the United States

A reflection of cancer care in the United States is seen in the development of the American Society of Clinical Oncology (ASCO). Started in 1964 by seven cancer physicians, it has grown to almost 45,000 members today and has increased funding for cancer research from less than $200 million to more than $5 billion every year (American Society of Clinical Oncology 2018). In 1971, President Nixon signed the National Cancer Act, and while he never uttered the phrase, it soon became known as the “war on cancer” (Sporn 1997, Marshall 2011). One of the goals of the National Cancer Act included a massive overhaul of contemporary scientific and medical knowledge, with an influx of money to help achieve this goal: from $190 million in 1970 to more than $600 million in 1975 (Maugh and Marx 1975). At the time, cancer knowledge and the research outlook was somewhat pessimistic. In the preface to their book, Maugh and Marx (1975) wrote:

“There is little prospect of an immediate cure for cancer. The seeds of destruction seem to be sown within all of us, and there is no consensus about how these seeds develop into tumors or what can be done to halt that development. Indeed, it is often difficult to find a consensus about any aspect of cancer research. The sharply conflicting views of investigators in different subdisciplines has been most aptly summarized by Charles Heidelberger of the McArdle Laboratory for Cancer Research, who argues that ‘the mechanism of cancer is a mirror into which each man looks and sees himself.’ This is true despite the fact that large infusions of money and manpower now come to the aid of cancer research.”

More recently, the call to “cure cancer once and for all” was renewed when President Obama signed a Presidential Memorandum in early 2016 creating the White House Cancer Moonshot Task Force (Wang 2016). More popularly known as Cancer Moonshot 2020, the program seeks to develop cancer vaccines, immunotherapy, and combination therapy; enhance data sharing; develop therapies for pediatric cancer; and perform genomic analysis of cancer tumors (Wang 2016). The 21st Century Cures Act, passed by Congress in
December 2016, provides $1.8 billion of funding for Cancer Moonshot over 7 years (National Cancer Institute 2018a).

While awareness and funding has grown and changed significantly since the 1970s, cancer has yet to be completely controlled or eradicated. The overarching cancer goal for Healthy People 2020 is to reduce the number of new cancer cases, as well as the illness, disability, and death caused by cancer (National Center for Health Statistics 2016). Under this broad umbrella goal were 27 more specific objectives such as reducing cancer death rates (i.e. lung, female breast, colorectal, prostate, melanoma), increasing the proportion of cancer survivors living 5 years or longer, and increasing the proportion of adults receiving cancer screening. During the midcourse review of Healthy People 2020, it was determined that most objectives were improving (n=9, 37.5%) or had been met or exceeded (5, 20.8%), while some were getting worse (3, 12.5%) or had little to no detectable changes (5, 20.8%) (National Center for Health Statistics 2016). Interestingly, the only three objectives that are getting worse are screening measures for women (Table 1).

Table 1. Midcourse progress for measurable cancer objectives from Healthy People 2020*

<table>
<thead>
<tr>
<th>Objective description</th>
<th>Baseline Value (year)</th>
<th>Midcourse Value (year)</th>
<th>Target</th>
</tr>
</thead>
<tbody>
<tr>
<td>Women receiving a Pap test within past 3 years</td>
<td>84.5% (2008)</td>
<td>80.7% (2013)</td>
<td>93.0%</td>
</tr>
<tr>
<td>Women counseled about mammograms</td>
<td>69.8% (2008)</td>
<td>61.4% (2010)</td>
<td>76.8%</td>
</tr>
<tr>
<td>Women counseled about Pap tests</td>
<td>60.2% (2008)</td>
<td>53.9% (2010)</td>
<td>66.2%</td>
</tr>
</tbody>
</table>

*adapted from National Center for Health Statistics (2016)

While overall cancer deaths have improved significantly since 2007 (National Center for Health Statistics 2016), other issues exist in regards to access to treatment. For instance, patented cancer drug prices in the United States have increased 5- to 10-fold from 2000 to 2014; in 2012, 92% of the new drugs approved for cancer were priced above $100,000 per year for therapy (Kantarjian et al. 2014). There are still large disparities in cancer death rates, with the rate being 15% higher in blacks than whites (based on 2014 data).
Although the racial gap is narrowing for some cancers, it is widening in others, such as breast cancer in women (DeSantis et al. 2016).

In attempting to understand the cultural forces that shape healthcare in the United States, which is a biomedical paradigm, it also helps to see how biomedicine is shaped from within. Del Vecchio Good et al. (1990) argue that “American oncology has its own ‘local’ culture”, which relies primarily on the symbol of “hope” (Del Vecchio Good et al. 1990:60). Coreil et al. (2012) similarly describe the dominant cancer narrative in the United States as one that centers around optimism and personal transformation through adversity. This seems to be in direct contrast to previous centuries and decades (and some current practices cross-culturally), where, “[R]evealing the diagnosis to a patient was considered cruel and inhumane because the patient would lose all hope and could cope better not knowing. This was viewed as an acceptable ‘white lie,’ although the patient’s family was always told” (Holland 2002:207). This “American discourse on hope”, I argue, is reflected in two important concepts – biomedicine and neoliberalism – which are important features to consider in the landscape of healthcare in the United States.

Biomedicine

This research follows Kleinman (1997) who uses biomedicine over “Western medicine” in order to underscore the “institutional structure of the dominant profession of medicine in the West, and today worldwide, while also conjuring the primacy of its epistemological and ontological commitments” (Kleinman 1997:25). As a sociocultural system, three main features of biomedicine exist for interrogation: as a domain of knowledge and practice, as a place of labor division and rules of/for action, and the means by which is it produced and altered (Gaines and Hahn 1985, Good 1995). Indeed, as a structural institution it is responsible for generating knowledge and training practitioners into this scientific paradigm, so while we may think of biomedicine broadly, it is important to understand that, in practice, it is a polymorphous ethnomedical system much like the numerous other ways in which humans practice medicine across the world. This is an important distinction as biomedicine, which is highly reliant on science, often holds the implication that it is belief and value free, and even acultural (Gaines and Davis-Floyd 2004). This could partly be due to the idea
of Cartesian dualism that so drives biomedical thought, with a strict (artificial) separation of mind/body, nature/culture, matter/spirit, and real/unreal (Scheper-Hughes and Lock 1987). However, through anthropological critique, biomedicine can be understood like any other ethnomedical system. In this case, the biomedical paradigm relies on technological advances to take humans beyond the limitations of biology or nature, putting emphasis on the body and the “real” and divorcing biomedical treatment from the person (Gaines and Davis-Floyd 2004).

There are two main aspects of biomedicine I have identified as important for this research context. First, feminist scholars critique biomedicine because it idealizes the male body as the epitome of function, and the female body as that of dysfunction (Gaines and Davis-Floyd 2004). Images of the male body still predominate major anatomy textbooks and visually support stereotypical gendered emotions, roles and settings (Parker, Larkin, and Cockburn 2017). Echoes of this may translate into direct, misogynistic actions during physician care, as described by Hahn (1985). In his “Portrait of an internist”, Hahn describes interactions of an internist with his female patients that suggests a “lack of intelligence” among his female patients, and calls them “Honey” or “Sweetie” (Hahn 1985:64; 103). Margaret Lock (1985) documents how menopause became medicalized by a system that is both condescending and paternalistic to female patients, where blame for the problems of menopause fall squarely at the feet of women and their attitudes about expected female roles (Lock 1985:116). A more recent look at the way women are treated in the medical system highlights many ways in which women’s symptoms are downplayed or simply not believed, such as labeling women as chronic complainers or giving anti-anxiety medication for heart attacks because women’s symptoms do not fit the “normal” presentation of disease (Dusenbery 2018). The idea of women having to perform their pain in certain ways to have providers believe that their chronic pain is real is something Amy McKiernan explores in her work with female fibromyalgia patients (McKiernan 2018). The treatment of women within a biomedical system can have repercussions during provider interactions, and thus healthcare outcomes, and therefore makes the culture of biomedicine an important foundation for this research.
Second, biomedicine focuses on that of the material, with a noticeable “discomfort with dialectical modes of thought” (Kleinman 1997:29). Dialectical in this case would be two people (e.g. the patient and the physician/medical practitioner) holding differing viewpoints (e.g. embodied knowledge and authoritative knowledge) but hoping to establish some sort of truth through discussion. With this kind of power dynamic, can true understanding ever be established? This discomfort with the dialectical is important in all areas of medicine but is especially true when we consider areas of medicine that overlap with the transmission of knowledge, such as genetic counseling. Rayna Rapp, while working with women and prenatal diagnostic technologies in the 1980s, perfectly states the uncomfortable position that patients may find themselves, stuck between biomedical authoritative knowledge and their own lived experience. She states that, “I was struck by the difficulties women had in working in a communicative system whose vocabulary was almost exclusively medical, whose grammar was technological, and whose syntax was as-yet unnegotiated” (Rapp 1998:47). In the case of the present research, I am similarly concerned with how women, when placed in a biomedical setting and confronted with a genetic testing technology and communication, make sense of that situation given the unfamiliar, and sometimes constraining, language and actions of biomedical hegemony. In the larger scheme, patients must make sense of this in order to successfully negotiate the system. There are pulls within biomedicine in seemingly opposing directions between genetic counseling for cancer syndromes (e.g. dialectical, narrative) and DNA (e.g. nature) which are particularly relevant.

Neoliberalism

Neoliberalism is a political-economic system, inexorably intertwined with globalization, which can be simplified to three core tenets: individualism, free market via privatization and deregulation, and decentralization (McGregor 2001), which more specifically deals with ideas of risk, governance, and responsibility (Ericson, Barry, and Doyle 2000). The history and breadth of neoliberal policies around the world is beyond the scope of this dissertation. Briefly, in the late 1970s and early 1980s, world leaders such as Deng Xiaoping in China, Margaret Thatcher in Britain, and Ronald Reagan in the United States implemented neoliberal economic policies which still reverberate globally today (Harvey 2007). In the United States, Reagan’s neoliberal policies resulted in a massive shift (and massive cuts) of funds from health and human
services, a systematic program of deregulation, and the support of “starving out” public programs in order to shift people to private entities (Terris 1999). When a neoliberal agenda is applied to a health care setting, this can mean “cost cutting for efficiency, decentralizing to the local or regional levels rather than the national levels and setting health care up as a private good for sale rather than a public good paid for with tax dollars” (McGregor 2001:83).

Privatization, deregulation, and decentralization are all important factors that can have direct impacts on individuals. However, when we look at the individualism tenet, it starts to become clear how a political-economic policy augments cultural ideals and practice, especially with health. Political-economic systems are obviously underpinned by cultural values, ideals, and norms. For example, neoliberalism highlights the individual, which thus impacts how we view citizens to be “good” or “bad” when it comes to health. Rose Galvin (2002) argues that the importance of the individual, which thus makes health primarily a personal responsibility, can impact how we view people with chronic illnesses. In a neoliberal society, a good citizen is “someone who actively participates in social and economic life, makes rational choices and is independent, self-reliant and responsible” whereas those with chronic illnesses are moral failures because they should have managed their health risks, and therefore need intervention (Galvin 2002:108). Self-care, then, can be viewed as a sort of disciplinary power of neoliberalism and enacted in biomedical settings (Leontini 2010). The individual must be a “producer, consumer, and citizen, as a social being as well as individual who cares for herself, her health, body, mind, and soul” (Freeman 2011). Specific to women in this study, we see how risk discourses, along with the acts that are strongly suggested/required to reduce cancer risk, are situated within a neoliberal society which assumes personal choice for health behaviors. In this context, women “are morally responsible for being aware of the risk factors and for taking steps to reduce them” (Dubriwny 2012:40).

Understood within a neoliberal context, risk-reducing actions may seem like a moral imperative to women because they are ensconced in an American healthcare culture that values patient autonomy and individual control over health, as well as a biomedical paradigm which does not leave room for questioning (Browner and Press 1996, Eisinger et al. 1999). The emphasis on individual responsibility to reduce or manage risk is evident in a number of studies situated in biomedical paradigms (Robertson 2001). Press,
Fishman, and Koenig (2000) likewise argue that our society exists under a twin paradigm of risk and surveillance, where risk factors are inherent in the individual, which increases the need for surveillance, prevention, and self-determination – the onus completely on the individual (Press, Fishman, and Koenig 2000:241, Stemerding and Nelis 2006). Risk-reducing action, whether through various “self-care” activities, or prophylactic surgeries to reduce the chance of developing cancer, are better understood when the principles of neoliberalism are applied to cultural values of health. In sum, neoliberalism as a political-economic policy has had cultural ramifications, impacting how we perceive our own healthcare responsibilities, the health of others, and the how healthcare providers deal with patients.

**Hereditary cancer-associated genes and genetic testing technology**

The overall cancer burden worldwide is classified as sporadic, with only 5-10% of cancers estimated to be inherited or hereditary (Nagy, Sweet, and Eng 2004). Characteristics of families with inherited cancer susceptibility include two or more relatives with the same type of cancer on the same side of the family, several generations afflicted, earlier ages of cancer diagnosis (typically under 50 years old for adult-onset cancers), or individuals with multiple primary cancers (Nagy, Sweet, and Eng 2004:645). Most hereditary cancer syndromes are autosomal-dominant, meaning that a patient’s first-degree relatives (parents, children, and siblings) will have a 50% risk of carrying the mutation as well (Rahner and Steinke 2008) due to a germline mutation. Autosomal-dominant hereditary cancer syndromes include hereditary nonpolyposis colorectal cancer (HNPCC, mutations in genes MSH2, MLH1, MSH6, or PMS2); familial retinoblastoma (mutation in RB1); and Li-Fraumeni syndrome (mutation in TP53) (Rahner and Steinke 2008). After the discovery of the BRCA1 and BRCA2 genes in the early 1990s, which are associated with hereditary breast and ovarian cancer syndromes, more than 200 other hereditary cancer susceptibility syndromes have also been discovered (Nagy, Sweet, and Eng 2004).

Critical to this research are the BRCA1 and BRCA2 genes, in which mutations give rise to what is known as hereditary breast and ovarian cancer (HBOC) syndrome (Shannon and Chittenden 2012). In 1990, after 17 years of research, the existence of a gene that predisposed some individuals to breast cancer was
found on chromosome 17q21 (Hall et al. 1990). BRCA1 and BRCA2 are tumor suppressor genes which are critical to maintaining genomic stability and preventing tumor development (Lindor et al. 2008, Mersch et al. 2015). Between 1 in 150 to 1 in 800 people in the general population carry a genetic susceptibility to breast cancer (Garber and Offit 2005). While breast and ovarian cancer are obviously the main cancers of concern with these mutations in these genes, pancreatic and prostate cancer screening is also recommended because of increased cancer risks (Mersch et al. 2015). The cancer risk associated with mutations in these genes has evolved over the years as more data has been gathered, but a recent prospective cohort study of almost 10,000 female BRCA1 and BRCA2 carriers provides updated cumulative cancer risks (Kuchenbaecker et al. 2017). For BRCA1 carriers, the cumulative breast cancer risk is 72% and the cumulative ovarian cancer risk is 44% to age 80; for BRCA2 carriers, the cumulative breast cancer risk is 69% and the cumulative ovarian cancer risk is 17% to age 80 (Kuchenbaecker et al. 2017). Figure 1 shows the location of the BRCA1 and BRCA2 genes.

![BRCA Gene 1](https://commons.wikimedia.org/w/index.php?curid=49029667)

![BRCA Gene 2](https://commons.wikimedia.org/w/index.php?curid=49029667)

**Figure 1.** BRCA1 and BRCA2 genes.*

*By Tesssa13 - Own work, CC BY-SA 4.0, https://commons.wikimedia.org/w/index.php?curid=49029667

Just as First Lady Betty Ford’s public spotlight and battle with breast cancer in the 1970s caused a rush of women to request breast exams and helped to destigmatize breast cancer (Beck et al. 2014), Angelina Jolie’s public showcase of her testing for BRCA1 in 2013, and subsequent risk reducing double mastectomy,
caused what researchers termed the “Angelina Jolie effect”, where clinics saw almost double the referrals in
the aftermath of her announcements (Evans et al. 2014, Jolie 2013). In fact, 2013 turned out to be a pivotal
year for BRCA1/2 for several reasons. In February, the Affordable Care Act clarified that BRCA testing and
counseling was covered as a preventative service for women who met agreed upon risk factors based on
family history (Chen et al. 2018). On June 13, the Supreme Court ruled that individual genes found in nature
(specifically, BRCA1/2) could not be patented by Myriad Genetics, a molecular diagnostic company
(Kesselheim et al. 2013). This ruling opened the door for other labs to enter the genetic testing market.
Overall in 2013, there was a 57% increase in BRCA testing among women, compared with average annual
increases of only 11% between 2009-2012 (Chen et al. 2018:5).

Parallel to these landmark events was the advancement of genomic technology, namely next-
generation sequencing (NGS), which enabled genetic testing to become both faster and cheaper than ever
before (Koboldt et al. 2013, Goodwin, McPherson, and McCombie 2016). NGS has had a remarkable impact
in the field of genomics, enabling a large amount of genetic information to be sequenced cheaply and quickly
compared to earlier technology such as Sanger sequencing (Reis-Filho 2009, Schuster 2007). The capability of
NGS in the context of GT/C allows for multiple genes to be tested at once, or what is also known as gene
panel testing, which can test up to 83 genes at once⁴. The onslaught of genomics has prompted changes,
including updating policy statements from the American Society of Clinical Oncology (ASCO) and updating
the definition of “genetic counseling” (Resta et al. 2006, Robson et al. 2010). However, as Roche (2012)
points out, “many figurative hands have been wrung” over how to best deal with the “imminent genomic
tsunami hovering off our shores, threatening to drown us all in downpour or sequence data” (Roche
2012:777). Indeed, it was Roche’s initial musings from 2012 that led to an entire special issue in the Journal of
Genetic Counseling to specifically address how NGS might impact GC practice. Specifically, the special issue
sought to “tap into the collective wisdom of the genetic counseling community to explore how NGS's clinical

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⁴ Invitae, which calls itself a “genetic information company”, offers the “Invitae Multi-Cancer Panel” which tests 83
genes associated with cancers in eight organ systems: breast and gynecological, gastrointestinal, endocrine, genitourinary,
skin, brain/nervous system, sarcoma, and hematologic. Their “Common Hereditary Cancers Panel” tests 47 genes.
application would impact current genetic counseling practices and ponder future implications by providing a platform on which the voices of genetic counselors could be heard” (Roche and Palmer 2014:439).

Finally, another important model of genetic testing technology is direct-to-consumer (DTC) genetic testing, which, as the name suggests, provides genetic information without a medical practitioner. Although DTC testing was not a focus of my study, there were participants who encountered and used specific DTC companies for health information, and this approach serves as an important actor in the genetic testing landscape. Genetic testing for ancestry has been around since 2000 (Smart, Bolnick, and Tutton 2017), yet it wasn’t until recently that these tests were also allowed to provide information on disease risks. In 2013, the Food and Drug Administration (FDA) imposed a moratorium on tests from genetics company 23andMe that provided genetic health information, insisting that the “company prove that its test were accurate, and that customers understood their results” (Kolata 2017). That moratorium was lifted in April 2017, allowing testing for genetic health risks such as specific BRCA1 and BRCA2 variants, Celiac disease, late-onset Alzheimer’s disease, and Parkinson’s (https://www.23andme.com/dna-health-ancestry/). While ancestry testing has gone largely unregulated, health testing has been scrutinized due to the potential implications for prevention, diagnosis, and treatment of disease (Smart, Bolnick, and Tutton 2017). With the approval of this “medical device”, other companies are entering the DTC fray, which has ethical, regulatory, and medical implications for consumers and practitioners alike (Allyse et al. 2018).

Breast cancer

Among primates, perpetually enlarged breast tissue is a curious phenomenon specific to Homo sapiens (and to a lesser extent, Pan paniscus), for which researchers do not have an answer for (Mascia-Lees 2009). While permanent breasts are primarily found in H. sapiens, they are also a sexually dimorphic feature found primarily on women. For this reason, breasts, and breast cancer, tends to be a gendered disease. For women, breast cancer is the most common cancer diagnosed in the United States and is the second leading cause of cancer death (DeSantis et al. 2017). Over a woman’s lifetime, she will have a 12.4%, or 1 in 8 chance, of being diagnosed with breast cancer, with the median age of diagnosis at 62 years (DeSantis et al. 2017). In 2018, it is
estimated that there will be 266,120 cases of invasive breast cancer diagnosed in the United States, with a resulting 40,920 deaths (Smith et al. 2018), yet only about 5% of these breast cancer cancers are due to a mutation in the BRCA1 or BRCA2 gene (Rahner and Steinke 2008). The recognition that breast cancer could occur more often in certain families was recognized by physicians in ancient Rome, and was an idea that was thoroughly documented by French surgeon and anatomist Pierre Paul Broca (1824-1880) using his own wife’s family history of breast cancer (Wagener 2009). While breast cancer mortality rates have declined since the late 1980s, black women still have a 42% higher mortality rate than white women (Smith et al. 2018) and worse 5-year relative survival rates by 10% (U.S. Cancer Statistics Working Group 2018). Overall, however, 5-year survival rates are quite good for female breast cancer – 88.6% versus 64.9% for all cancers (U.S. Cancer Statistics Working Group 2018).

In 1974, First Lady Betty Ford was diagnosed with breast cancer. Instead of hiding her diagnosis, she decided to share her journey with the general public, ushering in an era that led to destigmatization of breast cancer and screenings like mammography and breast exams (Beck et al. 2014). This destigmatization was followed by a breast cancer movement in the 1980s and 1990s that Maren Klawiter (2008) says, while narrow in scope because it was centered around one disease, is one of the “broadest of movements – bridging across institutional domains, disease regimes, fields of contention, and cultures of action” (Klawiter 2008:248). Feminist health activism in the United States coalesced around issues such as reproductive rights and disease-specific issues as a way to show how the body could become a politicized site of power struggles (Grigg and Kirkland 2015:332).

**Ovarian cancer**

Ovarian cancer is relatively rare, with only about 1.3 percent of women in the United States diagnosed over their lifetime (National Cancer Institute 2018b). However, it is responsible for more deaths than any other cancer of the female reproductive system, with an estimated 14,070 ovarian cancer deaths in 2018 (American Cancer Society 2018a, Henderson, Webber, and Sawaya 2018). This estimate is slightly higher than the 13,920 women who died of ovarian cancer in 2015 (U.S. Cancer Statistics Working Group 2018).
Part of what makes ovarian cancer so deadly is the lack of quality screening tests. Therefore, more than 60% of cases are diagnosed after ovarian cancer has already metastasized (Henderson, Webber, and Sawaya 2018) or else it is diagnosed at late stages (Torre et al. 2018). This delay in identification contributes to a relatively low 5-year relative survival rate of 46.1% (U.S. Cancer Statistics Working Group 2018). While a good screening test for ovarian cancer does not exist, the two most often used are transvaginal ultrasounds and CA-125 blood tests. Transvaginal ultrasounds can be used to identify masses in the uterus, fallopian tubes or ovaries. CA-125 identifies a protein in the blood that can be a good tumor marker for some women, but can also show up with conditions such as endometriosis (American Cancer Society 2018b). Signs and symptoms of ovarian cancer can include bloating, fatigue, pain during sex, pelvic or belly pain, trouble eating or feeling full, and urinary symptoms (American Cancer Society 2018b). Unfortunately, these symptoms can be attributed to many other causes and as a result, women may not follow up with a doctor or misdiagnosis occurs. In fact, misdiagnosis of these symptoms are seen as high as three-quarters of the time (Koldjeski et al. 2004). Depending on the type and stage of ovarian cancer, treatment options can include surgery, radiation, chemotherapy, and hormone therapy (American Cancer Society 2018b). While surgery is sometimes the first option, Goff et al. (2006) found after evaluating over 10,000 women that a “significant percentage” of women did not receive recommended surgical procedures, which can ultimately effect survival (Goff et al. 2006:383).

A Brief History of Genetic Counseling for Hereditary Cancer Syndromes

Genetic counseling (GC) is defined as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease” (Resta et al. 2006:77). Sheldon Reed, a geneticist at the University of Minnesota’s Dight Institute of Human Genetics, first coined the term GC in 1947 (Resta 1997) and the first master’s degree GC program was established at Sarah Lawrence College in 1969 (Stern 2009). At the time, GC was only accomplished through PhDs and MDs, and the master’s program received a “paternalistic form of professional gate-keeping” and pushback from medical geneticists prior to getting started (Stern 2009:3). Since then, GC has experienced rapid growth, with a 75% increase in the workforce since 2006 (Knapke et al. 2015) and a total membership of 3,174 with the National Society of Genetic Counselors (NSGC) in 2015 (National Society of Genetic Counselors 2015).
Overwhelmingly, GCs are white and female (Lega et al. 2005). There are currently 30 genetic counseling programs accredited by the American Board of Genetic Counselors (ABGC) in the United States, with another 11 programs newly accredited (who have not graduated a class yet), and 3 more under accreditation review (Accreditation Council for Genetic Counseling 2018). As a “leading voice” of genetic counselors, the NSGC reflects what is important in the world of GC. For example, in 2015 the NSGC kept track of the articles it released and found that they covered a variety of topics at difference frequencies. This included oncology (33%), prenatal testing (24%), genetic testing and genetic counseling (both 13.5%) as the most common, and Alzheimer’s disease, Angelina Jolie’s announcement, direct to consumer tests, and the Precision Medicine Initiative (all 5%) less frequently (National Society of Genetic Counselors 2015).

GC, typically vis-à-vis genetic counselors, has much to accomplish during a session: “convey medical and genetic information, translate implications, communicate in a way to optimize understanding, aid psychological adjustment, and ensure that decision making is informed” (Roche 2012:777). Numerous cancer genetic counseling practice guidelines have been published in order to provide recommendations for the medical, psychosocial, and ethical consequences of genetic counseling and potential testing (Riley et al. 2012, Berliner et al. 2013, Berliner and Fay 2007, Moyer 2014). As the leading professionals in the matter, the NSGC has published practice guidelines for cancer risk assessment for hereditary breast and ovarian cancer attributed to BRCA1 and BRCA2 mutations (Berliner et al. 2013, Berliner and Fay 2007). The NSGC has selected several components that should be addressed during the genetic counseling session, including:

1. **Intake and history**: personal and family history is documented to understand possibility of hereditary cancer risk.

2. **Psychosocial assessment**: a thorough evaluation in order to understand the full impact of genetic information on the individual and their family (e.g. specific cancer risks, concerns of discrimination, risks for children and/or other family members).

3. **Cancer risk assessment**: risk assessment models, based on personal and family characteristics, provide risk estimates (Berliner and Fay 2007).
Communication is an important aspect of GC. In a recent systematic review on communication during genetic consultations, Paul and colleagues (2015) determined that GC communication studies fall under three main objectives: 1) searching for structural patterns during consultations, 2) determining communication and GC concepts, and 3) linking processes with input- and outcome- measures (Paul et al. 2015). Each of these three main objectives also had identified themes. In the studies to identify structural patterns, it was determined that clinicians dominate the discussion and that communication tends to be mostly educational and scientific rather than psychosocial (Paul et al. 2015:19). Concepts and communication themes that arose included the communication of risk, negotiations of power and knowledge, and the use of “indirect speech” to remain non-directive (Paul et al. 2015:20; 28-29). Finally, the review found that studies that attempted to link process with input- and outcome- measures were not typically successful (Paul et al. 2015:20).

Importantly, while the systematic review by Paul et al. (2015) found that most counseling sessions were primarily educational/biomedical in nature, another review on risk communication interventions determined that the psychosocial aspects (supportive/emotional elements) of counseling provided the most benefit to counselees (Edwards et al. 2008). While counselees and counselors obviously have different agendas and goals for their session, the focus of communication (education vs. emotional) may need to shift in order to best address the counselee and their needs.

A particularly in-depth view of GC was undertaken by anthropologist Rayna Rapp who studied prenatal GC in the early 1980s. Her research, in part, was to study the discourse between genetic counselors, a relatively “new health profession” at the time, and the multilingual, multicultural clients they came into contact with in New York City. Specifically, she was interested in how amniocentesis, as a new reproductive technology, was not only being communicated in regards to risks and benefits, but how power, social knowledge, and popular meanings about disability played out in the hospital (Rapp 1988). One of the most important findings from Rapp’s research is that despite the emphasis on being nondirective and for a patient to make up their own mind regarding pursuing testing, it was instead found that “counselors stand in a contradictory position with respect to their clients’ decisions…for they are always making choices about what sort and how much information a pregnant woman needs and can use, as well as the form in which she can
best absorb it” (Rapp 1988:154). This situation is mirrored in the genetics clinic, for the genetic counselor cannot/do not go over every single type of test that is available to the counselee, nor do they provide information on the many different laboratories that conduct the testing (personal observation, 2014). Rapp also clearly and effectively presents power differentials that exist between counselor and patient, as well as calls into question the supposed universality of the language of science, which is portrayed as objective and fact-based (Rapp 1988). This research, clearly focused on communication and discourse, was enveloped within her larger research focus that contributed to assessing new reproductive technologies, disability rights, reproductive rights, and the role of scientific literacy in America (Rapp 1999).

Summary of literature review and theory

This chapter seeks to situate the reader among a variety of disciplines in order to understand the current research. As guiding frameworks, feminist theory and CMA are the interpretive lenses that this work is conducted through, impacting what data is collected and how it is interpreted. Feminist theory requires a gendered analysis to interrogate the ways in which power, gender, and privilege are constructed within a biomedical setting. While feminist theory can sometimes be used to understand individuals on the ground, CMA is then used to interpret individual actions under a larger framework. In this work, CMA reminds us that macrolevel forces are also at work which we cannot forget in the face of individual stories.

Part of the macrolevel approach is understanding the healthcare system in which this work is embedded, especially that of oncology care in the United States. Cancer care has grown by leaps (people) and bounds (money) since the National Cancer Act was signed in 1971. Recent renewed momentum in the form of the Cancer Moonshot 2020 indicate that while much progress has been made, more work has yet to be done. Racial cancer disparities are growing among some cancers, the price of cancer drugs is skyrocketing, and some Healthy People 2020 cancer objectives are slipping through our fingers.

Within cancer care there exists a relatively small portion of the pie which deals with hereditary cancers. While only 5-10% of cancers worldwide are estimated to be hereditary, more than 200 hereditary cancer syndromes have been identified since the early 1990s. Critical to this research are the BRCA1 and
BRCA2 genes and hereditary breast and ovarian cancer (HBOC) syndrome. Mutations for these genes increase cumulative risks for breast and ovarian cancer primarily. In the United States, breast cancer is the most common cancer diagnosed and is the second leading cause of cancer death. Ovarian cancer is rarer but is responsible for more deaths than any other cancer of the female reproductive system.

Finally, an understanding of genetic counseling (GC) is critical to this research. As a master’s degree profession, GC is relatively young having started in the late 1960s. However, it has experienced massive growth in both practitioners and programs. Guidelines for genetic counseling indicate several components that anyone ordered genetic testing should adhere to: personal and family history intake; psychosocial assessment to discuss impact of genetic information on the patient and their family members; as well as a cancer risk assessment, which is accomplished through various risk assessment models.
CHAPTER THREE:
METHODOLOGY AND PARTICIPANT CHARACTERISTICS

Introduction

This research explores the experiences of genetic testing and counseling by women determined to be at high genetic risk for hereditary breast and ovarian cancer. This study was first informed by work experience as a research assistant at a large cancer center on a study focused on hereditary cancer, which provided exposure to genetic counselors, genetic counseling appointments, and patients who had undergone genetic testing. For the dissertation research, semi-structured interviews were conducted between June 2017 and May 2018, complimented by attendance at various conferences and local groups specific to hereditary cancer.

Study participants and recruitment

Recruitment of study participants

Participants were recruited through local Tampa Bay breast and ovarian cancer organizations, which were established through previous research connections of Dr. Dinah Martinez-Tyson and by internet search. IRB approved (Pro# 00030472) recruitment material was emailed to the contact person listed for each organization, which could then be distributed to the organization members if the contact person so agreed. Before distribution of the recruitment materials, I offered to answer any questions or concerns that the contact person had. In addition to recruitment through local organizations, participants were encouraged to send recruitment information to other women they thought might be interested. Snowball sampling as a technique has been used to help recruit similar respondents in specific target groups with multiple eligibility requirements, such as the present study (Sadler et al. 2010). Altogether, this recruitment strategy and sampling technique helped to include both family members of some participants and other women in social media and support groups in states outside of Florida. Various snowball sampling frameworks included affinity social
groups (e.g., Facebook groups) and affinity organizations/resources (e.g., Faces of Courage Foundation, Celma Mastry Ovarian Cancer Foundation, National Ovarian Cancer Coalition) (Sadler et al. 2010).

Recruitment information was passed along through email listservs, word of mouth, and posted in private Facebook groups. An initial goal of 25 individuals was set, but after snowball sampling increased dramatically through ovarian cancer groups, the final participant total was 33. The increased sample size was approved by dissertation Chairs and IRB.

Forty-seven women made contact to participate in the study. Two (2, 4.3%) did not meet eligibility criteria; nine (9, 19.1%) did not interview due to scheduling/communication issues; and two (2, 4.3%) contacted me after I had closed the study for interviews.

**Inclusion criteria**

Inclusion criteria were: women who were 18 years or older, English speaking, and be determined to be at high genetic risk for hereditary breast/ovarian cancer. To determine genetic risk, participants were asked the following questions based on screening guidelines (adapted from the National Comprehensive Cancer Network Guidelines Version 1.2017, www.nccn.org). If participants answered yes to one or more of the following, they were considered high genetic risk and eligible to participate:

- Have you
  - Ever been diagnosed with ovarian cancer?
  - Ever been diagnosed with breast cancer at or under the age of 50?
  - Ever been diagnosed with two separate cancers?
  - Had one or more blood relatives with breast, ovarian or pancreatic cancer?
  - Any Ashkenazi Jewish heritage in your family?
  - Ever been found to carry a mutation in a cancer susceptibility gene?

- If you’ve **had** breast cancer, has
  - ≥1 close\(^a\) family blood relative had breast cancer ≤50 years old?
  - ≥1 close family blood relative had ovarian cancer (includes fallopian and peritoneal cancers)?
• If you **have not** had cancer, is any of the following true for your family (blood relatives):
  
  - There is a known mutation in a cancer susceptibility gene within the family
  - A close relative has had ≥2 primary breast cancers
  - A close relative has had ovarian cancer
  - A close male relative has had breast cancer

• Is there a personal and/or family history of three or more of the following diagnoses:
  
  - Breast, pancreatic cancer, prostate cancer, melanoma, sarcoma, adrenocortical carcinoma, brain tumors, leukemia, diffuse gastric cancer, colon cancer, endometrial cancer, thyroid cancer, kidney cancer, dermatologic manifestations, and/or macrocephaly, hamartomatous polyps of gastrointestinal tract

  *close blood relatives include first-, second-, and third-degree relatives*

**Study design**

Women who received the recruitment material were directed to contact me by email or phone. After this initial contact, inclusion criteria were established. If inclusion criteria were met, then a telephone or in-person interview was coordinated and scheduled. Most women opted for the phone interview, and four were done in person. Telephone interviews were offered as an option for multiple reasons. Because participants were not being compensated for their time, I hoped to make the interview as convenient as possible. Second, while I focused on the Tampa Bay area, snowball sampling led to some participants living in other states (at least 7, 21.2%). Before the interview, the study was explained, consent was reviewed, and participation was invited. I asked if there were any questions, addressed them, and ultimately obtained verbal informed consent. All interviews were audio-recorded for transcription.

To begin the interview, I first went over personal and family history of cancer in order to draw a three-generation pedigree (family tree, explained in more detail below). This was another check that eligibility
criteria were indeed met. Pedigrees also provided context for the participant’s knowledge and experience of cancer both personally and within the family. Once the pedigree was complete, the participant was offered a copy for their records. Next, participants were asked to describe their cancer experience (if they had been diagnosed). This provided information about cancer symptoms, diagnosis, and treatment, in addition to providing context for the GT/C experience. A semi-structured interview guide (see Appendix A) was used to elicit motivations for testing, determine who ordered testing, and other specifics of the GT/C experience. At the end of the interview, participants were asked if they would like to include anything else, which provided an open forum for feedback and/or a chance to let me know what they felt was important about their story.

Data saturation and nonprobabilistic sample

Research conducted in anthropology, especially cultural or health research, may not be concerned with statistical generalizability and will often rely on nonprobabilistic samples (Guest, Bunce, and Johnson 2006). Additionally, this research required participants to fit specific inclusion criteria and thus purposive sampling was used. As for guidelines for sample size, variability exists in the literature. Guest, Bunce, and Johnson (2006) note that they could only find seven sources that provided detailed guidelines for sample sizes, which ranged from 15-36 interviews as acceptable numbers. Morse (2000) suggests approximately 35 participants for ethnographies, grounded theory studies, and ethnoscience studies. In fact, in testing how many interviews are needed for qualitative research themes, Guest, Bunce, and Johnson (2006) found that six interviews provided high-level, overarching themes which they described as being enough to develop meaningful themes and useful interpretations. Regardless of the sample size, the researcher must also feel confident that saturation has occurred. I follow the definition provided by Glaser and Strauss (2017) of saturation, “that no additional data are being found whereby the sociologist can develop properties of the category” (p. 61). Toward the end of the interviewing stage, I had a good sense of what the answers to my questions would be, which indicated data saturation. Once I felt confident that saturation had been met, I felt comfortable closing the study to further interviews.
Pedigrees

In the history of anthropology, family and kinship have been a major area of study (Finkler et al. 2001). Pedigrees are used in biological anthropology to demonstrate patterns for epidemiological study or to display biological kinship patterns. It is a way to visually document direct ancestors, bloodlines, or lineages. Historically, it was an attempt to determine the “specific rights and duties, membership and status” of people (Epstein 1979:101). They have also been used in the study of political and economic anthropology. While critiques of kinship studies have rightly questioned the role of biology and culture in the actual construction of a “family” (Finkler et al. 2001, Peletz 1995), in researching hereditary diseases my goal was to gather information on biologically related blood relatives rather than the myriad types of “family” that can be constructed, regardless of biology. Further, pedigrees are used to visually capture both biological and cultural phenomena: biological variables (e.g., cancer diagnosis, age, genetic testing) along with the cultural milieu surrounding a cancer diagnosis through informal discussion of family history. These pedigrees are a snapshot in time, and instead of being regarded as static models, should be viewed as resources that can change over time (Finkler et al. 2001).

Pedigrees were obtained from each participant at the beginning of the semistructured interviews, which allowed participants to describe their personal and family history of cancer. This centered their cancer story within a larger familial framework and provided context for their GT/C experience and knowledge. As an interviewer, gathering pedigrees provided insights to various struggles associated with cancer – if the participant was the only person in the family with a cancer diagnosis, how did that impact them? If they had watched a loved one die of cancer, how did that effect their outlook for their own diagnosis? Inevitably, these stories came up during the interview. Given that I had a visual representation of the family and had spent 10-20 minutes discussing family history, I was much better equipped to understand the ripple effects of cancer in the context of each participant’s story.

These pedigrees provided data which was ultimately used in multiple ways. First, because inclusion criteria were multi-layered, I was able to double-check that the participant indeed met criteria. Second, it
provided context for the participant’s own personal and family history of cancer and/or genetic testing/counseling. This helped me understand the participant’s experiential orientation to the topic and often served as a way to initiate discussion. Did they watch a parent or sibling suffer through a cancer diagnosis? Were they the first one in their family to have a cancer diagnosis? If they were BRCA positive, what family members were they worried about telling (or not)? In providing this information, I hope it will help situate the reader visually and, potentially, empathetically to the history of each participant. This data is another way in which to possibly put ourselves into the shoes of another, an attempt to situate ourselves emically, or as the women interviewed defined themselves in the context of their biological family members.

Having drawn pedigrees for a hereditary breast and ovarian cancer study while working at a major cancer research center in the United States, I followed standardized pedigree nomenclature put forth by the National Society of Genetic Counselors (Bennett et al. 2008). While I attempted to gather the following basic information to draw a three-generation pedigree, some participants could not give all the information for each individual in their family for numerous reasons, including: estrangement, lack of information/knowledge, or reluctance to provide information. The information that was attempted to describe each individual is as follows:

- Identification of proband (ego/participant), indicated by a black arrow
- Month/year that pedigree was drawn
- Ancestry for both sides of family (maternal/paternal)
- Age or birth year
- Age of death or year of death
- Cancer diagnosis
- Age of cancer diagnosis
- Absence/presence of genetic testing

In the following pedigrees (see Appendix B), participants are denoted with a small black arrow. A legend is found in the upper left-hand corner of each pedigree to illustrate the types of cancers found in the family. In the case of every individual, I attempted to elicit their current age, their age at cancer diagnosis, their age at death, and if they had genetic testing. Numbers inside symbols represent multiple individuals, while a black

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5 In anthropology, this person is called the ego; in genetic counseling, the proband.
diagonal line indicates that the individual is deceased. Maternal ancestry is found on the right side of the pedigree, while paternal ancestry is found on the left. Due to a variety of reasons, some individuals are lacking information, and these pedigrees represent the best amount of information my participants could provide. Figure 2 shows common pedigree symbols and abbreviations that were used.

![Pedigree Symbols and Abbreviations](image)

**Figure 2.** Pedigree legend.

**Transcription of interviews**

A review by Halcomb and Davidson (2006) noted that while there has been a significant increase in interest of qualitative research, the transcription process has hardly ever been given a detailed description. To address this important concern, I will describe my transcription process.

I transcribed all interviews personally, which allowed for reflexivity and further field notes to be developed (Alcock and Iphofen 2007). Nonverbal cues, such as silences or body language, were not incorporated since the majority (n=29, 87.9%) of the interviews were conducted over the phone at the convenience of the participant. The interviews were not transcribed verbatim, in that they were not an exact word-for-word replication of the audio-recorded words (Poland 1995). Rather, a targeted transcription
approach was taken, which is in line with suggestions from Halcomb and Davidson (2006). For instance, I did not transcribe parts of the interview that did not directly pertain to the research objectives. Halcomb and Davidson question if interviews should be transcribed verbatim for every research project. For them, “the process of transcription should be more about interpretation and generation of meanings from the data rather than being a simple clerical task” (Halcomb and Davidson 2006:40). Emerson, Fretz, and Shaw (2011) also support the idea that selected paragraphs or stories that are relevant to the research question are all that are needed for transcriptions (McLellan, MacQueen, and Neidig 2003).

Transcription for this research project was focused on addressing the research objectives: the GT/C experience and women’s conceptualization of risk. For every interview, I transcribed women’s stories of how they were first diagnosed with cancer and their treatment. This was an open-ended question at the beginning of the interview intended to get women talking about their experience, which also provided context for the GT/C encounter. To find out more about the GT/C experience, I asked more detailed questions regarding the encounter and how the information was used by participants. All pedigree and transcription data were uploaded into Dedoose for coding and to assist analysis (Dedoose 2016).

Coding and Analysis

LeCompte and Schensul (2013) define codes as “names or symbols used to stand for a group of similar items, ideas, or phenomena that the researcher has noticed in his or her data set” (p. 121). Put another way, codes in qualitative research are used to describe an idea, theme, or anything that has a certain type of quality (LeCompte and Schensul 2013). Once transcripts were coded, I was able to easily search for patterns, relationships, similarities and differences (Lewins and Silver 2007). Analysis of the data included grounded theory (Glaser and Strauss 2017), vignettes, and thematic analysis to help organize and present dissertation results.

An example of codes and their usage can be seen in Table 2. Due to the semistructured interview format, a repeated structure exists among the transcript data and allowed for question-based coding to arise (Lewins and Silver 2007). Question-based codes included items such as: how to use test results, knowledge of
genetic testing, and reason for genetic testing. In addition, through the process of immersing myself in fieldnotes, listening to the interviews numerous times, and transcribing, I was able to inductively identify reoccurring elements in the data and assigned codes accordingly. These codes including items such as emotion, family, insurance, risk, and time.

Table 2. Codes and their operationalization

<table>
<thead>
<tr>
<th>CODE</th>
<th>DESCRIPTION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anything else</td>
<td>Open-ended question at the end of the interview to allow women to comment or expand on anything they thought was important</td>
</tr>
<tr>
<td>Cancer cause</td>
<td>Applied whenever participants offer a reason or cause of their cancer</td>
</tr>
<tr>
<td>Chemo brain</td>
<td>Applied when participants note memory issues related to chemo</td>
</tr>
<tr>
<td>Family</td>
<td>Applied when participants note importance of family</td>
</tr>
<tr>
<td>Genetic counseling</td>
<td>Applied only if participant reports being seen by a genetic counselor or geneticist, about their counseling experience</td>
</tr>
<tr>
<td>Genetic testing</td>
<td>Applied when genetic testing is discussed</td>
</tr>
<tr>
<td>Good quotes</td>
<td>Exemplar quotes</td>
</tr>
<tr>
<td>How to use test results</td>
<td>Applied when participant spoke about the ways in which genetic test results either could be used or were used in their situation</td>
</tr>
<tr>
<td>Insurance</td>
<td>Applied when insurance was discussed</td>
</tr>
<tr>
<td>Knowledge of genetic testing</td>
<td>How or what did the participant know about genetic testing/counseling?</td>
</tr>
<tr>
<td>Reason for genetic testing</td>
<td>Applied if something prompted testing (e.g., family member was positive) or wanted testing to use results for a specific reason (e.g., treatment, family).</td>
</tr>
<tr>
<td>Risk</td>
<td>Applied when risk is discussed (e.g. reducing/controlling risk, knowing risk factors)</td>
</tr>
<tr>
<td>Test results</td>
<td>Applied when discussion of genetic test results were discussed</td>
</tr>
<tr>
<td>Time</td>
<td>Applied anytime specific dates, such as months, days, or years, was discussed</td>
</tr>
</tbody>
</table>

With only one coder, there was no way to check intercoder reliability, stability, and reproducibility (Campbell et al. 2013), which is a limitation that many early-stage and un(der)funded researchers deal with. To combat this limitation, several steps were taken during coding and into analysis, while also recognizing that coding is analysis (Miles and Huberman 1994). First, a coding scheme was developed prior to the review and actual coding of transcriptions. These codes reflected research objectives and some of the questions in the semistructured interview (e.g., how to use test results, genetic counseling, genetic testing, risk). Using this initial coding scheme during the first cycle of coding, I was able to code both for previously anticipated
themes and also add extra codes as they became relevant (i.e., inductively generated codes). Some of the codes that were added during the initial coding process included “time” (when participants noted specific dates or times) and “chemo brain” (when participants spoke about being forgetful or having memory problems due to treatment). After this first cycle of coding, which included my preliminary codes in addition to new, inductively generated codes, I then started a second cycle of coding with my updated coding scheme, reflecting all the codes I had gathered after the first cycle of coding. The second cycle of coding ensured that new codes were applied to all transcription data, that preliminary codes were not missed during the first cycle of coding, and to ensure that no new codes emerged from the data. I revisited the transcripts more than twice in total at various points of analysis to manage, filter, and highlight salient data (Saldaña 2015).

After the second cycle of coding, I then explored the coded data within Dedoose using the “Analyze” tab, which quantifies and visualizes the codes into various charts. For example, I was able to view a matrix which showed “Code Co-Occurrence” (Figure 3) which allowed me to see what codes occurred together and at what frequency. This process helped me to gain a better sense of important themes present in the data that I may have missed if I were looking at code excerpts in isolation from one another.
Figure 3. Example of Code Co-Occurrence in Dedoose.

After exploring the data using the Analyze tab in Dedoose, I then exported specific code excerpts to view in a single column in an Excel file. Depending on the research objective I was attempting to address, I would split the codes into various groups in different Excel tabs: breast cancer versus ovarian cancer versus no cancer diagnosis, or NGP versus GP experiences. This splitting of the sample is an important component of grounded theory, which seeks to understand how people experience similar phenomenon under different circumstances (Starks and Brown Trinidad 2007). While these codes initially may have been more “lumped” and broad because they were focused on the answer to a specific question from the interview, breaking the code excerpts into various groups within my study allowed subcodes to emerge and “split” based on participant’s responses (Saldaña 2015). For example, once I exported the code “reason for genetic testing” and split the code excerpts into the different cancer diagnosis groups, I was then able to see more specific themes emerge as reasons for testing (discussed in Thematic Analysis in Chapter 4). This resulted in frequency counts to illustrate a basic descriptive statistical summary as well as importance of themes.
(LeCompte and Schensul 1999). For the thematic analysis, I split the “reason for genetic testing” code by cancer diagnosis for a couple reasons. One, splitting the codes into smaller groups made it easier to discern themes. Two, women’s reported experience of cancer diagnosis and treatment was vastly different between cancers, which I believed would translate into different reasons for wanting to have genetic testing done. This same technique was enacted for women’s reactions to their test results, except the groups were split into specific result they received and what kind of provider ordered testing.

Given the depth of data, thematic analysis was not the only type of analysis I used in order to understand and present this data. To understand women’s overall experiences with the providers ordering testing, and how providers impacted the experience and use of genetic testing, I felt that a vignette approach would be the most useful way to illustrate two very different experiences. A vignette “illustrates an interpretive theme within a research paper. Vignettes sketch images that through their detail illuminate ideas that seem inherently related to ‘being there’” (Graue and Walsh 1998:220). The interpretive theme for the vignette in Chapter 4 is to juxtapose the experience of seeing a GP versus a NGP, which is accomplished through one participant’s story after experiencing the genetic testing process twice – first, with a NGP and second with a GP.

Positionality

This research was informed initially by my 3½ year stretch as a research assistant at an NCI-Designated Comprehensive Cancer Center in the US Southeast. While there, I worked on a study focusing on hereditary breast and ovarian cancer (HBOC). I interacted with women who had a personal or strong family history of cancer, a positive BRCA1/2 test results, and who agreed to be followed longitudinally using survey data to describe their surveillance, surgeries, and a multitude of other factors. In addition to those study participants, I also experienced the clinical and medical side of HBOC, as I sat in on numerous monthly genetics case conferences where genetic counselors, geneticists, and other genetics professionals, both domestically and internationally, would present cases for comment and feedback. I also shadowed genetic counselors during their appointments and conducted a focus group with local genetic counselors. This initial
foray into the world of genetic testing and counseling was much more focused on the clinical side, which helped me to understand the demands of genetic counselors, the landscape in which testing is ordered, and the difficulties that can arise which may not always be apparent to the patient. This first round of fieldwork provided a clinical and medical understanding of HBOC and GT/C.

Incidentally, genetic testing crossed over from a research subject to a personal one. When I was practicing drawing pedigrees, which necessitated asking about family history, I discovered that both my maternal and paternal grandmothers had been diagnosed with ovarian cancer. I spoke to a genetic counselor friend of mine about this newly discovered family history, along with some other familial factors, and she indicated that I would be a candidate for genetic testing. However, I decided not to pursue it because of all the reasons we decide not to see a doctor: I didn’t want to deal with the costs, the appointment would be time consuming, and I also didn’t really know if I needed that information right now. About six months later, I learned that I qualified for free genetic testing and counseling through a research study with MD Anderson. With the inconvenience of time and cost removed, I decided to move ahead with testing. I spoke with a genetic counselor over the phone, received the testing kit in the mail, spit my DNA into a tub, and mailed it back. The friendly email reminders I received after every interaction with the company reminded me that results were expected after four weeks and gave me a specific day (November 27th) which I would receive my results. As much as I attempted to be nonchalant about the genetic information coming my way, I noticed that I was very on top of checking my email for my results on November 27th. When I first mailed my testing kit back, I tried to assure myself that most people come back negative, and that I would, too. When the date passed for when my results were supposed to be returned to me, the anxiety and worry set it, along with many internal conversations. I found myself much more anxious than I ever anticipated I would be. Part of me was trying to maintain the (fake?) façade of impartial, objective researcher. It's simply information, after all, and information in itself is objective and impartial. But this is information can have life altering impacts on my life. Different thoughts and excuses ran through my brain: If it is taking longer, does that mean I am positive? Is this delay because “they” have to have a discussion of how to break the information to me? (When I know that “they” do this all the time and I am not special). No, it must be taking longer because of
the holidays - Thanksgiving pushed back the timeline, no need to worry. Whatever the many excuses or reasons I told myself, it all came down to anxiousness. I was anxious even though I know that my risk for a positive gene mutation is low, even though I understand this topic more than most people who undergo the process, even though I am a logical, thoughtful researcher who knows how to scientifically approach problems. But what this narrative discounts, and what I am ashamed to realize given that I am an anthropologist, is the supreme role that emotions play in our research. Not only the emotions participants may express, but the emotions that we also inevitably bring to the table. Why should I pretend that these are sides to me that would be completely separate? The “scientist” and the “human” - these are not personas that cancel the other out. As a scientist who studies humans, understanding the wide range of emotions that we exhibit and feel should be of the utmost importance to much of the work that we do. This is also in line with much of the reflexive nature of our work - just as we question our positionality, our impact on people and communities, should we also grapple with how we feel during this very (important, stressful, fruitful, engaging) fieldwork?

By this point in reading, you may also be anxious. Ultimately, I came back negative. However, the experience provided first-hand insight into the anxiety and uncertainty that some women may feel while undergoing the testing process. About a year after I had gone through the testing process, a cousin of my father let us know that two of her daughters had tested positive for a BRCA mutation. One of my paternal second cousins had been diagnosed with breast cancer at 48; the other is undergoing a totally hysterectomy at 43 to “eliminate this threat”. While anxiety has been lessened for me and my cancer risk, I am actively working with my close family members to decide our genetic testing path going forward.

**Ethical considerations**

The research proposal for this study was approved by the dissertation committee at the University of South Florida (USF) and also received approval through USF’s Institutional Review Board (IRB). Per IRB requirements, study participants were not contact directly by me. Rather, they were required to make first contact with me via phone or email after recruitment information had been distributed through a contact
person of a local organization. IRB approved verbal informed consent was read to the participants to explain the study, explain potential risks and benefits, and what would be expected of the participant for the interview. At the end of the consent, I would ask if the individual had any questions or concerns that I could address, which allowed time for individuals to decide if they wanted to participate or not. If they decided to participate, I received verbal informed consent. These steps ensured voluntary participation, competence to participate, and informed consent in line with the ethical principle of respect for persons (Whiteford and Trotter 2008). No personal or identifiable information was collected during the interviews, and family trees have been de-identified. Each participant was assigned an identification number and pseudonym. I am the only researcher who had access to the collected data. This ensured confidentiality to protect the individual and their family (Whiteford and Trotter II 2008).

Fluehr-Lobban (2013) states that “thinking and planning ethically conscious work should be a part of every professional act and activity” (pg. 7). During my ethics coursework, we were taught to think preemptively about situations which might arise during fieldwork so that we might be better prepared to react and deal with difficult situations. Given the nature of this research, I knew that I would be dealing with potentially emotional stories and may be asked questions I might not have the answers to. Thinking about these situations, I put together resources which I thought would be helpful to direct participants to if they voiced concerns. I did direct one participant to Facing Our Risk of Cancer Empowered (FORCE), which has peer support services. Others I directed to the National Society of Genetic Counselors (NSGC) website to find a certified genetic counselor near them, because they had questions that would be better answered by a genetics professional. And at the end of the interview, I always encouraged participants to reach out to me if they had any questions or concerns. None of the participants contacted me after the interview with concerns, and no participants voiced negative repercussions due to the research questions and procedure.

**Participant demographics**

The average age for participants was 56.9 years old at the time of interview, with most reporting European ancestry (Table 3). Race in the typical US Census categorization was not gathered; rather, I asked
participants to describe their major maternal and paternal ancestry. This reflects standard genetic counseling pedigree guidelines and calls into question the use of race as any sort of medically relevant category (Root 2003). Out of 33 participants, 31 (93.9%) underwent genetic testing (Table 4). All participants had completed at least some college or an Associate degree, with most completing a 4-year degree (14, 45.2%) or graduate/professional school (9, 29%), making this a highly educated study sample. Most reported being married at the time of genetic testing (20, 64.5%) and almost all had insurance at the time of testing (31, 96.9%). Of the women diagnosed with cancer in this study (27, 81.8%), 11 were diagnosed with breast cancer (40.7%, M=43.3 years old) and 16 were diagnosed with ovarian cancer (59.3%, M=51.7 years old, Table 5).
Table 3. Individual data of study participants including ancestry, cancer diagnosis, year of genetic testing, and provider type

<table>
<thead>
<tr>
<th>Pseudonym</th>
<th>Paternal Ancestry</th>
<th>Maternal Ancestry</th>
<th>Age at interview</th>
<th>Cancer Diagnosis (age)</th>
<th>Genetic testing (result)</th>
<th>GT year</th>
<th>GP or NGP</th>
</tr>
</thead>
<tbody>
<tr>
<td>April</td>
<td>Scottish, Irish, English, German</td>
<td>Scottish, Norwegian</td>
<td>39</td>
<td>Breast (31)</td>
<td>Negative</td>
<td>2010</td>
<td>GC</td>
</tr>
<tr>
<td>Katherine</td>
<td>Dutch, Italian</td>
<td>Polish</td>
<td>49</td>
<td>Breast (35)</td>
<td>Negative</td>
<td>2003</td>
<td>GC</td>
</tr>
<tr>
<td>Lisa</td>
<td>Ashkenazi Jewish, Russian</td>
<td>German</td>
<td>66</td>
<td>Breast (55)</td>
<td>BRCA2+</td>
<td>2007</td>
<td>GC</td>
</tr>
<tr>
<td>Helen</td>
<td>Sicilian, Italian</td>
<td>Hungarian</td>
<td>43</td>
<td>Breast (40)</td>
<td>Negative</td>
<td>2015</td>
<td>GC</td>
</tr>
<tr>
<td>Joann</td>
<td>Unknown</td>
<td>Unknown</td>
<td>47</td>
<td>Ovarian/Fallopian (44)</td>
<td>BRCA1+</td>
<td>2014</td>
<td>NGP</td>
</tr>
<tr>
<td>Marjorie</td>
<td>Nigerian, Cameroon</td>
<td>Nigerian, Cameroon</td>
<td>56</td>
<td>Breast (56)</td>
<td>Negative</td>
<td>2016</td>
<td>NGP</td>
</tr>
<tr>
<td>Lilian</td>
<td>Scottish</td>
<td>Italian</td>
<td>48</td>
<td>Breast (42)</td>
<td>Negative</td>
<td>2011</td>
<td>NGP</td>
</tr>
<tr>
<td>Gretta</td>
<td>Irish</td>
<td>French Canadian</td>
<td>42</td>
<td>Breast (38)</td>
<td>Negative; PALB2 VUS</td>
<td>2014; 2017</td>
<td>NGP; GC</td>
</tr>
<tr>
<td>Becky</td>
<td>Unknown</td>
<td>Irish, English, Sicilian, Italian</td>
<td>30</td>
<td>-</td>
<td>BRCA1+</td>
<td>2016</td>
<td>GC</td>
</tr>
<tr>
<td>Julie</td>
<td>Ashkenazi Jewish</td>
<td>Ashkenazi Jewish</td>
<td>71</td>
<td>-</td>
<td>BRCA1+</td>
<td>2002</td>
<td>NGP</td>
</tr>
<tr>
<td>Sissy</td>
<td>Scottish</td>
<td>English</td>
<td>62</td>
<td>Breast (49)</td>
<td>Negative; Lynch VUS</td>
<td>2007; unk</td>
<td>NGP; GC</td>
</tr>
<tr>
<td>Melanie</td>
<td>English, Irish</td>
<td>English, Irish</td>
<td>60</td>
<td>Breast (42); Ovarian (56)</td>
<td>Negative</td>
<td>2014</td>
<td>GC</td>
</tr>
<tr>
<td>Martha</td>
<td>Scotch-Irish</td>
<td>German</td>
<td>73</td>
<td>Ovarian (63)</td>
<td>Negative</td>
<td>2014</td>
<td>GC</td>
</tr>
<tr>
<td>Courtney</td>
<td>Irish</td>
<td>Scottish</td>
<td>37</td>
<td>-</td>
<td>Unknown VUS</td>
<td>2016</td>
<td>GC</td>
</tr>
<tr>
<td>Megan</td>
<td>German</td>
<td>German</td>
<td>59</td>
<td>Ovarian (39)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Angie</td>
<td>Spain, Portugal, Sephardic Jew</td>
<td>Ashkenazi Jewish, Polish</td>
<td>70</td>
<td>Ovarian (66)</td>
<td>Negative</td>
<td>2014</td>
<td>GC</td>
</tr>
<tr>
<td>Roxanne</td>
<td>Polish, Greek</td>
<td>Czechoslovakia</td>
<td>61</td>
<td>Ovarian (52)</td>
<td>BRCA1+</td>
<td>2010</td>
<td>GC</td>
</tr>
<tr>
<td>Stacey</td>
<td>Swedish, French, Canadian, Scotch-Irish</td>
<td>English, Welsh, German</td>
<td>77</td>
<td>Ovarian (67)</td>
<td>Negative</td>
<td>2008</td>
<td>NGP</td>
</tr>
<tr>
<td>Lynne</td>
<td>English, Native American</td>
<td>English, Scotch-Irish</td>
<td>60</td>
<td>Ovarian (54)</td>
<td>BRCA2+</td>
<td>2011</td>
<td>NGP</td>
</tr>
<tr>
<td>Joyce</td>
<td>Scotch-Irish</td>
<td>English, Scotch-Irish</td>
<td>60</td>
<td>Ovarian (57)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Leigh</td>
<td>Irish</td>
<td>German</td>
<td>27</td>
<td>-</td>
<td>BRCA VUS</td>
<td>2007</td>
<td>GC</td>
</tr>
<tr>
<td>Clara</td>
<td>Ashkenazi Jewish, Italian</td>
<td>Ashkenazi Jewish</td>
<td>57</td>
<td>Ovarian (54)</td>
<td>Negative</td>
<td>2015</td>
<td>GC</td>
</tr>
<tr>
<td>Gwen</td>
<td>Ashkenazi Jewish</td>
<td>Ashkenazi Jewish</td>
<td>53</td>
<td>Breast (45)</td>
<td>BRCA1+</td>
<td>2002</td>
<td>GC</td>
</tr>
<tr>
<td>Kristina</td>
<td>Swedish, German</td>
<td>Swedish</td>
<td>60</td>
<td>Ovarian (21); Breast (55)</td>
<td>Negative</td>
<td>2013</td>
<td>NGP</td>
</tr>
<tr>
<td>Jill</td>
<td>Spanish, Dominican</td>
<td>German, Welsh</td>
<td>64</td>
<td>Ovarian (51)</td>
<td>Negative</td>
<td>2015</td>
<td>GC</td>
</tr>
<tr>
<td>Diane</td>
<td>Lebanese</td>
<td>Lebanese</td>
<td>71</td>
<td>-</td>
<td>Negative</td>
<td>2014</td>
<td>NGP</td>
</tr>
<tr>
<td>Marie</td>
<td>Dutch</td>
<td>Trinidadian</td>
<td>57</td>
<td>Ovarian (51)</td>
<td>Negative</td>
<td>2012</td>
<td>GC</td>
</tr>
<tr>
<td>Debra</td>
<td>Italian</td>
<td>English</td>
<td>62</td>
<td>Ovarian (34)</td>
<td>Negative</td>
<td>2000</td>
<td>GC</td>
</tr>
<tr>
<td>Gail</td>
<td>Irish, German</td>
<td>Irish, German</td>
<td>71</td>
<td>Breast (42); Ovarian (67)</td>
<td>BRCA+</td>
<td>Unk</td>
<td>NGP</td>
</tr>
<tr>
<td>Kristy</td>
<td>Ashkenazi Jewish</td>
<td>Ashkenazi Jewish</td>
<td>71</td>
<td>Ovarian (69)</td>
<td>Negative</td>
<td>2017</td>
<td>NGP</td>
</tr>
<tr>
<td>Yvonne</td>
<td>Irish</td>
<td>Irish</td>
<td>49</td>
<td>Ovarian (46)</td>
<td>BRCA2+</td>
<td>2016</td>
<td>GC</td>
</tr>
<tr>
<td>Bridget</td>
<td>Spanish, Dominican</td>
<td>German, Welsh</td>
<td>66</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Bonnie</td>
<td>German</td>
<td>German</td>
<td>61</td>
<td>Ovarian (59)</td>
<td>BRCA2+</td>
<td>2017</td>
<td>GC</td>
</tr>
</tbody>
</table>
Table 4. Demographics of study participants

<table>
<thead>
<tr>
<th>Total (N=33)</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Freq or mean</td>
<td>% or range</td>
<td></td>
</tr>
<tr>
<td>Age</td>
<td>56.9</td>
<td>27-77</td>
</tr>
</tbody>
</table>

Cancer diagnosis
- No cancer: 6 (18.2%)
- Breast cancer: 11 (33.3%)
- Ovarian cancer: 16 (48.5%)

Second cancer diagnosis: 3 (9.1%)

Genetic testing
- No: 2 (6.1%)
- Yes: 31 (93.9%)

Genetic test results
- Positive: 11 (35.5%)
- Negative: 18 (58.1%)
- VUS: 2 (6.5%)

Genetic counseling
- Yes: 17 (54.8%)
- No: 12 (38.7%)
- No then yes: 2 (6.5%)

Education**
- Some college or Associate's degree: 8 (25.8%)
- College graduate (4 years): 14 (45.2%)
- Some graduate or professional school: 9 (29.0%)

Insurance at time of genetic testing*
- Yes: 31 (96.9%)
- No: 1 (3.1%)

Marital status**
- Married: 20 (64.5%)
- Divorced: 5 (16.1%)
- Single: 3 (9.7%)
- Cohabitating: 3 (9.7%)

*missing 1; **missing 2

Table 5. Cancer demographics of study participants

<table>
<thead>
<tr>
<th>Total cancer diagnosis (N=27)</th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Freq</td>
<td>%</td>
<td>Mean age at diagnosis</td>
<td>Range</td>
</tr>
<tr>
<td>Breast cancer</td>
<td>11</td>
<td>40.7</td>
<td>43.2</td>
<td>31-56</td>
</tr>
<tr>
<td>Ovarian cancer</td>
<td>16</td>
<td>59.3</td>
<td>51.7</td>
<td>21-69</td>
</tr>
</tbody>
</table>
Descriptive statistics of the pedigree data is also made available (Table 6; for pedigrees, see Appendix B). The 33 pedigrees I drew represent a range of family sizes and cancer histories. Among the relatives included in the three-generation pedigree (siblings/half-siblings, parents, aunts, uncles, and grandparents) there was an average of 4.1 cancer diagnoses per family (range 0-9). In all, there are 135 cancer diagnoses represented among the pedigrees. This includes both cancers related to hereditary breast and ovarian cancer (HBOC) syndrome (i.e. breast, ovarian, prostate, pancreatic, and melanoma) and other types of cancers. Other cancers represent almost half of the cancer cases, with 62 cases (45.9%). Cancers related to HBOC are the majority and represent 73 cases (54.1%). By far the most commonly reported HBOC cancer was breast with 34 cases (25.2%), followed by prostate (12, 8.9%), ovarian (9, 6.7%), pancreatic (7, 5.2%) and melanoma (3, 2.2%). Figure 4 is further visualization of the distribution of cancer diagnoses in the pedigrees, clustered by type of cancer.

---

6 While participants sometimes included great-aunts/uncles or people outside the three-generation tree, they were not included in the descriptive statistics.
7 One person could have multiple cancer diagnoses.
Table 6. Relationships and cancer frequencies based on pedigree data

<table>
<thead>
<tr>
<th>Relationship and cancer diagnosis</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Sibling</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Breast</td>
<td>4</td>
<td>3.0</td>
</tr>
<tr>
<td>Ovarian</td>
<td>2</td>
<td>1.5</td>
</tr>
<tr>
<td>Prostate</td>
<td>2</td>
<td>1.5</td>
</tr>
<tr>
<td>Pancreatic</td>
<td>0</td>
<td>0.0</td>
</tr>
<tr>
<td>Melanoma</td>
<td>0</td>
<td>0.0</td>
</tr>
<tr>
<td>Other*</td>
<td>5</td>
<td>3.7</td>
</tr>
<tr>
<td><strong>Maternal side</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Breast</td>
<td>21</td>
<td>15.6</td>
</tr>
<tr>
<td>Ovarian</td>
<td>7</td>
<td>5.2</td>
</tr>
<tr>
<td>Prostate</td>
<td>4</td>
<td>3.0</td>
</tr>
<tr>
<td>Pancreatic</td>
<td>1</td>
<td>0.7</td>
</tr>
<tr>
<td>Melanoma</td>
<td>3</td>
<td>0.7</td>
</tr>
<tr>
<td>Other*</td>
<td>26</td>
<td>19.3</td>
</tr>
<tr>
<td><strong>Paternal side</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Breast</td>
<td>13</td>
<td>9.6</td>
</tr>
<tr>
<td>Ovarian</td>
<td>2</td>
<td>1.5</td>
</tr>
<tr>
<td>Prostate</td>
<td>8</td>
<td>5.9</td>
</tr>
<tr>
<td>Pancreatic</td>
<td>6</td>
<td>4.4</td>
</tr>
<tr>
<td>Melanoma</td>
<td>0</td>
<td>0.0</td>
</tr>
<tr>
<td>Other*</td>
<td>31</td>
<td>23.0</td>
</tr>
</tbody>
</table>

*lung, skin, leukemia, colon, bone, endometrial, neck, stomach, testicular, jaw, rectal, liver, GI, cervical, uterine, glioblastoma, bladder, unknown

Figure 4. Family history of cancer grouped by cancer type
Summary of methodology and participant demographics

This study required a very specific set of inclusion criteria and had an initial goal of 25 participants, both of which were attained (and participant goal surpassed) through successful outreach and snowball sampling. While embedding myself in the lives of participants would have added to the depth of my data, it was simply not feasible. The objective of the dissertation research was to gather women’s recollections of their experiences with genetic testing, a process that occurred, in some cases, many years before the interview. Rather, I attempted to understand my participants through their interview narratives, through my personal genetic testing journey, and various hereditary cancer meetings. Participants are not reflective of the wider US population as they were primarily white, highly educated and medically insured. However, they are reflective of the population of people who have historically had access to genetic testing and counseling (Han and Jemal 2017).
CHAPTER FOUR: 
INTERVIEW RESULTS AND ANALYSIS

Introduction

The primary research objective of this dissertation is to understand how women at high genetic risk of cancer view and/or have experienced genetic testing and counseling (GT/C) for cancer syndromes. In order to understand the experiences of women who fit this genetic risk criteria, I conducted semi-structured interviews with women that elicited their cancer diagnosis and treatment story (if diagnosed with cancer), since for the majority of women this is when they were exposed to genetic testing opportunities. The cancer story process was open-ended, as I wanted to allow women to share whatever details they felt comfortable sharing about their experience. After, I asked more detailed questions about their GT/C experience – who ordered testing? What did participants expect to happen? What do they remember? How did they feel about their results? These more pointed questions allowed me to uncover the salient themes of the GT/C experience. These results represent the GT/C process from the perspectives of my participants.

Research objective: Women's views and experiences of genetic testing and counseling

One objective of this project was to understand how women at high genetic risk of cancer view and/or have experienced genetic testing and counseling (GT/C) for cancer syndromes. As an exploratory approach, participants were asked open-ended questions from an interview guide: Could you tell me about your GT/C experience and why you decided to undergo GT/C? What did you know about GT/C before undergoing testing? What was the most useful part of your experience? The least useful? Because these were semi-structured interviews, participants would explain their motivations and hopes for GT/C through the interview and not always in response to the question. Through inductive coding, major themes emerged from the raw data. First, main codes of reason for testing and how to use results were used to organize the interview data. Within these groups, I then identified subcodes.
The following are the results of women’s reasons and/or motivations for testing, how they planned to use their test results, followed by their reactions to their genetic test results. As shown in Table 7, the major themes (as represented by total frequency in the last column) women discussed as motivations for testing centered on family (present in 26 interviews, or 81.3%), treatment purposes (20, 62.5%), surveillance of other cancers (14, 43.8%), general information/research purposes (13, 40.6%), or because of Ashkenazi Jewish heritage (6, 18.8%). More than one theme was sometimes mentioned during the course of an interview. Additionally, many of these themes emerged as the reasons for having testing and/or how women planned to use these results. In the thematic analysis section, not all women underwent genetic counseling (defined here as having seen a genetic counselor or genetic professional, GP). For thematic analysis, women’s responses are not separated into those that saw a GP versus those that did not (non-genetics professional, NGP). Later results will review women’s experiences with GP versus NGP. Finally, I am attempting to fit women’s experiences and reasonings (qualitative data) into fixed categories (quantitative boxes). As will be discussed, these categories are by no means impermeable and static (and in many ways, do not mean the same thing to different people).

Table 7. Frequency of themes* reported when discussing genetic testing/counseling

<table>
<thead>
<tr>
<th>Theme</th>
<th>Breast cancer n=11</th>
<th>Ovarian cancer n=16</th>
<th>No cancer n=5</th>
<th>Total n=32</th>
</tr>
</thead>
<tbody>
<tr>
<td>Family</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Family was reason for testing (i.e. prompted testing) or want to give genetic information to family members</td>
<td>63.6 (7)</td>
<td>87.5 (14)</td>
<td>100.0 (5)</td>
<td>81.3 (26)</td>
</tr>
<tr>
<td>Treatment</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Want to use genetic information for treatment (e.g. chemo, surgery, etc.)</td>
<td>72.7 (8)</td>
<td>62.5 (10)</td>
<td>40.0 (2)</td>
<td>62.5 (20)</td>
</tr>
<tr>
<td>Surveillance</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Want to use genetic information for knowledge of other cancers</td>
<td>63.6 (7)</td>
<td>31.3 (5)</td>
<td>40.0 (2)</td>
<td>43.8 (14)</td>
</tr>
<tr>
<td>Information</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Want more information about their cancer, possible treatments, or to give back to research</td>
<td>45.5 (5)</td>
<td>43.8 (7)</td>
<td>20.0 (1)</td>
<td>40.6 (13)</td>
</tr>
<tr>
<td>Ashkenazi Jewish ancestry</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>AJ ancestry discussed as factor in testing</td>
<td>18.2 (2)</td>
<td>18.8 (3)</td>
<td>20.0 (1)</td>
<td>18.8 (6)</td>
</tr>
</tbody>
</table>

*more than one theme could be present in an interview
Theme: Family

Family was by far the most important themes across all three groups, evidenced by the fact that the word *family* showed up in over 80% of the interviews (which did not include a search of other specific family members such as mom/mother, daughter, sister, brother, etc.). During analysis, it became apparent that there were two distinct subcategories in how family was discussed in relation to GT/C: (1) for family to be the reason for testing (i.e. they prompted testing), or (2) participants hoped to use the genetic information for, not only themselves, but for their family members as well. As part of my interview, I asked participants who they shared their genetic test results with, and almost all the women reported discussing their results with family members. However, unless they previously spoke about wanting to share their results with their family as a motivation to have testing, who they spoke to about their results is not included here.

For women without a personal cancer diagnosis (n=5), mothers, sisters, and half-sisters were cited as instigators for testing. In 100% of interviews, participants mentioned family as a reason for testing, with participants further identifying wanting to know their genetic information for their daughter’s or granddaughter’s sake (if they had those relatives). For example, both Becky (30, BRCA1+) and Leigh (27, BRCA negative), two of the youngest participants, had mothers who were diagnosed with breast cancer. Becky’s mother underwent genetic testing and was found to be a BRCA carrier, which spurred testing for Becky and her four sisters. In Leigh’s case, her mother passed away from cancer before Leigh underwent tested, so with urging from her father and conversations with her aunt, Leigh decided to have testing. She recounted:

“I think I kind of resisted it at first because it was something my dad kind of kept pushing for after my mom passed, he would kind of push me and say, ‘Oh you know, at your next doctor’s visit ask them about this cuz your mom had all these indicators and your grandma had this too so you have a family history of it.’”

In both cases, a family member’s genetic testing or cancer diagnosis instigated testing, which prompted testing of the participant, which was thus shared with family members.
Family as a motivation for testing was also high in the breast and ovarian cancer groups, although not quite universal like the group did not have a cancer diagnosis. The breast cancer and ovarian cancer groups provided data which indicated they either were prompted to have testing by family members or hoped/planned to use that information for their family at fairly high rates (breast cancer, 63.6%; ovarian cancer, 87.5%). A major difference between the cancer and no cancer group is that the cancer group took a different route to testing, primarily through providers rather than family. However, family was still an important reason for seeking testing, with many participants indicating that they would like to give the genetic information to their family members. For instance, Marjorie (56, BRCA negative, breast cancer at 56) told me that:

“I was asking her [the oncologist] generally speaking, without knowing the source of why I had cancer, was there automatically a risk to my daughter? And we got into the conversation about her, and then we started talking a little bit more about my family history similar to what I shared with you, and she said you know what, let’s go ahead and do that kind of testing to understand that.”

Other participants with a cancer diagnosis voiced similar concerns and indicated that they wanted genetic test results for the sake of their sons, daughters, sisters, nieces, and granddaughters.

In the group with a cancer diagnosis, the provider was usually the one to prompt testing. This may be way family was not a universally stated motivation to undergo testing, unlike the group without a cancer diagnosis. However, other reasons such as lack of biological children and estrangement of family members (rare in my sample but present), may also be factors. While family was not the initiator for testing in this group, family was still an important theme for the intended use of genetic test results, as illustrated by Marjorie and others (for more quotations, see Appendix C1).

**Theme: Treatment**

Treatment options for breast and ovarian cancer can be grouped into local treatments (e.g. surgery, radiation) and systemic treatments (e.g. chemotherapy, hormone therapy, and targeted therapy). While systemic treatments can reach cancer all throughout the body, local treatments attempt to minimize systemic
impact and solely treat the tumor (American Cancer Society 2018c). Surgeries such as lumpectomy, mastectomy, hysterectomy, and/or bilateral salpingo-oophorectomy are the main treatments for women diagnosed with breast and ovarian cancer, and other treatment options may depend on the type of surgery they initially had, the stage of the cancer, if the cancer had spread to lymph nodes, hormones present in the blood (i.e. ER-positive breast cancer), if the woman has gone through menopause, or even age. The treatment process can be complicated, a sentiment (and frustration) that was brought up by many participants.

Frustrations regarding past or current cancer treatment are not covered here. Rather, this section aims to understand how women connected the idea of receiving a certain course of treatment based upon the results of their genetic testing.

In speaking to women during this research it became clear that the idea of treatment was intertwined with prophylactic surgeries. Prophylactic surgeries are not considered treatment in a medical sense because it is a surgery to remove healthy tissue, thereby reducing the risk of developing disease in the future. However, these surgeries also do not fit into the surveillance category (discussed next), either. Prophylactic surgeries would technically fit into a prevention category, but through the use of corpus analysis software called AntConc (Anthony 2018), it was found that the word “prevent” and its various forms (i.e. prevention, prevented) only appeared 5 times in the entire corpus of interview transcripts (from 4 unique interviews). The term “prophylactic” was present 20 times, but only from six unique interviews. From this standpoint, prophylactic surgeries inhabit a sort of no (wo)man’s land, between treatment and surveillance, as a preventative strategy that was not explicitly stated as such in interviews. Since prophylactic surgeries were most commonly brought up in the context of treatment, I have included them in this category. Further, if talk of prophylactic surgeries was not included, then women without cancer (n=5) would not be represented in how they hoped to use their genetic test results, in regards to access to surgeries; the responses of “wanting surgery” due to their genetic test results would be absent.

Women in the breast and ovarian cancer groups spoke about treatment in light of their genetic testing at about the same frequency. Eight women (72.7%) in the breast cancer group and ten women (62.5%) in the ovarian cancer group spoke about the ways in which genetic testing either informed treatment,
or how they hoped it might help treatment in the future. Treatment as a result of genetic testing was seen by women as influencing surgeries, chemotherapy, and the types of prophylactic surgeries a woman should consider. In a couple cases, prophylactic surgeries turned out to not be prophylactic, as cancer was found after removal of supposed “healthy” tissue and would be then classified as treatment (another reason to include prophylactic surgeries in this section).

Surgery as treatment was an important topic for how genetic tests could be used. Women in all three groups (breast, ovarian, or no cancer) spoke about how they either planned to, or did use, their test results to inform different surgeries. For instance, after Lisa’s (66, BRCA2+) breast cancer diagnosis at 55, she stated that she thought she would have a lumpectomy and radiation like her mother had gone through; she in fact did have the lumpectomy. However, when her test results came back BRCA2 positive a couple weeks after her lumpectomy, she then went back for a mastectomy and ultimately did not have radiation. In another case, Lillian (48, BRCA negative, breast cancer at 42) recalled that genetic testing:

“…would determine what type of surgery I was gonna have, mastectomy or lumpectomy vs. what type of treatment might be needed, so I was very aware of what the results, what the final results presented would determine in terms of treatment and surgery options.”

Lillian acknowledged that while she was not sure exactly what the steps in her treatment journey would, she knew that genetic testing results would be used in decisions with her OBGYN, oncologist, and breast surgeon/oncologist. In fact, Lillian may have used her genetic test results to push against the recommendations she received from two different providers saying she should have a bilateral mastectomy. In the end, she did not have a mastectomy and did not undergo chemotherapy, which is what she wanted. In both cases, the results of genetic testing were used in very specific ways by my participants to determine the course of treatment.

While many women diagnosed with cancer were concerned with what kind of surgery they would have based on their genetic tests results, women diagnosed with ovarian cancer also mentioned other specific
types of treatment, including chemotherapy and PARP inhibitors. Among women with germline BRCA1/2 mutation, PARP inhibitors have produced significant improvement in long-term control of ovarian cancer (Staropoli et al. 2018). Given these improved outcomes, it may not be a surprised that women diagnosed with ovarian cancer were hoping (and knew how) to use their BRCA test results to help them gain access to this type of targeted therapy. Angie (70, BRCA negative, ovarian cancer at 66) mentioned that she was aware that if “I tested positive there were trials for PARP inhibitors that I could then participate in”. Mentions of PARP inhibitors occurred ten times in six unique interviews.

For other women the uses of genetic testing were a bit vague and were talked about in terms of future use. First, prophylactic surgeries were mentioned as potentially occurring in some indeterminate future, only being considered due to the knowledge of a woman’s genetic testing results. When Joann (47, BRCA1+, ovarian/fallopian tube cancer 44) received her BRCA1 positive results at age 44, she said her doctor:

“…was very forthright with me. He said you need to get everything removed. He said you need to get a mastectomy, and you need to…he said you don’t need your ovaries, he said you don’t need these things anymore.”

While Joann was told she “needed” to have these surgeries, women not diagnosed with cancer also expressed a desire for surgery. Julie and Becky both spoke about “wanting” surgery based on their genetic test results. For others, they hoped that they could use their results to help in the case of cancer recurrences. Roxanne (61, BRCA1 positive, ovarian cancer at 52), who had already been through one ovarian cancer recurrence, stated that if she had another recurrence or was diagnosed with another cancer, that she hoped the information could be used to target her therapy better than if she had not had the genetic testing. This was an idea repeated by Melanie (60, BRCA negative, breast cancer at 42, ovarian cancer at 56) that, as she understood it, genetic testing could help specifically in the cases of ovarian cancer recurrence, because doctors would “know better how to treat people with a reoccurrence that had the genetic mutation”.

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8 According to the National Cancer Institute’s dictionary of cancer terms, poly (ADP-ribose) polymerase or PARP, an enzyme which helps repair damaged DNA, can be blocked to help keep cancer cells from repairing their DNA, which causes them to die. It is a type of targeted therapy. For more see: [https://www.cancer.gov/publications/dictionaries/cancer-terms/def/parp-inhibitor](https://www.cancer.gov/publications/dictionaries/cancer-terms/def/parp-inhibitor)
Theme: Surveillance

Medical surveillance is not meant to prevent cancer, but rather to catch cancer in its earlier stages in order to have better treatment outcomes. These surveillance techniques include screenings such as clinical breast exams, MRIs, mammograms, CA-125 (tumor marker), and transvaginal ultrasounds. Surveillance guidelines are put together by the National Comprehensive Cancer Network® (NCCN®), a not-for-profit consortium of cancer centers which develops resources for patients and practitioners (www.nccn.org). For example, NCCN® guidelines for a woman with a BRCA positive or likely pathogenic variant are: (1) clinical breast exams every 6-12 months starting at age 25; (2) breast screenings of an annual breast MRI screening with contrast between age 25-29, annual mammogram and MRI between age 30-75; (3) and discussion of options for risk-reducing mastectomy and salpingo-oophorectomy (Daly et al. 2018). For women who have been diagnosed with ovarian cancer and finished treatment, guidelines for monitoring include: (1) visits every 2-4 months for 2 years, then 3-6 months for 3 years, then annually for 5 years; (2) physical exams including pelvic exams; (3) chest/abdominal/pelvic computerized tomography (CT), MRI, positron emission tomography (PET) scans as clinically indicated; (4) chest x-rays as clinically indicated and (5) CA-125 or other tumor markers (Armstrong et al. 2018).

An important distinction should be made at the outset that medical terminology and ideology of medical surveillance and how participants where describing their planned uses of GT/C were quite different from one another. This is an important distinction that only close, qualitative work can uncover, and is important if we are attempting to improve clinician dialogue with patients to improve health outcomes and understanding. Medical surveillance is not meant to prevent disease but rather to catch it in its earlier stages and is accomplished through various screening techniques. Medical surveillance, in medical fields, is a narrow hallway encompassing specific procedures with specific outcomes. In my interviews, however, surveillance seems to be enmeshed with ideas of preventing cancer (as discussed above, see Treatment). For example, women stated that they were glad they had the GT/C information because they then knew their risks for other cancers; ergo, they would then know what their options were to “hopefully prevent future cancers” (Lisa, 66, BRCA2+, breast cancer at 55). Other participants were told by various healthcare professionals that
they should consider panel testing for multiple genes so that they could be assessed for their risk of other cancers, some of which they had no prior idea that they were at risk for. In this sense, surveillance is more closely tied to the Latin word from which it is derived, *vigilare* – to keep watch. Vary rarely did women bring up the specific screening measures tied to medical surveillance. One younger participant, Becky, mentioned self-breast exams, and others mentioned “screenings” and “monitoring” for these other cancer threats. While the looming threat of other cancers caused many of the participants to want to “keep watch” for these diseases, they may not have known by which specific medical pathways they would keep watch.

While many women spoke specifically about the surveillance they had done or currently were doing for their cancer diagnoses, I sought to understand how they considered surveillance in light of GT/C. Among the three groups, women in the breast cancer group brought up surveillance at the highest frequency: 63.6% in the breast cancer group, 31.3% in the ovarian cancer group, and 40% in the no cancer group. Based on the interviews, there appears to be a sense that GT/C will provide some sort of heads up for other cancers, either for the women themselves or their family members. This is understandable given the repercussions for women and their family members if a positive mutation is found. For instance, Joann (47, BRCA1+, ovarian/fallopian tube cancer at 44) stated that knowing they have this “thing” (BRCA mutation) in the family means that other family members need to “do your homework and stay on top of it.” Other than warning family members, women also reported being more aware of other cancers. If the participant had breast cancer first, ovarian cancer was cited as a concern. Vice versa, if they had ovarian cancer first. Lynne (60, BRCA2+, ovarian cancer at 64) stated, “I wasn’t going to fight ovarian cancer and then turn around and get breast cancer if I could help it.” Other cancers that women mentioned being concerned about include pancreatic and colon cancer. Hereditary breast and ovarian cancer syndrome, which is principally associated with mutations in the BRCA1 and BRCA2 genes, does confer an increased absolute lifetime risk for certain cancers such as breast (46-71%), male breast (2.8%), ovarian (17-46%), prostate (7.5%), and pancreatic (1-7%) (Centers for Disease Control and Prevention 2015). Melanoma (absolute cancer risk 0.1-2.4%) is also at increased risk, albeit one of the lower risks within the HBOC cluster, but was not mentioned specifically.
Some misunderstandings, or misinformation, were also discussed during interviews. For example, Yvonne (49, BRCA2+, ovarian cancer at 49) stated that she was “80% more likely to get breast cancer with this gene”. A prospective cohort study of almost 10,000 BRCA mutation carriers found that the cumulative breast cancer risk to age 80 was 69% for BRCA2 carriers (72% for BRCA1 carriers) (Kuchenbaecker et al. 2017). Second, Sissy (62, BRCA negative/Lynch VUS, breast cancer at 49), who had genetic testing two times and came back with what she described as a Lynch VUS (variant of uncertain significance). VUS results are reported about 5% of the time after BRCA1 and BRCA2 testing, and there have been documented cases of physicians misinterpreting VUS results (Culver et al. 2013). With proper training in genetics, one should know that a VUS result is usually counseled as a negative since they are typically reclassified as negative, which is supported in a recent study that looked at over 1 million individual test results (Mersch et al. 2018). However, Sissy hopes to use her VUS result as a reason to have her doctor give her a colonoscopy every year rather than every ten. NCCN guidelines recommend colonoscopy every 1-2 years only if the person is a MLH1, MLH2, MSH6, PMS2, or EPCAM mutation carrier (which Sissy is not) (Provenzale et al. 2018). It is interesting to note that the only other person to mention a percentage in regards to their increased risk was correct. Joyce (60, BRCA2+ and NBN+, ovarian cancer at 57) noted that her pancreatic cancer “risk is 7% greater than the average person’s risk of pancreatic cancer”. These specifics are important as they indicate potential misunderstandings of women’s risk for other cancers as well as misconceptions for how genetic testing may be used for surveillance purposes.

**Theme: Information**

Education, knowledge, studies, research – these types of ‘information’ were mentioned by participants when they described how and why they pursued genetic testing, how they hoped to use their genetic testing information, and/or outcomes of having gone through the testing process. In short, many women seemed to agree with the statement that, “[K]nowledge is a good thing” (Melanie, 60, BRCA negative breast cancer at 42, ovarian cancer at 56) or that “knowledge is power” (Yvonne, 49, BRCA2+, ovarian cancer at 49), which Sissy is not).

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9 Lynch syndrome, or hereditary nonpolyposis colorectal cancer (HNPCC), is an inherited disorder which increases the risk of cancers of the colon, rectum, stomach, small intestine, liver, gallbladder ducts, upper urinary tract, brain, skin, ovaries, and endometrium. [https://ghr.nlm.nih.gov/condition/lynch-syndrome](https://ghr.nlm.nih.gov/condition/lynch-syndrome)
cancer at 49). Among the different groups, the idea of information/research was brought up at about the same frequencies: 45.5% in breast cancer (n=5/11), 37.5% in ovarian cancer (n=6/16), and 20% with no cancer (n=1/5).

Overwhelmingly, women viewed the information that genetic testing generated as a “good thing”, saying that it would potentially help solve various medical mysteries – either their own or for others, citing cancer and other diseases such as multiple sclerosis – and/or empower them. One participant stated that she could not understand why someone would refuse testing, “unless they had something to hide” (Helen, 43, BRCA negative, breast cancer at 40). Only one participant expressed ambivalence at the knowledge she was gaining through the testing process. April (39, BRCA negative, breast cancer 31), while “thrilled” about all the information the genetic counselor was telling her, did not feel like she had the necessary context to be able to understand and use the information. In her words, “I don’t have a box to put this in, I don’t have filter to run this through to translate it into my words, or how to make it apply to my life or my body.” Even then, April was sure to tell me that she was happy to have the information given to her. This reaction seems to fit the idea of science as a cultural authority, that “we” should be happy to contribute to; that “Science Makes the Modern World” (Shapin 2007). In this mode of thinking, the laity is not supposed to truly understand exactly what “science” does, but instead what should be apparent is “knowing where to look for it, knowing who are the relevant authorities, knowing that we can and should assent to what they say, that we can and should trust them in their proper domains” (Shapin 2007:441). Participants’ responses for someone to “figure this [disease] out” (Katherine) by “collect[ing] all the information you want” points to a trust in science and its purveyors.

**Theme: Ashkenazi Jewish heritage**

The final and least frequent theme that was discussed as a motivation for GT/C was Ashkenazi Jewish (AJ) heritage. BRCA mutations in the general population are quite rare, with an estimated 1 in 300 to 500 women, or about 0.2% to 0.3%, being carriers. Among women with AJ heritage, the prevalence of BRCA mutations increased to 2.1% (U.S. Preventive Services Task Force 2015). As a result, women with AJ heritage
are considered eligible for GT/C per NCCN© guidelines (Daly et al. 2018). Because of the increased prevalence of BRCA mutations, there have been studies to assess the impact of population-based genetic testing for those with AJ heritage (Manchanda et al. 2015), with a recent study supporting the cost-effectiveness of population testing in people with 1-4 AJ grandparents (Manchanda et al. 2017). In this study sample there were six participants (18.2%) who identified some amount of AJ ancestry (Table 8). While these participants relayed that they had AJ ancestry, not all of them spoke about it in the context of GT/C.

**Table 8.** Characteristics of participants who reported Ashkenazi Jewish heritage including cancer diagnosis, BRCA status, and if they discussed AJ heritage during the interview

<table>
<thead>
<tr>
<th>Pseudonym</th>
<th>Paternal Ancestry</th>
<th>Maternal Ancestry</th>
<th>Cancer</th>
<th>BRCA mutation</th>
<th>Discuss AJ?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lisa</td>
<td>Ashkenazi Jew, Russian</td>
<td>German</td>
<td>Breast</td>
<td>BRCA2</td>
<td>Yes</td>
</tr>
<tr>
<td>Gwen</td>
<td>Ashkenazi Jew</td>
<td>Ashkenazi Jew</td>
<td>Breast</td>
<td>BRCA1</td>
<td>No</td>
</tr>
<tr>
<td>Angie</td>
<td>Spain, Portugal, Sephardic Jew</td>
<td>Ashkenazi Jew, Polish</td>
<td>Ovarian</td>
<td>Negative</td>
<td>Yes</td>
</tr>
<tr>
<td>Clara</td>
<td>Ashkenazi Jew, Italian</td>
<td>Ashkenazi Jew</td>
<td>Ovarian</td>
<td>Negative</td>
<td>Yes</td>
</tr>
<tr>
<td>Kristy</td>
<td>Ashkenazi Jew</td>
<td>Ashkenazi Jew</td>
<td>Ovarian</td>
<td>Negative</td>
<td>Yes</td>
</tr>
<tr>
<td>Julie</td>
<td>Ashkenazi Jew</td>
<td>Ashkenazi Jew</td>
<td>No</td>
<td>BRCA1</td>
<td>Yes</td>
</tr>
</tbody>
</table>

For those with AJ ancestry, most testing was prompted in part because of their AJ ancestry. During discussions with medical providers, it became apparent to Lisa, Angie, Kristy, and Clara that their AJ ancestry was an important indicator for pursuing GT/C since providers explicitly asked them about Jewish ancestry. In the case of Gwen and Julie, this does not seem to be the case. Even though Gwen (53, BRCA1+, breast cancer at 45) shared that she had both maternal and paternal AJ ancestry, when I asked her whether she knew about the higher risk for BRCA mutations among AJs she relayed that growing up she didn’t know about her ancestry and then “all of a sudden it became a thing.” Gwen had testing around the time it first became available in 2002, and it was then that she learned that “Ashkenazi Jews have this defect on this gene and whatever”. To further complicate the issue, she told me that her mother’s father’s family was estranged so she wasn’t getting much familial information from them. Likewise, Julie (71, BRCA1+, no cancer) also had testing around 2002 and stated that, after prompting from me about her AJ ancestry, stated that while her family’s ancestry was possibly an indicator to her doctor, it was also the “early days of information”. Since genetic
testing had only recently become available, more important to her was her mother’s cancer diagnoses (breast cancer at 64, ovarian cancer at 62), which was her primary reason for being closely followed by her doctor.

**Women’s reactions to genetic test results**

Explanations for why women want to undergo GT/C is only a small part of the GT/C experience. Their reactions to receiving test results varied and were sometimes wildly different. A total of 31 participants underwent genetic testing, with 18 women (58.1%) receiving negative test results, 11 (35.5%) receiving positive BRCA results, and 2 receiving a variant of uncertain/unknown significance (VUS) result (6.5%).

While seeing the participants divided initially by cancer diagnosis is useful, it may also be useful to visualize the breakdown of participants by genetic testing status and whether they saw a GP or NGP. Figure 5 shows that most of the participants had genetic testing (93.9%), and most saw a GP (51.5%).

![Figure 5. Diagram of participant’s genetic testing status, provider type, and test results.](image)
Women reported a wide range of reactions to their test results, from indifference, to shock, to anger, to relief, to a variety and mixture of feelings. In order to ascertain the impact the provider has on women’s experiences, groups are divided into positive and negative test result (VUS results are grouped with negative results), and then further subdivided by whether the participants had seen genetics professionals (GPs) or not (NGP).

Positive result, non-genetics professional (NGP)

While there were only 5 women (16.1%) who received a positive result in the NGP group, it is striking that the main emotions elicited were largely negative – shock, fear, anger – or simply to not report a reaction. When I asked Gail (71, BRCA+, breast cancer at 42, ovarian cancer at 67) how she felt about her BRCA positive results, she stated, “I don’t know. I just remember Dr. K told me I was BRCA positive.” This response reflects a few possibilities: either Gail was not informed of how these genetic test results would impact her medical treatment, care, and surveillance; Gail was informed but does not remember; or else Gail was not concerned about a positive BRCA result one way or the other. Regardless, these possibilities imply that she had no frame of reference, and thus no discernable reaction, to arguably very important information about her health. Julie (71, BRCA1+, no cancer), on the other hand, had quite a strong reaction to her results. Julie described that receiving news of her results was like getting the wind knocked out of her. In her case, she did not have genetic counseling, she had testing through a surgeon, and was told her test results by her husband when she came home one day. She recounts:

“So, I was tested and I found out…actually my husband is the one who told me because the doctor called and talked to him. Well my husband is a doctor and they were colleagues, but it’s not the way to find out. So, I wasn’t really prepared. So that was the extent of my genetic counseling, which was none.”

Julie stated that she did not expect her results to be positive, meaning she was not prepared for the potential outcomes of the test; this is a point that is usually covered during a genetic counseling appointment. Not only did she turn out to be a BRCA carrier, but she arrived home to have her husband tell her the news out of the
blue. On top of that, she said her husband was “really upset” by the news because he knew what it entailed, mainly that she would likely undergo prophylactic surgeries. The lack of preparation and respect for Julie’s autonomy by the surgeon who ordered testing culminated into a very distressing situation when Julie was told her results. In fact, the lack of preparation by ordering providers for most of the women in this group seems to be directly related to the reactions they had to their test results – at best, an innocuous reaction and at worst, a distressing one.

Joann (47, BRCA1+, ovarian/fallopian tube cancer 44) may be an example of how a lack of preparation during genetic testing can have consequences further down the road. Like Gail, Joann did not have a strong reaction to her test results mainly because she didn’t seem to understand what they meant. In her interview, she mentioned multiple times that she did not know what BRCA was. She said after she received her positive test results her “brain like shut down, I’m like surgeries? Double mastectomy? Remove ovaries, fallopian tubes, all that why? I’m hearing, menopause, and I’m like what? I don’t want to go into menopause!” For her, the information that stood out were the prophylactic surgeries she was being told about. Given that she had testing through a breast surgeon, this may reflect his bias and expertise. When I asked her to explain how she made sense of BRCA, she said, “it’s this horrible gene” and “he [breast surgeon] said it’s good I never had children…and he said to me, this is not something that you want to pass down. I just know it’s not a good thing to have.” Joann had watched one of her sisters die of cancer, another be diagnosed and undergo treatment for breast cancer, and was then advised to undergo prophylactic surgeries soon after receiving her test results, with the understanding that BRCA is a “horrible gene” and it was good she never had children to pass it on (per her breast surgeon). When we spoke, Joann confided that she was having a very hard time after her surgeries – a prophylactic double mastectomy with reconstruction, and a supposed-to-be prophylactic bilateral salpingo-oophorectomy in which they found cancer in her ovaries/fallopian tubes. She told me, “the doctors save your life, they do what they need to do. I was very blessed to find these wonderful doctors but now I’m left to figure it all out and it’s a big struggle now, and it’s hitting me big time. Big time. So now I’m like a mess.” We spoke about how she felt rushed through the whole process, and while she felt like her body was taken care of, her mind was not. While genetic testing is a
small piece of Joann’s larger medical journey, it was one that she did not feel well prepared for. Her positive BRCA1 results were the snowball to start an avalanche, one which left her disoriented, depressed, and struggling with body issues.

**Positive result, genetic practitioner (GP)**

In stark contrast to the women who had testing with an NGP, women in this group (n=6, 19.3%) appeared to be much more accepting of their carrier status, claiming they “were not surprised” by the results, or they already knew they would be carriers. Yvonne (49, BRCA2+, ovarian cancer at 46) told me that when she received her BRCA2 positive test result, she “already knew it. It was just confirmation of something I already knew, so it was like, okay.” Bonnie (61, BRCA2+, ovarian cancer at 59), went so far as to say that being BRCA1 positive made her “feel good, like it wasn’t my fault. It wasn’t what I ate…it gave me a reason why I got it, cancer.” One woman (Gwen, 53, BRCA1+, breast cancer at 45), rather than describing her own reaction, went into much more detail about how one of her sisters tested negative and felt guilty that she was the only sister to come back negative. Becky (30, BRCA1+, no cancer) also expressed guilt. In this case, it was because she thought she had not pushed hard enough to receive genetic testing 10 years previously. When her mother was diagnosed with breast cancer, Becky said she felt it could have prevented if only she had gone through with genetic testing, which would have identified her as a BRCA carrier and thus prompted testing for her mother before her breast cancer diagnosis. The striking contrast between this group, with their relatively benign and/or positive reactions to positive genetic test results, and the previous group discussed (positive results, NGP), speaks to the possible role that GPs have in preparing women for receiving and dealing positively with genetic test results.

**Negative/VUS test result, non-genetic practitioner (NGP)**

Women in this group (n=9) reported being relieved and even “thrilled” about their test results, but most reported having mixed feelings. As Leigh (27, BRCA negative, no cancer) told me, “I wasn’t as relieved as I thought I would be getting the test results, I was still kind of freaked out about it.” Some of Leigh’s anxiousness may have stemmed from her mother’s aggressive and late breast cancer diagnosis, which had
ultimately been fatal. Diane (71, BRCA negative, no cancer) voiced a similar concern as Leigh after receiving her results. She told me that when she was told her results, “it made me feel a little bit less stressed about getting it [cancer] but then not really because I said, ‘mother didn’t have it and she had ovarian [cancer],’ know what I mean? It’s sort of like you think, okay well I’m in the clear but I’m not.” For these women, the first-hand experience of their mother’s cancer diagnoses was as prominent (if not more) than their negative genetic test results.

While some of the respondents in this group mentioned that part of them felt relief from the test results, there was lingering hesitation or questions about why they got cancer. For Kristina (60, BRCA negative, ovarian cancer at 21, breast cancer at 55) a negative result was confusing and not what she was expecting. She told me that:

“… it was really weird to hear back that no, I didn’t have those markers. And the breast specialist said, ‘well it’s probably just some kind of a genetic flaw but they haven’t found that particular gene, they’re not testing that particular gene.’ I was just stunned! It was sort of like, should I do that test again? Could we do a redo? I was so expecting it to say yes, and when it was no it was, I thought that was gonna be the thing that made my decision for me. So, I don’t know if you could say I was disappointed, but I was surprised, I was really surprised.”

Kristina’s early ovarian cancer diagnosis at 21, with her second cancer diagnosis of breast cancer at 55, led her to believe that there must be a genetic reason for her cancer. And she was not the only participant who hoped that a positive genetic test result would give them a reason for their cancer diagnosis, even though cancer is multifactorial and complex. Marjorie (56, BRCA negative, breast cancer at 56) told me that, “I had mixed feelings about the results and that probably sounds crazy,” because while she was relieved that her daughter and future grandchildren were safe from hereditary disease, she was frustrated because she was not “able to understand how this [cancer] happened.” While some of this ambivalence may have come from lack of discussion on the part of the NGP, equivocating may have stemmed from the practitioners themselves.

Kristy (71, BRCA negative, ovarian cancer at 69) reported that the “doctor [gynecological oncologist]
explained to me that it [test result] was good and bad,” indicated that while negative would be good for her family, not being a BRCA carrier could potentially limit “some of the avenues that were available for treatment.”

**Negative/VUS test result, genetic practitioner (GP)**

Women in this group (n=11) exhibited some of the ambivalence present in the negative test result, NGP group, speaking about the relief the test results gave them but also the disappointment in the negative result. This ambivalence and need for answers is very similar to the negative test result-NGP group. Two women specifically, Katherine (49, BRCA negative, breast cancer at 35) and Clara (57, BRCA negative, ovarian cancer at 54) wanted answers for why they got cancer – answers they perceived might come from a positive genetic test result. Katherine, with triple-negative breast cancer, hoped that genetic testing may help her figure out what was “feeding or driving” her cancer, while Clara reported that a positive test result would “give you a place to put the blame”. Angie (70, BRCA negative, ovarian cancer at 66) also confided that she:

“…was disappointed which sounds strange, because I felt there was more research on BRCA positive patients and there would be more trials for me. But on the other side I was relieved that I didn’t have it. I was ambivalent…I should say, I was relieved that I hadn’t passed it on.”

Other women reported ambivalence and relief. Relief for family members and daughters who were not at risk for inheriting the BRCA gene. Or relief from the anxiety of waiting and thinking about the *what ifs* of genetic testing. In Courtney’s (37, BRCA negative, unknown VUS, no cancer) case, she articulated that she “was relieved given that build up from the initial visit [pre-test genetic counseling]. There’s just a tension that’s built into the, you build the scenario up and there is no certainty”. For others, genetic testing was almost like a routine test to have done, an ‘i’ to dot, as Jill (64, BRCA negative, ovarian cancer at 51) put it. For her, the test was “not a big deal” and was a confirmation of what she already felt to be true.
Women’s experiences with a genetic practitioner (or not)

During these interviews, I attempted to clarify what kind of provider women were referring to – if she said doctor, I attempted to clarify what kind of doctor. If they spoke about having genetic testing done, I clarified who ordered the testing. In this way, I was able to determine, according to self-report, what kind of providers participants had genetic testing through. This allowed me to divide genetic counseling experiences into groups of those who (1) saw a genetics professional (GP) (n=17, 51.5%); (2) those that did not (n=12, 36.4%); and (3) a smaller group who had testing done twice – who initially did not see a genetics professional but then did (n=2, 6.1%). Based on these interviews, there is a stark difference in the information that women receive prior to and after having genetic testing depending on the type of provider they are seeing. As shall be discussed, women who saw NGPs (e.g. OBGYNs, gynecological oncologists, surgeons) usually received much less information than women who spoke to GPs (genetic counselor or some other type of genetics professional). However, even though it is apparent that less information and time is being given to women who had testing through NGPs, women remember little of their actual encounter with the GPs. The following vignette is a good illustration of how women reported their interactions with both GPs and NGPs.

Vignette: Joyce’s experience(s)

Joyce (60, BRCA2+ and NBN+, ovarian cancer at 57) did not have genetic counseling initially but because of her positive genetic test results she encouraged her daughter to also undergo testing. Her daughter, on the other hand, did go to a genetic counselor and Joyce accompanied her, affording her two very different perspectives of the process. Joyce detailed how she had to initially fight to get genetic testing:

“I said, ‘are you [oncologist] going to do it?’ ‘Well, we’ll do it when frontline chemo is over. But you probably really don’t need it.’ I don’t know why he thought that, but they didn’t think I needed it. See the kicker to me of the whole thing is my normal gynecologist who I saw every year for 20 years, I told him every year, don’t forget: mother had breast cancer, her sister had ovarian cancer. Because to me, regardless of genetics, that was a big red flag. And every year the gynecologist would say, ‘were they diagnosed before the age of 50?’ And I would say, ‘no’, and he’d say, ‘then you don’t have anything to
worry about.’ Well, we all now know that that’s not accurate science and he should have known better, long before I was diagnosed. So that’s why I insisted through the oncologist that I get the genetic testing done.”

Joyce was well aware of her family history and how she may have an increased risk of gynecological or breast cancer. Once she started to experience symptoms of what would eventually be diagnosed as ovarian cancer – but was alternatively diagnosed as hypothyroidism, diverticulitis, and appendicitis for over a year – she finally had genetic testing through her oncologist, but only after she asked directly. Joyce described her genetic testing experience with her oncologist and contrasted that experience with her daughter’s genetic counseling:

“The onc [oncologist] ordered it [genetic test], the blood was drawn, sent off, came back, and he tells me the results. I don’t remember what I was asking him, but I was asking him some questions about the results, and he said, ‘do you want to see a genetic counselor?’ I said absolutely. So, he sends me across town to another one who says, ‘oh yes I do all the genetic counseling for this cancer center.’ Well, she was a hematologist, and I’m not saying that she didn’t know what she was talking about, but I recently went to a genetic counselor that my daughter, I went with my daughter to her appointment with a true genetic counselor. Biiiiig difference in how the conversations went. The basic science information was provided to me. I think what wasn’t provided was more of the emotional support, and how do you feel about this kind of interaction.”

First, Joyce describes how the testing process went with the oncologist as incredibly succinct – he ordered it, drew blood, sent it off, it came back, and she was told results. She did not report any psychosocial assessment, nor a discussion of the possible test outcomes, medical implications, or medical management options. Joyce describes the genetic testing through her oncologist almost like any other blood test that is ordered for a patient. And yet, this was a of blood test that the oncologist, when asked questions about the outcome, did not feel comfortable discussing results, as evidenced by his referral of Joyce to someone else. When Joyce accompanies her daughter, she realizes that one thing her daughter got, which she did not, was emotional support “and how do you feel about this kind of interaction”. Per Joyce’s recollection, the
discussion about what was occurring during genetic testing was had with her daughter, but not during Joyce’s initial testing experience. When I asked Joyce if there was anything else she noticed that was different between her two experiences she stated:

“There was a lot more education, my daughter got a lot more education about what the science is currently knowing about her mutation, which are the same as mine. The BRCA2 and the NBN. So, she got a lot more education from that lady [genetic counselor]. I basically got yup, that’s what you’ve got, and here are your additional risk factors for breast and melanoma and pancreatic and colon, I think that’s it.”

Joyce’s experiences provide crucial insight into the basics of how women described their experiences. In the case of women who had testing through a NGP, the process is described much like any other test that a doctor would order for a patient without much explanation of why it is being done. Sometimes the NGP would have women read brochures and packets of information rather than explain the process or implications in person (Kristina, Marjorie, Table XXXXX). In Leigh’s case (27, BRCA negative, no cancer) her OBGYN ordered testing for her, which she said they described as, “Oh we’ll send it off to this lab, they’ll do XYZ testing, and you’ll get your results in a month. They didn’t really cover too much of the details.” When her results did arrive, she received a packet that had, “information on what the test results mean, just kind of what the indicators were and why you should get tested which I thought was a little after the fact, like well I already did the test that’s why I got this packet.” While the packet did not have her test results, Leigh received a phone call from a nurse at her OBGYN office who called and:

“…told me that my test results were back and that I was negative and asked me if I had received the packet and I was like yeah, and she was like okay. I was kind of, I don’t want to say they were cavalier about it, but it was all very like…I don’t know. It all just seemed kind of quick and just like, okay well we’re done with this.”

Leigh described this interaction as feeling very “procedural”, which could be applied to most of the NGP genetic testing encounters. Genetic testing was simply another procedure that was ordered by their doctor.
which at best, women described as a routine test with discussion of family history or insurance in rare cases. At worst, it is a procedure that is done without the knowledge of the patient. In an extreme example, one woman reported that she was completely unaware that she was having testing done. Her gynecological oncologist took her blood during her surgery and conveyed her results later (Gail, 71, BRCA+, breast cancer at 42, ovarian cancer at 67). In the end, Joyce gives me her final verdict on her experience(s):

“Not being able to go initially to a true genetic counselor was kind of a waste of my time because I could do the research and get the same stuff that oncologist gave me on my own. It was kind of useless. And it was more than just useless because she didn’t really say, you should see this doctor or that doctor and this is what they ought to do. She kind of left it to me to figure out well, if you have a higher risk of melanoma maybe you should see a dermatologist, versus when my daughter saw the genetic counselor, she actually said, let me help you make an appointment with the following physicians, let me send my report to them. I think that makes a big difference.”

**Genetic counseling/testing with a genetic practitioner**

While women who had genetic testing through a NGP describe the process as a routine procedure with little information divulged regarding the consequences or potential implications of testing, women who had testing through a GP also were sometimes hard-pressed to “recall the details of that day” (Courtney 37, unknown VUS, no cancer). During these interviews, I had to probe to get any details about the genetic counseling experience, and even then, the details were vague. Participants had genetic counseling as early as 2000 and as recent as 2017, with nine having had genetic testing in 2014-2017.

Roxanne (61, BRCA1+, ovarian cancer at 52) and Courtney (37, unknown VUS, no cancer), who have two different contexts and stories, both described the encounter as somewhat impersonal. In Roxanne’s experience, she did not feel like the GP situated her counseling within her experience with cancer. She told me that:

“The spiel was, to me, it was the spiel you would give to someone who had never had, or never battled cancer. To me it was kind of…she had to tell me all the facts and, you know, she gave me the"
sheet with all those little DNA markers that are flawed, you know? I looked at it and we talked about it, and it was kind of...okay, now I’m gonna go on with living.”

Courtney, who did not have a cancer diagnosis and underwent testing at the behest of her half-sister, described her counseling experience as very “call-centerish” and generic:

“The person [genetic counselor] was taking me through this flip notebook with all these tabs, and it seemed as though, [they] was trying to be very delicate in terms of talking about probability and what various things could possible mean and that nothing is a guarantee, and it seemed like [they were] gonna say the same thing to anybody who came back with my particular result regardless of who I was. And it felt very impersonal, and very scripted. I was surprised by that. I don’t know why I was surprised by that. But I was moved by that. There didn’t seem to be any recognition of what I already knew, or what I was capable of knowing, there was just a lack of recognition of me. And I was pretty surprised that anything that was revealed in that follow up I could have read online, or been emailed, or...there was really no necessity to that follow up, I didn’t learn anything there other than the DNA sequencing or whatever suggested I hadn’t been a recipient of that genetic mutation, and I was like well that probably could have been handled in a very different way just in terms of like, what they were considering the patient education. It didn’t really feel to me like, educational.”

In both of these cases, the women’s experiences, knowledge, or viewpoint may not have been taken into consideration for the counseling session. Rather, the focus was on purportedly neutral facts and information, which unfortunately didn’t even accomplish the educational aspect of counseling because Roxanne and Courtney were so put off by the impersonal nature of their experience.

For other women, genetic testing was simply another medical procedure to be completed. Lisa (66, BRCA2+, breast cancer at 55), described her counseling as a means to an end in order to get her genetic test results. Lisa stated that, “I didn’t really need much because I wasn’t depressed, I was just ready to do whatever I had to do.” Lisa described her genetic counselor as “very good” but felt that she didn’t need “mental counseling”. Similarly, Helen (43, BRCA negative, breast cancer at 40) said that she has “no idea how
genetics works, it doesn’t matter to me” because when the doctors told her they’d like her to have testing done she “just agreed”. However, Helen did remember that the genetic counselor “explained what the BRCA meant, and the HER2\textsuperscript{10}, and likelihood of recurrence if you’re positive” along with the “higher risk if you’re certain ethnicities”. Helen noted that sometimes she would be in the hospital for 12 hours and would feel rushed from appointment to appointment, so the genetic counseling appointment was not something she felt was particularly important in the scheme of long days, blood draws, and “chaos”.

Other women were able to remember some specific details about their genetic counseling experience. They described learning about their likelihood of cancer recurrence based on their genetic test results, potential increased risk for other cancers, discussing family history, testing implications for family members, and statistics. Melanie (60, BRCA negative, breast cancer at 42, ovarian cancer at 56) was grateful that she was provided with recommendations for how her daughter, granddaughter, and nieces could be monitored for breast and ovarian cancer. Gwen (53, BRCA1+, breast cancer at 45) who had testing in 2002, probably had the most specific recollection of most of the women, remembering that they spoke about increased cancer risk with BRCA carrier status, prophylactic surgeries, and other “statistics”.

Multiple women noted that the information was useful and interesting. However, most of the women were vague about the details of what occurred during their genetic counseling appointment. For some, this may have been because they were not sure how to fit the information that was being given to them into their vision of their life moving forward. April (39, BRCA negative, breast cancer 31) told me that she:

“[w]as fascinated with what she [genetic counselor] was saying, but so much of it was, there was no context for it really. I don’t have a box to put this in, I don’t have a filter to run this through to translate it into my words, or how to make it apply to my life or my body. I know that I was negative for BRCA1 and 2, and my risk was very low for ovarian cancer, especially without family history, but past that, for that half hour, 45 min or hour or I don’t even remember, whatever we were in there…I don’t know that I took much more out of it.”

\textsuperscript{10} Human epidermal growth factor receptor 2 (HER2) is a gene which plays a role in the development of breast cancer. More: https://www.breastcancer.org/symptoms/diagnosis/her2
April had genetic counseling in the midst of her diagnosis and treatment for breast cancer at 31 years old. For her, the most important information was that she was BRCA negative and her ovarian cancer risk was low. Beyond that, she was unsure how to “apply it to her life”. She acknowledged that while she appreciated all the information the genetic counselor told her, genetics is “super complicated, and I think it’s difficult to explain to the average person who doesn’t know much about it, and I don’t know much about science.” April, who has a master’s degree, is certainly not average when it comes to education attainment in the United States and yet she still felt like the topic, and the way it was explained, was much too complex. Or, it simply was not information she felt she needed to retain after receiving the news that she was not a BRCA carrier.

**Research question: Effect on women being labeled “high-risk” for cancer**

The fourth research objective was to investigate how being labeled “high-risk” for cancer affects women. This question was identified as an important area of study by the U.S. Preventative Services Task Force, which stated that “[w]hat happens after patients are identified as high-risk in clinical settings is unknown. The consequences of genetic testing for individuals and their relatives require more study” (Moyer 2014:276). Women in this study could only participate if they matched inclusion criteria that identified them as being at elevated breast and ovarian cancer risk. Because this is an identified area that needs more study, participants were asked an open-ended question to allow them to respond to what risk meant to them. Per the semistructured interview guide, this question was along the lines of: your provider may have suggested testing because you were considered to be high-risk – what does that mean to you? Is that something you think about today? How does that effect you?

**Conceptualization of risk**

Participants identified specific risk-reducing behaviors – such as limiting alcohol intake, eating healthy, watching their weight, and exercising – as ways that they felt they could personally reduce risk, whether that was preventing cancer, preventing another cancer, or preventing a recurrence and/or metastasis.

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11 In 2015, only 32.5% of adults 25 years or older had a Bachelor’s degree or more; only 12% had an advanced degree (Ryan and Bauman 2016).
of cancer. Women also reported that in their everyday life risk of cancer was not something they dwelled on, although cancer risk seemed to loom to varying degrees. I separated women’s responses to risk based on their genetic test outcome (positive versus negative) and their cancer diagnosis or status (no cancer versus ovarian cancer versus breast cancer) to see if there might be a difference in how these groups spoke about risk. Overwhelmingly, almost all women spoke about some amount of worry associated with their perceived risk of cancer. Only two women reported that they did not worry about cancer (Julie 71, BRCA1 positive, no cancer; Sissy, 62, BRCA negative, Lynch VUS, breast cancer at 49). I will discuss the two major themes – ways to mitigate risk and looming risk – below.

**Ways to mitigate risk**

Unlike some other diseases, breast and ovarian cancers can rarely, if ever, be traced back to a specific moment of “getting” the disease. Because there are so many factors and pathways that lead to cancer, the ability to control the risk factors that are associated with cancer may provide some level of comfort to people who believe they are at risk for developing cancer. Indeed, many of the women reported “healthy” behaviors they ascribe to. As Marjorie (56, BRCA negative, breast cancer at 56) put it, “I’ve taken a really strong effort at controlling the things I can control to prevent it. My exercise level, my weight levels, my diet, I’ve become just close to fanatical on all of those things because I know my risk is there.” Overall, ten women, almost a third, mentioned either healthy habits (exercise, diet, less alcohol, less stress, etc.) or staying on top of their annual exams and checkups as ways they felt they could control or mitigate risk.

While healthy habits were reported as ways to control risk, these habits did not always control worry. In fact, sometimes they could contribute to worry if done in excess. As Leigh (27, BRCA negative, no cancer) reported:

“I try not to do anything in excess, but I still think you should just live your life and not worry about that stuff. So I try not to think too much about trying to control it because if I did that I think I’d probably lose my mind, I’d rather just live my life and if I get anxious sometimes, if I think well, what happens if I do get diagnosed…those are usually passing moments, it’s not like an obsessive thought.
I think the main things are just doing my regular checkups and trying to remember to do my self-exams when I can and if I don’t not to beat myself up about it. But to also not be afraid to live my life and enjoy the things I enjoy. If I try to worry about everything then I’m not going to enjoy anything and then what’s the point of prolonging my life?”

The idea of balancing healthy habits with the enjoyment of life was repeated by Roxanne (61, BRCA1+, ovarian cancer at 52) who told me she doesn’t “overdo drinking and I watch my saturated fat intake, and I eat more fruits and vegetables.” But after a recurrence of her ovarian cancer, she thought, “I did all that for what? What was I missing? Was I really suffering? I thought hell, if I want a glass of wine, glass of beer, I’m gonna have it. I’m not gonna forgo it. I’m gonna live.” While these healthy habits are reported by many women, most of them are careful not to overdo it. In fact, women from both the BRCA positive and negative groups reported that they were careful to change their lifestyle too dramatically because they felt like the reason they got cancer was because of something genetic. Yvonne (49, BRCA2+, ovarian cancer at 46) stated that:

“I really haven’t changed my lifestyle, and I don’t take a lot of supplements because I’m just like, ya know, the reason I developed cancer is not because of my lifestyle or what I did or didn’t do, it’s because I have this gene. And I have no control over that so if you have no control over something there’s no point to fussing and worrying about it, in my opinion.”

Yvonne, who is a BRCA carrier, completely attributes her cancer to her BRCA status. Stacey (77, BRCA negative, ovarian cancer at 67), who is not a BRCA carrier, told me that she thinks that “all cancers are some kind of genetic mutation”, which for her was proven by her extremely healthy lifestyle wherein she still got cancer. Stacey, who was influenced by her grandmother’s “health nut” ways, described how she will not get x-rays at the dentist unless absolutely necessary, and how she has never dyed her hair for fear of the chemicals. This belief that all cancer is genetic may underlie some of the responses by women who were BRCA negative (above) who were frustrated at not having an answer for their cancer.
Looming risk

Much like the limbo BRCA negative women find themselves in after receiving their test results, the idea of risk for most of these women is a day-to-day assessment. While participants reported that it wasn’t something they typically dwell on, the associated risk of cancer consequences, was something that loomed, omnipresent. Gretta (42, BRCA negative, breast cancer at 38) told me she was doing everything she possibly could for her breast cancer now (meaning surveillance and checkups), but she’s not actively keeping an eye out for “new tests” (e.g. genetic, surveillance) because “it gets to be kind of a full time job managing my treatment now, even in remission and the things I’m trying to look out for, or sometimes even just something that comes across my newsfeed can cause me anxiety because it starts to make me worry again, or think about longevity.” New tests, or pieces of information that she sees on her newsfeed, could trigger anxiety and worry for Gretta. For others who had already been diagnosed with cancer, this anxiety and worry could be triggered by pains in their body. Gwen (53, BRCA1+, breast cancer at 45) described trying to not think about cancer, but getting worried when faced with an unusual bodily pain:

“Yes, I worry, I try to put it out of my mind, I try not to think about it. I think that in the back of your mind is always, is this gonna come back? I try not to live everyday thinking, you know, is it coming back? I can’t live like that. I choose not to. But when you get a little pain or you get a little whatever, it’s like oh my god what’s the matter?! So you can tend to be more scared. It’s always in the back of your head.”

Many women diagnosed with cancer spoke about looming anxiety associated with bodily pains and forgetfulness, and whether that might mean their cancer had returned (either as a totally new cancer or a recurrence of their previous cancer). For Diane (71, BRCA negative, no cancer), the trigger was approaching the age that her mother was diagnosed with cancer. Diane found out she was not a BRCA carrier so while genetic testing gave her some comfort, it didn’t totally erase her cancer anxieties. Joann’s solution to this worry is to try and remove herself from the medical sphere entirely. She told me:
“What do I do for the rest of my life, test myself? And if you have something, I’m afraid, like I have a pain, or I don’t feel right, what do I do? Oh, it’s cancer, it’s cancer, it’s cancer. So, I have personally chosen to do nothing. Now I’ve gone to the other extreme, I don’t go to the doctors at all. I’m not going to worry about it. I’m just tired.”

By removing doctors and blood tests, Joann hoped to remove the fear that she feels whenever she may “feel different”. While avoidance at this level was not voiced by many women, it is important to note that almost all the women, regardless of BRCA or cancer status, described risk in relation to cancer – whether it would be a new cancer or a recurrence. Triggers would most commonly come from the woman’s own body and something not feeling right.

Summary of findings

This chapter presented the results for two research objectives. The first was to understand and present women’s experience of genetic testing and counseling. Through thematic analysis, it was found that family, treatment, surveillance, information/research, and Ashkenazi Jewish heritage were the main motivations for testing, as well as how women planned to use their genetic test results. Multiple themes were often present in one interview, and the qualitative data regarding these themes provides insight into how women hope to use genetic test results. It is apparent that the women in this study comprehend the uses of genetic tests and have their own ideas about how to use their results, especially around treatment and surveillance options. However, treatment and surveillance, as spoken about by the participants, are conceptually different than how they have been defined in the medical realm. Specifically, prophylactic surgeries are tied up with notions of treatment. Medically speaking, these are two separate concepts, yet women spoke about them interchangeably. Surveillance for other primary cancers or cancer recurrences was also an important topic and is another area where women’s conceptualization of surveillance may differ slightly from medical professionals. Rather than focusing solely on specific screening mechanisms that would catch cancer at an early stage, women reported that they hoped genetic testing would somehow allow them to be vigilant for other cancers, for themselves and their family members. It is noteworthy that women in the
breast cancer group spoke about surveillance at double the frequency of the ovarian cancer group (63.6% vs 31.3%).

Thematic analysis provided data for women’s motivations and aspirations for genetic testing, while reactions to receiving genetic test results uncovered the ways in which women may (or may not) have been prepared for their results. These reactions depended on who their ordering provider was (GP or NGP), and whether they received positive or negative test results. Of the women who had genetic testing in the overall sample (n=31, 93.9%), slightly over half saw a GP (17, 54.8%). Women who received a positive test result through a NGP (5, 16.1%) reported negative emotions – shock, anger, fear. In contrast, women who received a positive test result through a GP (6, 19.3%) largely reported ambivalence at their results, saying it was just confirming something they already knew. Negative results drew similar responses regardless of if a GP was seen or not. The participants in this group described being relieved and even thrilled about not being BRCA carriers, and yet an undercurrent of ambivalence and questioning was present. While they received a negative test result, there remained lingering questions of why they developed cancer.

Undergoing genetic testing and undergoing genetic counseling are two very distinct experiences. 54.8% of this sample had genetic testing with a GP, leaving 38.7% having testing through other providers such as OBGYNs, surgeons, and oncologists. Two participants had testing twice – once with a NGP, and then with a GP. Joyce, a participant who experienced testing initially with an oncologist and then later with her daughter through a genetic counselor, provided a vivid recollection of what occurred during both appointments, contrasting her experiences. Joyce’s vignette was used as a proxy for the data presented for those that had genetic counseling versus those that did not. Her initial testing appointment with her oncologist did not include any discussion of psychosocial impacts, of how the results may impact her medical treatment or surveillance, or any of the other boxes that are supposed to be ticked per genetic counseling guidelines before ordering genetic testing. Joyce recounted that after accompanying her daughter to her genetic counseling appointment, she realized that the emotional and educational aspects of the testing process had not been provided to her. Women who had testing with a NGP reported not understanding what BRCA was, being left with brochures and informational material to read about genetic testing, not expecting to be
positive (but then being positive and feeling unprepared), and even having blood samples taken during surgery without realizing testing would be done. Women who had genetic testing with a GP did not report situations like the NGP group experienced. Rather, this group reported a variety of perspectives: that it was simply a medical procedure to complete, that it was call-centerish and impersonal, that it was unnecessary, that they did not understand how genetics works, that it was no big deal, that it was interesting. The women in this group also reported specific topics that they covered during their appointments: statistics, family history, increased cancer risks, and repercussions of testing for the patient and their family members; these were specifics that were not brought up by women in the NGP group.

The final section addressed the research objective which aims to understand how women make sense of being labeled ‘high-risk’ for cancer. Inclusion criteria for this study is for women who are at an elevated risk of breast and ovarian cancer, and an open-ended question was asked in order to elicit their conceptualizations of risk broadly. When discussing risk, almost all (29, 93.5%) of the responses included varying levels of worry about cancer. In worrying about cancer, there were two main themes: that risk was constantly looming, and they ways in which cancer risk might be mitigated. In order to moderate perceived cancer risks, women in this study reported adhering to “healthy habits”, focusing on things like their diet, exercise, alcohol intake, and stress, as well as attending to annual exams and checkups. In women’s responses to this question, it was apparent that risk was a day-to-day and ultimately embodied experience which was closely tied to worry and anxiety. Anxiety about cancer risk could be triggered by pains in the body, social media posts that would trigger anxiety, medical tests, or spats of forgetfulness.
CHAPTER FIVE:
DISCUSSION OF FINDINGS

Overview of study findings

The purpose of this study was twofold: to determine women’s views and experiences of genetic testing and counseling (GT/C) and to determine how being labeled “high-risk” for cancer affects women. At first glance, this objective sounds extremely straightforward. However, given the semistructured interview format (which allowed for talk that could sometimes meander to topics I did not foresee) and inductive coding (which allowed themes to emerge), there is now a wealth of knowledge regarding women’s experiences and perceptions of GT/C. Significantly, this study has some important outcomes. First, it adds to an area that has been previously neglected: that of women and their testing experiences with non-genetics professionals (NGPs). Second, given the data of the results of women’s reactions to their test results, I believe there is another demonstrated difference in undergoing testing with a genetics professional (GP) versus a NGP. This chapter will discuss these findings in light of existing scholarship. Additionally, it will consider contributions to theory, specifically midrange theory within a feminist and critical medical anthropology scope. Finally, I hope this study may help guide GPs in their interactions with their patients, as a way to truly tailor the GT/C experience in a way that is helpful and practical for the patient.

Thematic analysis

Through the process of semi-structured interviews with participants, followed by inductive coding, I was able to allow important themes to emerge in the context of GT/C, rather than providing pre-conceived boxes for participants to tick. Themes discussed, in order of overall frequency, were family (81.3%), treatment (62.5%), surveillance (43.8%), information/research (40.6%), and Ashkenazi Jewish ancestry (18.8%). Since these topics represent frequencies, they represent important topics that this sample population considered
when deciding to pursue and undergo genetic testing. Quite often, women explicitly stated that they had specific objectives for their genetic test results.

In terms of previous research, the themes I discovered over the course of my interviews are not necessarily new or unstudied by those who have been interested in the motivations of women undergoing genetic testing. An early study by Clark et al. (2000) asked participants to rank their reasons for and against having genetic testing. The top five reasons for why genetic testing was “somewhat or very important” to that cohort slightly reflects my own results: (1) aid in cancer research (98%); (2) information for others in the family besides children (92%); (3) offer of free testing (91%); (4) information for children or future children (86%); and (5) protect health through screening/prevention (85%) (Clark et al. 2000:228). Family was such an important reason for women to undergo testing in this cohort that it shows up twice in the top five reasons.

More recent research by Hesse-Biber (2018) likewise found that family risk, including children’s risk and to tell family, was a primary motivation for testing. Considering the hereditary implications of genetic mutations, and the sometimes familial nature of multiple cancer diagnosis witnessed in one family, this finding should not be surprising, and has been found elsewhere (Dancyger et al. 2010, Etchegary et al. 2009, Foster et al. 2002, Garg, Vogelgesang, and Kelly 2016) and in systematic reviews (Sweeny et al. 2014). Foster et al. (2002) likewise found that women felt a need to take responsibility for their own health through prevention or screening, as well as a responsibility towards family members and children, when choosing to undergo genetic testing. This idea of “genetic responsibility” to family has been thoroughly studied in general (Hallowell 1999, d'Agincourt-Canning 2006, Etchegary et al. 2009, Foster et al. 2002) and among Ashkenazi Jewish women specifically (Mozersky 2012). Other research also supports the importance of family in this context, reporting that some family members even felt obligated to undergo testing in order to benefit their family, rather than to the benefit of themselves personally (Dancyger et al. 2010, d'Agincourt-Canning 2006). While a few respondents did say that they underwent testing solely to help their family members, they were in the minority. Most of the participants elicited other reasons for genetic testing.

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12 In the context of genetic testing for hereditary cancer syndromes. While testing for BRCA1/2 became available in the mid-1990s, Cho et al. (1999) described testing as “controversial among clinicians, patients, ethicists, and policy-makers” in 1999 (pg. 158).
Treatment was the next most frequently mentioned theme, with 20 participants (62.5%) wanting to have genetic testing information due to the influence it could have on treatment (72.7% in breast cancer, 62.5% in ovarian cancer, and 40% in no cancer group). When women spoke of treatment, they brought up how they hoped their genetic test results could help inform current treatment, such as surgeries and types of chemotherapy, potential future treatment of cancers (either a recurrence or new diagnosis), and prevention of future cancers through prophylactic surgeries. Although treatment is defined as attacking a current disease and not future ones (prevention), I grouped prevention (i.e. the idea of prevention, prophylactic surgeries) with treatment because they were very often spoken of in the same sentence. A recent study assessing patients’ views of treatment-focused genetic testing (TFGT), which is the “mainstreaming of genetics into cancer care” similarly found that women expressed hoping to use their genetic test results to prevent future cancers, both in the patients themselves and among their family members (Wright et al. 2018). From this same study, I will quote an extensive portion of their discussion because there are multiple points I wish to address. Wright and her co-authors state that:

“We observed in participants’ accounts a common lack of appreciation of the treatment function of TFGT, with breast cancer patients rarely understanding the treatment role of the test and ovarian cancer patients appearing to be completely unaware of this. Thus, when it came to the issue of treatment – the element of TFGT that distinguishes it from other forms of germline BRCA testing – there was a general lack of awareness and understanding in participants’ reflections. Not only was immediate treatment of current cancer rarely expressed as a motivator for undergoing TFGT, but in the minority of instance where treatment was understood, it was ambiguously described, and almost indistinguishable from individual’s hopes for prevention.” (Wright et al. 2018:1468, emphasis in original).

First, it seems that if the point of the paper, as described in the title (Patient Views of Treatment-Focused Genetic Testing) is to assess patients’ views of TFGT and you refer patients to clinical genetics for pre-test counseling, then whoever is doing the counseling did not do a good job of describing how women could use this information in their treatment pathway. This is entirely on the medical practitioners to explain. In fact,
the authors explain that when introducing TFGT to patients in the gynecology unit during their “new patient” appointment, “there is little discussion of familial implications, or treatment implications” unless the tests come back as a positive or VUS. In the breast unit, they also describe that it “varies” how much patients are informed about “the possible implications for TFGT for surgery, adjuvant therapy, and family” (Wright et al. 2018:1463). Given this information, of course patients “lack an appreciation of the treatment function of TFGT”…for some, it simply was never explained to them. For those that were provided an explanation of TFGT, the very thing they were supposed to have “views” on, was it explained in a way that made sense? There is copious literature and academic vigor assessing patient-provider communication, with much of it pointing out that providers aren’t actually speaking in a way that makes sense to patients (for genetic counseling specific examples see: Joseph et al. 2017, Rapp 1988, Hunt, de Voogd, and Castañeda 2005). In work conducted by Browner et al. (2003) with Mexican-origin pre-natal patients and genetic counselors, they found that, “[M]iscommunication is often the result of asymmetric communication…the best way for counselors to improve comprehension and win a hearing for their message is to give a better hearing to the words of their clients” (pg. 1945). In the case of Wright and colleagues, it may not have even been miscommunication, but the complete lack thereof. Additionally, other work with cancer patients and treatment decisions have found that while patients buy into the role of patient as customer/partner in decision making, they also can resort to delegation of information, reframing, deflecting, or outright rejection of responsibility for these decisions (Sinding et al. 2010). The authors of this study note that past scholarship “on treatment decision making typically characterizes people who prefer not to take an active role as overwhelmed, misinformed about the nature of treatment decisions, or more generally lacking capacity to participate” (Sinding et al. 2010:1099), which completely echoes the findings by Wright et al. (2018).

I find this patronizing tone of “patients just not understanding treatment”, and therefore wrong or lazy, an old paradigm that places blame on patients rather than as a constructive critique. Blaming patients has taken on various forms in biomedicine over the years. It has been an argument for “frivolous, entirely avoidable, or irresponsible” emergency room visits (Oliver 2015), in doctor’s views of obese patients as “unmotivated and non-compliant” (Teixeira, Pais-Ribeiro, and Maia 2015:218), in nurses attitudes towards
pain management (Holley et al. 2005), and as the “cause” of homelessness (Lyon-Callo 2000). Additionally, the idea that patients do not understand treatment contrasts with the conversations I had with participants who were active agents who have a good grasp on how they would and could use their genetic test results for treatment options. Participants described using their test results to advocate for specific surgeries and discussed genetic test results’ impacts on drugs such as chemotherapy and PARP inhibitors. There was a recognition that there could be a difference in how you treat someone that carries a BRCA mutation versus one that did not. True, ambiguousness exists about what those treatment pathways might be (and was in fact recognized by several women), but who would expect patients to be intimately knowledgeable about all the possible treatment options, when it is in fact so very complicated? Sinding et al. (2010) likewise found that at a certain point, some women decided to opt-out of making medical decisions by relegating that responsibility to a caregiver, or else by assigning treatment decision making responsibility to a collective one (based on doctor’s experience and deciding to trust healthcare practitioners) (pg. 1096). Disengaging from the treatment decision making process, then, is not necessarily a sign of an unengaged or uniformed patient, but rather shows trust in the knowledge and experience of healthcare professionals (Swainston et al. 2012, Mendick et al. 2010).

The third most frequent theme was discussion of surveillance (43.8%) as a way to use their genetic test results – particularly around the idea of “keeping watch” for other cancers and/or preventing them outright. The way that women spoke about surveillance contrasts with surveillance in medical usage, which seeks to detect cancer at earlier stages to have better treatment outcomes. This is done using various screening techniques such as breast exams, mammograms, and CA-125. Some specific surveillance techniques were mentioned, but more often than not surveillance was spoken about as in more ambiguous terms, as simply understanding your “increased chances” of developing certain cancers as a result of genetic testing (Gwen, 53, BRCA1+, breast cancer at 45). While surveillance was the third most frequent theme discussed, it was much more frequent in the breast cancer group versus the ovarian cancer group (63.6% breast cancer, 31.3% ovarian cancer, 40% no cancer), and women in the both of the cancer groups reported specific cancers they wanted to watch out for – ovarian, colon, pancreatic. In contrast, two women in the no cancer group did
speak about specific screenings and when/how often they should occur, but only spoke about one cancer (breast).

The fourth most frequent theme centered around using genetic test results for more information and research, with 40.6% overall frequency (45.5% breast, 43.8% ovarian, 20% no cancer). Women in this study spoke about genetic testing as a “good” thing, which could potentially solve medical mysteries and empower them with knowledge. These findings do not support the idea that women feel “constrained” after undergoing genetic testing, as found by (Hallowell 1999). While Hallowell argued that women may feel constrained based on their responsibility to help determine genetic risk for others, women in this sample spoke about how their genetic information could be used for not only family but for other people in similar situations. There exists an idea that knowledge is good, even if it will not help immediately or even personally. What women do feel constrained by, I would argue, is when test results come back as negative, and they are left questioning why they developed cancer (more below).

Helping with research was found to be the top reason to undergo testing by Clark et al. (2000). While research was not the number one reason women in my study cited for undergoing genetic testing, important themes of information and research remain. Namely, Clark et al. found that an “unexpected outcome of the study [was] that 70% of participants thought the researchers wanted them to have testing” (Clark et al. 2000:233).

The above discussed themes are the why women in this study decided to pursue genetic testing, as well as the how they planned to use the genetic information. Some of these reasons and uses are not surprising, and have been observed in the literature, while other may provide a bit more nuance that cannot be captured in quantitative research. In adding to the this literature, I am encouraged by a statement by Sweeny et al. (2014) in their systematic review and critique of the literature of predictors of genetic testing decisions. They state that:

“…people’s self-generated explanations for testing (and not testing) are far more consistent than the findings from quantitative attempts to predict testing decisions. Qualitative studies may not provide
the precision or statistical conclusiveness desired by many researchers, but our initial overview of the literature revealed the value in simply asking people why they made the decision they made.

Considering the potential for interventions to increase interest in testing (to the extent that testing is beneficial in a particular context or for a particular person), the existing quantitative findings provide only a few appropriate targets (i.e., predictors that have received largely consistent support) for interventions. In contrast, qualitative studies paint a clear picture of how best to promote effective, informed and value-based decision making about genetic testing” (Sweeny et al. 2014:282).

This information is particularly important for medical providers – both genetic and non – to help recognize which areas are salient for women undergoing genetic testing (namely family, surveillance issues, and treatment) rather than traditionally measured outcomes such as knowledge, quality of life, anxiety/distress, and satisfaction (Madlensky et al. 2017, Meiser and Halliday 2002, Oberguggenberger et al. 2016). Knowing that these may be important themes (among others, see Roshanai et al. (2012), providers should seek to include this information when discussing why genetic testing can be important, and how it can be used specifically.

**Fluidity of women’s experiences**

There will always be complications when attempting to put lived experiences into discreet boxes as above. One of these complications arose when participants spoke about prevention of cancer, which was spoken in the same breath as ideas of treatment and surveillance. As stated before, when participants spoke of treatment uses of genetic testing, they also spoke about prophylactic surgeries. When we spoke about surveillance uses of genetic testing, it was often in terms of being vigilant for other cancers in order to prevent them. I considered coding prevention into a separate theme, but prevent* (-ion, ed, etc.) only showed up five times in the entire corpus of interviews. Prevention, then, is intimately intertwined and embedded with ideas of both surveillance and treatment, a place where, biomedically speaking, it does not necessarily belong.
This is an important concept to understand in its complexity, as research around prevention acceptability addresses specific methods of prevention (e.g., chemoprevention, prophylactic surgeries) rather than women’s experience and understanding in greater context (Julian-Reynier et al. 2001, Koch and Svendsen 2005). This is what a systematic review of women’s decision making regarding risk-reducing strategies suggested, explicitly calling for more qualitative and longitudinal work to be done around the subject (Howard, Balneaves, and Bottorff 2009). Longitudinal work on the subject is particularly important considering women may wait up to nine years after they receive their results to undergo risk-reducing mastectomy or oophorectomy (Howard et al. 2010). Additionally, understanding that patients/women conceptualize these applications differently than providers is imperative. While the literature on patient-provider communication is vast, work to address miscommunication or misidentification of concepts cannot begin unless the disparity is uncovered.

Women’s overall experiences – what’s genetic counseling got to do with it?

A strength of this study was in gathering qualitative recollections of women who did not have GT/C at a genetics clinic or with a genetics professional (GP), which allowed for greater understanding of how women remember the genetic testing experiences with non-genetics professionals (NGPs) and GPs alike. As a group, women who had testing through a NGP are potentially hard to reach. Past research has relied on convenience sampling of individuals being seen in a genetics clinic, by reaching out to NGPs and genetic counselors directly (Vadaparampil et al. 2015, Bensend, Veach, and Niendorf 2014, Douma, Smets, and Allain 2016) or through assessment of providers’ knowledge through systematic reviews (Hamilton et al. 2017).

Several studies have identified numerous areas of concern when it comes to NGPs understanding appropriate screening for and ordering of genetic testing (Hamilton et al. 2017, Wideroff et al. 2005, Tan and Fitzgerald 2014, Klitzman et al. 2013, Trivers et al. 2011). In terms of actual genetic counseling, Cragun et al. (2015) found a difference between people who saw a GP and NGP in their pre-test discussion and recall of GC elements, specifically drawing of a pedigree, discussion of laws against genetic discrimination by health insurers, discussion of issues relating to life and/or disability insurance, and having a summary letter of the
appointment (Cragun et al. 2015:4). Another study, which assessed all BRCA testing providers in Florida, the majority of whom did not have formal training in genetics, found several knowledge gaps. Importantly, it was found that the majority of these providers lacked knowledge relating to comprehensive rearrangement testing after a negative result, as well as appropriate family testing following a VUS result (Pal et al. 2013). I very much support the notion that genetic counseling, as a complex and somewhat long process, should be done by trained professionals both pre- and post-test (Christinat and Pagani 2013).

There has been some research that has looked at self-reported pre-test GC practices from NGPs by Vadaparampil et al. (2015). In their sample of 81 providers, they noted that the “lack of adherences to clinical guidelines in the delivery of genetics services among this sample is concerning” (p. 475). Based on the responses from women I spoke to, I would say “concerning” is an understatement. In this study, participants reported having blood drawn during surgeries without any discussion of the test whatsoever. Participants also reported being left with brochures and other packets of information in lieu of a discussion of the testing implications. In the sample reported by Vadaparampil et al. (2015), 61% of providers reported always discussing implications of test results for family members, and 64% said they discuss the benefits and limitations of risk management options. Less than half (49%) discussed psychological impacts of testing. While my sample is small (12 who saw a NGP, 2 who initially saw a NGP), their descriptions regarding their genetic testing experiences are striking (see Appendix C10). Women reported being unsure about what BRCA is, being left with brochures in lieu of discussions, not feeling prepared for their results, only covering family history in their pre-test discussion, having blood drawn for genetic testing during surgery, and not knowing if they were testing for genes other than BRCA. Bensend, Veach, and Niendorf (2014) noted that empirical evidence of negative outcomes as a result of seeing NGPs for genetics services was limited, but these possible negative outcomes could include psychosocial effects, testing and screening errors, unnecessary prophylactic surgeries, advancement of disease, and unnecessary use of resources (Bensend, Veach, and Niendorf 2014:59). After speaking to genetic counselors in the state of Minnesota, Bensend and colleagues found that these negative outcomes were preceded by “lack of counseling about genetic test or screening results; failure to provide a thorough explanation of the genetic testing being considered, including all the components
necessary to obtain informed consent such as the risks, benefits, and limitations of testing; failure to present all of the testing or screening options in an unbiased manner; insufficient education regarding a genetic condition and the way in which it is inherited; and lack of articulation of risks for family members” (Bensend, Veach, and Niendorf 2014:60). I do not know if specific negative outcomes that Bensend, Veach, and Niendorf (2014) detail such as testing errors or unnecessary prophylactic surgeries came to pass in my sample, but there is a definite lack of many of the suggested counseling guidelines from the women I spoke to, at least in their recall of the testing events. I would, however, argue that adverse psychosocial outcomes are present in my data when considering the reactions women reported to their test results. Women with positive results that did not see a GP overwhelming reported negative reactions to their results, while positive women who saw a GP were much more likely to report some ambivalence to their results (Table 9). This is similar to results found by Bonadona et al. (2002) who found that 52% of women in their population, who saw a geneticist, felt reassured after receiving their positive genetic test results. Given women’s recollections and descriptions of their testing experience, paired with their reported reactions to test results, it is clear that NGPs are not properly preparing women to receive positive genetic test results. Fully addressing the psychosocial aspects of genetic testing is important in all cases, but especially for women who were recently diagnosed with cancer, as van Roosmalen et al. (2004) found that reported well-being is lowest for this group (in relation to unaffected [no cancer] women).

Table 9. Women’s reactions to genetic test results grouped by provider type and test result

<table>
<thead>
<tr>
<th></th>
<th>Positive</th>
<th>Negative/VUS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>NGP</strong></td>
<td>shocked, scared, knocked the wind out of me,</td>
<td>relieved, thrilled, freaked out,</td>
</tr>
<tr>
<td></td>
<td>angry</td>
<td>surprised, mixed feelings, stressed</td>
</tr>
<tr>
<td><strong>GP</strong></td>
<td>upset, good, not surprised, confirmation</td>
<td>elated, relieved, ambivalent, no big</td>
</tr>
<tr>
<td></td>
<td></td>
<td>deal, disappointed</td>
</tr>
</tbody>
</table>

Another important outcome of the data is how women in both GP and NGP groups who received negative/VUS test results reported a wide range of emotions. While the women in the positive group were
fairly homogenous in their responses, women in the negative/VUS group reported numerous emotional reactions, sometimes multiple feelings at once. Early work by Lodder et al. (2001) concluded that for “most non-mutation carriers, psychological follow-up might be of lesser importance” (Lodder et al. 2001:15), perhaps because non-mutation carriers have been found to have less distress than carriers (Lynch et al. 2006). Based on my results, I do not agree that those who receive negative results should not be considered equally (psychosocially) as those who receive positive results. It appears that some women, when presented with a negative genetic test result, felt that they did get the “answer” for their cancer diagnosis. This may reflect a genetic deterministic view that “genes determine health” (Parrott et al. 2012:763). Importantly, women searching for an answer for their cancer existed in both the GP and NGP groups, indicating that genetic counseling may not be doing enough to address ideas of genetic determinism and disease essentialism. In their critique of contemporary public discourse around genetics, Nelkin and Lindee (1995) write that “the images and narratives of the gene in popular culture reflect and convey a message we will call genetic essentialism. Genetic essentialism reduces the self to a molecular entity, equating human beings, in all their social, historical, and moral complexity, with their genes” (Nelkin and Lindee 1995:2). For example, rather than attributing obesity to a complex relationship to between genes and environment, media, such as cartoons, show a researcher isolating the “obesity gene”. In a biomedical system that understands disease as a malfunction of biology, genes may be viewed simplistically as the ultimate cause (Lippman 1992). That seems to be the case for many of the women who I spoke to who presented mixed feelings about their negative results (Table 10).
Table 10. Reports of mixed feelings among women with negative test results

<table>
<thead>
<tr>
<th>Quote</th>
<th>Participant</th>
<th>Age</th>
<th>Cancer Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>“It was really weird to hear back that no, I didn’t have those markers…So I don’t know if you could say I was disappointed, but I was surprised, I was really surprised.”</td>
<td>Kristina</td>
<td>60</td>
<td>ovarian cancer at 21, breast cancer at 55</td>
</tr>
<tr>
<td>“I had mixed feelings about the results and that probably sounds crazy…there’s a little bit of frustration in not being able to understand how this happened [breast cancer diagnosis].”</td>
<td>Marjorie</td>
<td>56</td>
<td>breast cancer at 56</td>
</tr>
<tr>
<td>“I was also disappointed which sounds strange, because I felt there were more research on BRCA positive patients and there would be more trials for me.”</td>
<td>Angie</td>
<td>70</td>
<td>ovarian cancer at 66</td>
</tr>
<tr>
<td>“I was kind of hoping that they were gonna find…something? A reason? Because it’s all a mystery for me, at least, with my sub-class [triple negative].”</td>
<td>Katherine</td>
<td>49</td>
<td>breast cancer at 35</td>
</tr>
<tr>
<td>“There was definitely some sense of relief and there was a little bit of, huh, like a little disappointment, I’m surprised, I thought that was gonna help us there, right?”</td>
<td>Clara</td>
<td>57</td>
<td>ovarian cancer at 54</td>
</tr>
</tbody>
</table>

I believe these “mixed feelings” are a vital outcome of qualitative work that can be potentially overlooked or misunderstood by quantitative measures and scales. For instance, Mella et al. (2017) described “contradictory” findings, with high levels of anxiety, depression, anger-hostility, and fatigue-inertia and confidence and serenity in their sample of women’s emotional reactions to BRCA genetic test results. They explain the existence of outwardly diametrically opposed emotions on “instruments with different sensitivity” and “emotional constraint due to having started a genetic counseling process”, with no mention that it might simply be normal for people to exhibit multiple emotions simultaneously (Mella et al. 2017:6). Rather than measuring women on discreet emotional dimensions, this qualitative work allows a more nuanced view of a traditionally overlooked group in genetics research. I would argue for more work to be done to address the liminality that women with a cancer diagnosis, who also received a negative result, may be feeling.

Which is not even addressing the other critiques of using quantitative measures and scales in research. For just one example, an important scale which is used in many studies to measure anxiety and distress, the Hospital Anxiety and Depression Scale (HADS), has “numerous structural, conceptual, and psychometric problems” (Coyne and van Sonderen 2012:77).
Given the present and extant research, I am an advocate for true genetic counseling\textsuperscript{14} to take place both before and after genetic testing for cancer. Some of the experiences of genetic testing by NGPs that were relayed to me were, at best, procedural and at worst, unethical. However, I also want to reiterate the point that women who had genetic counseling with a GP sometimes had a hard time recalling events of that appointment. During interviews, I had to probe on numerous fronts, during almost every interview, to gain more insight into the appointment proceedings – did you talk about family? Did you talk about insurance? Did you talk about repercussions of the test? Several of these questions were directed at components which are considered “imperative” elements to include in a counseling session, for instance (Berliner and Fay 2007:250):

1. Details of the genetics of cancer in general
2. The medical and family history of the client and the specific syndromes being considered (if any)
3. The likelihood of a mutation being present in the family
4. Possible test outcomes and the implications of these outcomes
5. Medical management options

The data presented of women’s experiences with genetic counseling with GPs (see Appendix C11) makes apparent how little is remembered from the appointment(s). This finding may be supported by work from Ellington et al. (2011), who analyzed counselor-patient communication and found a lack of in-depth cognitive processing and understanding on the end of the patients. Of course, some women remember specifics such as notions of increased risks for certain cancers and some statistics, but most of the recollections can be tied back to the thematic analysis that that was covered previously. Understanding how genetics and DNA works was never mentioned as important (in fact, it was listed as something that was confusing and not useful) yet has been observed as an educational tactic in observation of counseling sessions by the researcher.

Recognizing and understanding what genetic counseling practices can be changed, updated, or modified, is an important area of research, and one that GPs have been encouraged to take part in (Biesecker 2018). Riley et

\textsuperscript{14} When I say “true”, I mean for counseling to take place with someone who has genetics training, preferably a certified genetic counselor.
al. (2012) identified a multitude of “essential elements” of genetic counseling for cancer, some of which were never brought up by my informants. These elements were potentially never discussed. Or else, they simply were not remembered, which indicates they were not useful to the participant at the time of counseling. Figuring out exactly what should be covered, so as to not overburden the patient, should be of utmost importance to GPs and can be accomplished through more robust, personalized sessions. Research conducted in Germany with women about 3.5 years after their initial genetic consultation likewise recommended that more emphasis and time should be put into making the counseling session more personalized. One way to do this is to provide the information on DNA and genetics, typically covered during the pre-test counseling appointment, before the appointment in email, paper, or web-based platforms (Eismann et al. 2016).

Risk

“Everywhere – while reading the science pages in my daily newspaper, taking in health tips at my local health food store or during a routine visit to the gynaecologist – I run the risk of being saddled with a risk” (Samerski 2006:198)

As Samerski underscores in the above quote, risk (or the risk of risk) appears to be omnipresent in today’s society, which, it may be argued, is a consequence of existing in late modernity (Beck 1992, Giddens 1991). Some risks, like those associated with genetic testing, cannot be uncovered or known unless through technological intervention – in this case, putting the person genetically at risk (Novas and Rose 2000). Yet risk is not an objective measure, it is simultaneously scientifically and socially constructed, and it must be considered within its social construction (Beck 1992). It is also an embodied concept “connected to…class positions, sense of vulnerability, and social resources” (Castañeda and Mulligan 2017:15). This co-construction of risk, along with the individual’s values and world views, must all be considered (Nelkin 2003). This is elegantly showcased in research by (Hashiloni-Dolev 2006), who juxtaposes how German and Israeli genetic counselors’ notions of risks associated with sex chromosome anomalies are mediated by their own culture’s understanding of what constitutes normal and pathological. In this case, broader cultural ideas about the importance of fertility impacted how the counselors described sex chromosome anomalies (normal versus
pathological) and thus impacted whether the counselors stressed potential risks (as they did in Israel) or downplayed them (as they did in Germany). For these reasons, risk must be understood in its cultural context, and “cannot be considered a neutral term” (Lupton 1993:425). When I asked women about risk, their answers very much reflect how individuals in our society are supposed to think and react to risk. This presented a potential mismatch of risk communication that may take place during GT/C, which may focus exaggerated perceptions of personal cancer risks (Croyle and Lerman 1999).

When asked about risk, participants spoke mostly about how they felt it was looming sword of Damocles, and that they attempted to mitigate risk through individual behaviors. As stated before, risk seems to be everywhere. As Douglas and Wildavsky (1983) stated, “[W]hat are Americans afraid of? Nothing much, really, except the food they eat, the water they drink, the air they breathe, the land they live on, and the energy they use” (Douglas and Wildavsky 1983:10). Risk was spoken of as something that most participants attempted to not think about all the time, but medical tests, something on the news, forgetfulness, or body aches could trigger anxiety about cancer. This anxiety was tied to the assumption that there might be a cancer reoccurrence, or a new primary cancer diagnosis. As Lupton (1993) states, the “discourse of risk is weighted toward disaster and anxiety rather than peace of mind” (Lupton 1993:433), which most certainly occurred in the lives of participants.

The other angle presented by participants of risk was that of the individual controlling risk through specific healthy behaviors that one could control – exercise, weight, diet, alcohol, stress. This idea of controlling your risk for cancer through lifestyle choices through “health-enhancing behaviors” is explicitly stated as a goal of genetic counseling for common diseases such as cancer (Biesecker 2001:329) and is copacetic with the larger public health discourse (Lupton 1993). The flipside of knowing your risk, and thus knowing how to control of one’s behaviors, is that even though it is your choice to engage in healthy behaviors, to not engage in those behaviors is not really a choice as seen through the eyes of a risk adverse society. Women in this study struggled with that idea, attempting to balance the right amount of healthy habits (“I try not to do anything in excess”), but also sometimes just saying to hell with it (“if I want a glass of wine, glass of beer, I’m gonna have it”). This habitus, as thought about in regards to contemporary lifestyle
theorizing, presents an interesting nexus of individual agent lifestyle choice against/in midst of the larger structure of cultural influences and expectations of how to stay healthy (Hinote 2015). Leontini (2010) likewise points out in her work with women undergoing genetic testing for Huntington’s Disease that an idea of “care of the self” arises during the genetic counseling encounter. This notion importantly binds the individual to their own self-care and to that of others, who would benefit from the care the individual can then provide. While this notion was not explicitly stated by participants in this study, family was obviously an important theme during interviews, and the literature on women’s obligations to the health of their family and “duty to disclose” genetic test results is vast (d’Agincourt-Canning and Baird 2006, Gibbon 2006).

And yet, while almost a third of the women in my sample spoke about individual healthy behaviors they engaged in, the idea that cancer has a solely/primarily genetic explanation persists. As discussed earlier, women with negative results were confused or disappointed in their test results; they wanted an answer for their cancer diagnosis, which they felt they would get through genetic testing. And while some women mentioned their healthy lifestyle choices they opted in, others specifically mentioned that they did not change their lifestyle at all because they had a genetic mutation. Women in both groups, negative and positive, mentioned lifestyle behaviors to control risk.

Implications for future research

First, I would like to use this data to help recommend updates to the way that genetic counseling is conducted (more in Chapter 6), which can take place through individual GPs or through changes to masters’ programs teaching curricula. Obviously, I am an advocate for GPs and counselors. Just like any other discipline or profession, however, I think there are ways in which to improve. While this study did not directly observe genetic counselors in action, what women remember about their encounter is telling. Further, it is important that reflexive research continues to be conducted, as women’s motivations and desires for genetic testing will change over time. Based on this data, however, I think there are some concrete applications. For one, I question how much background on genetics is given to patients. Based on personal experience and observations of GC appointments, along with comments from participants in this study, it
does not seem to be a priority for most patients. Rather than spending the time on how genetics works, I would like to see a true conversation take place between GPs and patients. The NSGC website tells interested parties to “[T]hink of genetic counseling as a conversation. Your input – your goals, your feelings, your concerns – are all very important” (National Society of Genetic Counselors n.d.). What makes a conversation successful is making sure both parties are prepared. Is there more that can be done to prepare women for the genetic counseling experience beforehand, especially in the midst of a cancer diagnosis? Once the counseling session is underway, how can we make sure it is an actual conversation and not a one-sided educational lecture? From this data, it is apparent that women have ideas of how they hope to use their genetic information. Ensuring that these points are addressed and interrogated with each patient, is vital. I am not going to change genetic counseling practices by myself, but I hope to be in dialogue with those that could affect change.

Exploratory research indubitably discovers further avenues of research, and this study is no different. First, the current research can be refined and explored via different avenues and the triangulation of observed genetic counseling practices, feedback from patients, and feedback from practitioners/genetic counselors. Rather than focusing on one time-point, this work should focus longitudinally to see how women remember their experience, incorporate (or not) the information, and then be used to improve genetic counseling practice. NGPs should also be a focus of research in order to truly capture their impact on patients and to evaluate their genetic practices. New avenues of research include an examination of gender within biomedicine, as well as close qualitative research with ovarian cancer patients. During the course of interviews with ovarian cancer patients and survivors, it was apparent that participants felt more needed to be done for their disease – more awareness on the part of practitioners and women along with better screening and diagnostic capabilities. It was also among this group that I was told some frankly horrifying stories about providers during diagnosis and treatment of ovarian cancer. These stories were much more common in the ovarian cancer group and suggests that more work needs to be done to understand this deadly disease within the context of biomedical hegemony, patriarchy, and power. It was apparent that women diagnosed with ovarian cancer were passionate about their plight. At the end of every interview, I always asked if there was
anything else they wanted to share with me, either to help clarify something they said earlier, or something they felt would be important for other people to know about their situation. While only 7 (63.6%) of the women in the breast cancer group added something, all 16 (100%) women in the ovarian cancer group had something to share. These additions were usually a call for more women and providers to understand the symptoms of ovarian cancer, to work towards better screening measures for the disease, and just overall better outcomes for those diagnosed with ovarian cancer.

**Contributions to theory**

Theory is “an explanatory framework, a body of systematic suppositions derived from observations” (Ellen 2010:391). Using feminist theory and critical medical anthropology (CMA) as guiding theoretical frameworks, I hoped to bridge a microlevel analysis, at the level of the individual participant lived experience, with that of a macrolevel analysis, aimed at biomedicine. These two theoretical perspectives are intertwined and have several common goals including questioning the practices of those in power, critiquing biomedical hegemony, and attempting to incorporate the lived experiences of people into analyses. Both of these theoretical perspectives are important when dealing with a biomedical system which ultimately exerts control over and during the process of genetic testing and counseling. I see application of this dissertation research at a midrange theory level. Midrange theories “are attempts to identify the important patterns of thought or behavior in specific domains of a culture – patterns that are representative of an identified group of people in a designated context” (Trotter II, Schensul, and Kostick 2014:620). In respect to the current work, I attempted to identify patterns of women’s aspirations for, and experiences of, genetic testing within the United States, with both GP and NGPs providers.

A feminist theory approach necessitates an assumption that “gender is a dimension of societies and cultures that is key to understanding human relations” and therefore power dynamics exist that are built on assumptions about gender (Gailey 2014:148). For example, it foregrounds the importance of gender inequalities, such as the fact that today, only one-third of licensed physicians in the U.S. are female (Young et al. 2017). While I did not gather information on the gender of ordering providers, the majority of genetic
counselors are female (Legà et al. 2005) and evidence exists that women receive more comprehensive genetic counseling when they see a female provider versus a male provider (Zare, Sorenson, and Heeren 1984).

Regardless of gender, both women and men are acculturated into a male-dominated cultural and biomedical system which imbues particular values and praxis (Hahn 1996, Risberg, Hamberg, and Johansson 2006, Harding 2013). Much deeper than who the actual providers are, biomedicine is predicated on science that has historically excluded females as experimental subjects, which has focused on health problems of interest to men, which then becomes a part of mainstream scientific thought and literature (Rosser 2000). Feminist scholars have argued that one way in which we can see the results of a patriarchal medical system is through the process of medicalization and women’s bodies as a site for technological intervention, especially around reproduction and childbirth (Lock and Nguyen 2010, Rapp 2001). CMA likewise requires that we question the motives and practices of those in power. Given a male-dominated medical system, the treatment of women may be impacted by assumptions, beliefs, and behaviors of its practitioners, regardless of gender. Other scholars, too, have argued that specific to breast cancer research, problems with women’s health has come at the “hands of a male-dominated, hierarchical health system that is based on a biomedical model of medicine. The biomedical model focuses on anatomy and physiology and causes disease at the cellular, hormonal, and genetic levels rather than behavioral, social, and environmental contributions to disease. Since breast cancer is impacted by behavioral and environmental factors and is not a major health problem for men, it has received low priority, funding, and attention” (Rosser 2000:245-246). Genetic counseling, then, could be seen as a distillation of the biomedical model, as it is an entire field devoted to the individual and their genetics. Without situating the individual within larger social, economic, and political processes, other explanations for the disease and its progress may be lost or misunderstood. Evidence of this can be seen in participant’s responses to risk, which focused on individual behavior change and control. These responses reflect both cultural attitudes of risk control and the narrative of genetic counseling. This was also found in participant’s views on the cause of their disease, which some attributed solely to their BRCA mutation, or felt uneasy about the cause of their cancer when they came back negative. In both cases, women looked inward to the self – as the locus of control to combat further cancer risk, or as the sole cause of their disease.
All of the participants in this study were female and most were diagnosed with a mostly or completely gender-specific disease: either breast or ovarian cancer. Are the patterns identified in this study reflective of a larger, biomedical, male-dominated culture? I would argue so. At the very least, this study identifies patterns of care – for better and for worse, depending on provider type – among women undergoing genetic testing. The inability or unease of NGPs in having conversations with women about genetic testing is reflected in the extant literature and in this study. Women reported being left alone to digest brochures, or not even being told that they were undergoing genetic testing, which reflects aspects of biomedical hegemony and the assumptions that might be made of citizens who live under this paradigm. Do providers assume women understand risk assessments innately? Viewed as autonomous, rational actors, is it presumed that they understand what decisions can (should?) be made after receiving their genetic test results? Notably, biomedical training demonstrates a noticeable “discomfort with dialectical modes of thought” (Kleinman 1997:29). Dialectical thought, which is a discourse between two people who hold different points of view about something but hope to establish some sort of “truth” through reasoned arguments, is not what most biomedical practitioners are taught. Even genetic counselors, who are taught to hold conversations with their clients, lean towards education rather than a tête-à-tête between equals (Joseph et al. 2017). It is evident that this discomfort with dialectical discourse pervades the patient-provider encounter, often to the detriment of the patient. By understanding that these patterns of care reflect larger themes of biomedical thinking and training, it is my hope that actionable, applied interventions can be developed (discussed in Chapter 6).

Limitations

Despite the contribution to the literature, this study is not without limitations. This research only provides the voice of the patient in a complex web of medical interactions, which includes family, providers, and genetic counselors. It also replicates a voice which is the dominant one in genetic testing and counseling research – that of a middle-class white female. Given the health disparities present within the United States healthcare system, more work should be done to incorporate diverse voices to methodologically support an intersectional approach. Thinking of diversity diversely – linguistic, racial, sexuality, class, ability – would only strengthen future research. While I did reach out to local African-American cancer support groups in Tampa
Bay, I never received a response. Future research should attempt to incorporate a diverse set of voices which I failed to accomplish at present.

Another limitation specific to anthropological work was the lack of a “community” to fully immerse myself in. Interviews were the only contact I had with my participants, and because IRB mandated that participants be the one to contact me directly, participation may have been skewed towards women who were outspoken or were involved in cancer advocacy. Although I attempted to combat this lack of bounded community with attendance to a multi-day hereditary cancer conference and multiple hereditary cancer group meetings, I was never able to establish long-term rapport with my participants, although many indicated I should reach out to them again if I wanted. Methodologically akin to this is the fact that most interviews were conducted over the phone. Due to the self-funded nature of the research, compensation was not offered. In order to make the process as easy as possible for willing participants, I offered to conduct interviews in person or over the phone. While this avenue opened up conversations with women outside of Florida, it may have impacted the level of openness or in-depth quality of the interview.

Finally, I was attempting to elicit feedback about genetic testing and counseling. For some women, this occurred in the early 2000s; for others, within the last year. Additionally, some women had cancer diagnoses from 20 years ago, while others were actively battling new or recurrent diagnoses. While recall is certainly a factor, I think there is value in women’s recollections of their experience, even from almost 20 years ago. However, in future studies it would be helpful to understand these experiences along the continuum of genetic testing and cancer diagnosis.

**Summary of discussion**

Using thematic analysis, we are provided a clearer picture of how women hope to use their genetic test results. While some of the topics discussed have been covered in the literature, others such as treatment and cancer surveillance uses for genetic test results should be explicitly addressed during conversations around GT/C. Additionally, it is clear that there are differences in how women reacted to genetic test results depending on what kind of provider ordered testing. Participants who received a positive result through a
NGP reported largely negative reactions, whereas those who saw a GP were much more blasé. Interestingly, regardless of provider, women who received negative results reported mixed feelings. This warrants further research and may indicate a previously unknown need among some individuals who receive negative results. Finally, risk was discussed as an embodied, individualized behavior. Cancer risk is omnipresent in this sample. If risk is to be controlled, it is typically through individualized behaviors with the aim of being “healthy”, which reflects larger societal and cultural notions of risk.
CHAPTER SIX: CONCLUSION AND FUTURE RECOMMENDATIONS

Summary of major findings and their significance

This study, broadly concerned with women’s health, focused on the experiences of women who underwent genetic testing for hereditary cancer syndromes vis-à-vis various providers, who are either formally trained in genetics or not. First, I presented evidence of how women come to be tested as well as how they hope to use their test results. This data has implications for GPs and NGPs and can help guide their discussions with women undergoing genetic testing. Second, I argue that there is an important difference between women who undergo genetic testing with a GP versus those that do not. Women who see GPs are, unsurprisingly, better prepared for the genetic testing process. What is surprising, perhaps, is the myriad ways in which we see this preparation impact women’s experiences. The results of this preparation (or lack thereof) can be seen in women’s reactions to their test results as well as their recall of their testing/counseling experience. While I argue that GC is supremely important in engendering positive outcomes from the GT/C process, I also suggest that based on women’s recall of GT/C and their motivations for usage of their genetic test results, modifications to the counseling process should be considered.

Additionally, this study explores risk from a slightly different angle than previous research on risk in GT/C. Past work has assessed the communication and presentation of risk and uncertainty during the actual GT/C appointment (Paul et al. 2015). Rather than assessing if women could accurately recall their risk, this study sought to understand how women conceptualized risk. According to participants, risk is conceptualized in relation to cancer and as such is an expressed anxiety that is constantly in the back of women’s minds, to varying degrees. Participants do deeply worry about cancer risk, which is an entity that ebbs and flows day to day and can be triggered by external and internal factors such as medical tests and feelings within the body. Participants also reported engaging in specific individualized behaviors in order to control their perceived
cancer risk. How women think about and deal with risk also has implications for NGPs and GPs, especially when discussing cancer risks. Rather than presenting risk in relation to cancer, which may lean towards a “disaster and anxiety” narrative, perhaps more time should be spent with helping women gain more “peace of mind” as Lupton (1993:433) suggests. This could include discussions on how to control and think about risk in healthy, moderated ways. This may also include frank discussions of how cancer risk cannot be entirely controlled by women and their choices, which may alleviate some of the burden women reported feeling in regard to the ways in which they attempt to control their cancer risk.

**Applied recommendations**

A qualitative approach to this topic has provided rich data for the improvement of the GT/C process and experience. This approach not only supports past findings related to this area but also provides a more nuanced understanding of women’s experiences during the GT/C process. For example, participants spoke about using their genetic test results in relation to treatment and surveillance options. Not only did this provide a space for women to voice their own in-depth aspirations for genetic test results, but this research highlights that women’s conceptualizations of treatment and surveillance are slightly different than that of the medical community. This is an important distinction that providers need to understand in order to improve patient-provider communication. Allowing a space for women to voice the (sometimes) multiple ways in which they hope to use their genetic information provides a framework of topics for providers to discuss, some of which may not have been clearly articulated previously.

In addition to a qualitative approach, this research provides a longitudinal approach to women’s recollections of the GT/C experience. This was useful in multiple ways. First, I was able to interview women with a variety of experiences – most importantly, those who had testing through an NGP. This perspective is hard to reach and adds to existing literature on the deficiencies of NGP and genetic testing practices/lack of proper genetic counseling. Second, rather than simply assessing women on their recall of events or specific GT/C topics, this format permitted women to speak about what was important to *them*. If they did not find something important or applicable at the time of GT/C, it follows that that topic will not be something they
remember long term. The things that women described during their interviews are indicative of the things they found most important, interesting, or startling. Conversely, what was not reported during interviews is just as important. Topics that may be covered before undergoing genetic testing, such as education on the actual genetics of cancer (Berliner and Fay 2007), were not brought up as important. In fact, the few participants who spoke about that aspect of counseling said it was confusing. Critically assessing the counseling process to help determine exactly what information should be covered, and how, is important if we are to make improvements to the process. I believe that data from these interviews provides important insights into what is remembered and what is not.

With these findings, I see many areas of application. First, I hope this data can be used in advocacy efforts for GPs to be the only practitioners allowed to order and explain genetic testing. This research adds to the literature that has identified problems that can and do arise when medical providers, who are not formally trained in genetics, order genetic testing. This can include insufficient knowledge of genetics, being unable to recognize appropriate patients for BRCA testing, and being unprepared to counsel patients about their genetic test results (Bensend, Veach, and Niendorf 2014, Douma, Smets, and Allain 2016, Klitzman et al. 2013, Prochniak et al. 2012, Pal et al. 2013, Bellcross et al. 2011). As of now, any medical practitioner – from medical geneticists to nurse practitioners – can order testing, regardless of if they have had specific training in genetics. Unlike the United States, there are several developed nations which require the involvement of a GP prior to genetic testing (Rantanen et al. 2008). As my data reflects, without proper counseling there can be adverse effects: patients are left to disentangle and interpret complex medical and genetic information by themselves, they do not understand what they are being tested for, and the implications for family members are not elucidated. Additionally, and as Joyce pointed out, GPs are instrumental in coordinating follow-up appointments based on genetic test result and medical recommendations. GPs, and especially genetic counselors, do not just attend to the psychosocial needs of patients. They stay up to date on which genetic testing labs take which insurance, helping the patient navigate a complex arena of providers, insurance

15 Which is not only incredibly important but is an area of weakness identified by NGPs themselves (Klitzman et al. 2013, Douma, Smets, and Allain 2016).
coverage and laboratories. Because genetics is their primary focus, GPs stay up to date on various testing options available to patients. They understand how to correctly interpret results and can competently explain a positive, negative, or VUS test result. Not only can they explain those results, but they can then provide the next steps for recommended screenings and surgeries for patients and their family members. They also provide letters to help explain genetic test results to other family members and speak with patients about how to communicate those results. GPs are skilled practitioners that have a narrow focus within the medical system. Given that genetic testing options have changed so rapidly since 2013, it makes sense that these should be the only practitioners dealing with this information. And while there are limitations to where GPs can be accessed in person throughout the country, telephone and telehealth counseling are options. I believe that providers who do not have training in genetics should not be ordering genetic tests and should rely on experts to help navigate this complex terrain.

**For genetic counseling programs**

I recognize that genetic counseling master’s programs will have slight variations between them, but there are some general guidelines that are followed (Scott et al. 1988). Additionally, guidelines and coursework necessarily change over time. For instance, curricula have been updated to reflect the use of large-scale, clinical genomic sequencing (Hooker et al. 2014). Riconda et al. (2018) likewise acknowledge the many ways in which genetic counseling and genetic counseling training will change in the coming years. As genetic counseling moves forward as a discipline, reflexive work should be done in order to incorporate the changing needs and wants of the people receiving this genetic information, recognizing that genetic counselors can contribute to research in many ways (Biesecker 2018). This can be accomplished through ongoing research, which GPs should be a part of.

Based on this dissertation research, I recommend that a fundamental reassessment of the communication practices of GPs should be undertaken. It appears that GC programs may still rely heavily on the “teaching model” which is focused on health education rather than counseling (Kessler 1997). Evidence of this exists from a review of the literature which found that genetic counseling providers were speaking
more than clients, and used biomedically focused communication rather than psychosocial communication (Meiser et al. 2008). Other research has found that during counseling sessions, only 4.23% of counselor talk aimed to help patients apply genetic testing information to their own lives (Ellington et al. 2011, Roter et al. 2006). While GCs should have specific aims to accomplish during the counseling session, I think these sessions should also reflect how patients hope to use their genetic information. Unfortunately, there appears to be a disconnect between patient aspirations and the goals of the genetic counselor/GP. This idea is reflected in recent research by Joseph et al. (2017) who found that there is a fundamental mismatch of what information patients want versus what information is provided by counselors. For example, their main qualitative findings were that (1) patients felt that too much information was provided; (2) that the information was complex and that its presentation was conceptually difficult; (3) that the information presented was not relevant; (4) patient’s perceived that they could not ask questions or otherwise engage with the counselor; and (5) discussions of screening and prevention recommendations were vague (Joseph et al. 2017). Although the study by Joseph et al. represents a more racially and linguistically diverse sample than this one, the findings by Joseph et al. are strikingly similar.

Given the data from this research, along with Joseph et al and others, I recommend that detailed, concrete discussions regarding genetic testing information’s impact on family, patient treatment options and surveillance (both screening protocols as well as other cancer risks) take place. While some GPs may feel like these are topics that they already cover, I would ask that they evaluate their counseling techniques because the literature does not seem to support that notion.

Additionally, I would recommend that the scope of risk should be expanded during GC. Rather than focusing narrowly on presentation of risk, it may be helpful to acknowledge the impact of cancer risk on patients in the future. Preparing women for the potential anxiety that may accompany cancer risk, as well as healthy ways of coping and managing cancer risk, is not outside the purview of GPs (Croyle and Lerman 1999). This sort of longitudinal thinking could also extend to the cases of women who received negative test results. My results found that many of the women, both those who saw GPs and NGPs, struggled with negative test results. On the one hand, they were happy that they did not carry a mutation; on the other hand,
they expressed that they felt like they didn’t receive an answer for why they developed cancer. Rather than assuming that women who receive negative test results will not need psychosocial support or counseling, I recommend that women should be directly asked about how their test results make them feel, in a way to foster conversation and answer patient’s questions about why they were diagnosed with cancer.

**For non-genetics professionals (NGPs)**

Ideally, I would recommend that providers who do not have formal training in genetics stop ordering tests if they do not know how to properly prepare patients for the results or cannot accurately interpret the results. At the very least, GPs should be involved prior to the ordering of genetic tests. Based on this research and existing scholarship, I believe it is an agenda that should be addressed through legislation and active application of professional guidelines, several of which exist (Daly et al. 2018, Smith et al. 2018, U.S. Preventive Services Task Force 2015). A recent systematic review of primary care providers’ knowledge and communication of cancer genetic testing found that primary care providers had “incomplete or inaccurate knowledge about the inheritance and characteristics of hereditary cancer syndromes and interpretation of genetic test results” (Hamilton et al. 2017:318). Not understanding medical genetics yet ordering genetic testing anyway is unethical; it can put the patient at risk, as well as the physician at risk for malpractice (Guttmacher, Porteous, and McInerney 2007). Referring back to the story in Chapter 1, mistakes do happen and can have dire consequences. Less dramatic than the story of Elisha Cooke-Moore (Ducharme 2017) from Chapter 1 are some of the consequences documented here – women who are not told that they are undergoing genetic testing, women who do not understand what they are being tested for, or women who are told about their test results in a way that “knocks the wind out of them”. In fact, it is these ‘less dire’ situations which are a result of NGPs non-compliance with suggested clinical guidelines for discussing hereditary breast and ovarian cancer (which can be as high as 64% for some components) (Hamilton et al. 2017). NGPs who order genetic testing obviously feel that they are competent in ordering and delivering genetic test results (Douma, Smets, and Allain 2016). However, I would highly encourage NGPs to rely on experts with formal genetics training to accomplish the many tasks associated with preparing patients both pre- and post-test, especially given the reported “difficulties managing patients’ emotions” by NGPs (Douma,
In 2018, 59% of genetic counselors reported using phone counseling (National Society of Genetic Counselors 2018). With the rise of telehealth and telephone genetic counseling, options exist to counsel even without a counselor being physically present. I recognize that there are barriers in accessing GCs, primarily due to the relatively small number of GCs in the US\textsuperscript{16}. If those barriers exist and a NGP cannot access any sort of GC service, I would then highly encourage them to take it upon themselves to adequately perform the tasks necessary to properly prepare patients for the GT/C experience. I loathe to make a recommendation that potentially adds barriers to the US healthcare system. However, for patient safety I think it would be ideal to only allow GPs to order tests, rather than various doctors or specialists. This recommendation, which would ultimately be a policy-level change, is ambitious. However, given the potential negative consequences to patients as well as the documentation in the literature, it is a recommendation with patient safety as the priority. I imagine that GCs would also advocate for this change, as well. Not only would it serve to help ensure the physical and emotional health of patients undergoing genetic testing, but it could offer more job security and prestige and a way to help control a fast-changing genetic testing landscape.

\textbf{Conclusion}

This research sought to understand women’s views and experiences of genetic testing and counseling, as well as to determine how women think about risk. The results of this research enrich our understanding of women’s experiences during genetic testing for cancer syndromes in many ways. First, it adds to our understanding of women’s experiences with NGPs during genetic testing. Second, it supplements our understanding of what women remember from genetic counseling, and the impact that may have on women and their reactions to genetic test results. Third, it helps clarify women’s reasons for wanting to undergo genetic testing, and their ideas of how they hope to use their genetic test results. Lastly, we gain insight into how risk is conceptualized, and how it is informed through cultural notions of risk management.

\textsuperscript{16} The Professional Status Survey administered in 2018 by the National Society of Genetic Counselors (NSGC) was sent out to all 4,780 members of NSGC, the Canadian Association of Genetic Counselors, and diplomates of the American Board of Genetic Counseling (National Society of Genetic Counselors 2018).
Main points from the research show us that women have numerous motivations and aspirations for genetic testing and its uses, which should be incorporated into genetic counseling practices. Importantly, this research highlights areas within genetic testing and counseling which can and should be modified in order to improve (mental and physical) health outcomes of those undergoing testing. These modifications could take place on an individual level with currently practicing GPs, or through modification of genetic counseling master’s program curriculum.

While genetic tests are available through various providers, I argue that only providers with proper genetic training should order testing. The many facets of genetic testing and counseling—psychosocial counseling, informed consent, medical management, and familial implications—are components that need GP oversight. GPs will also be important in combating the anxiety almost all of the women in this sample expressed over their cancer risks. Anxiety about cancer risk uncovered notions of what it means to be at “high-risk” for this population. Additionally, women described how they attempt to temper that risk, mostly at the expense of the individual. The embodiment of cancer risk and anxiety highlights larger cultural notions of individual risk and responsibility, even when faced with hereditary and familial cancer syndromes.
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APPENDICES
Appendix A: Semi-structured interview guide

My name is Dana Ketcher. I am a doctoral student researching women’s experiences and perceptions of genetic testing and counseling for hereditary cancer syndromes. I’d like to invite you to participate in a voluntary research study to talk about your experiences and opinions. Although your participation may not benefit you directly, the information you share may help us identify ways to improve women’s experiences. All comments will be kept confidential.

This interview is expected to take about 30-45 minutes.

I am going to audio record our conversation so I don’t miss any information you share, but if you want me to turn it off at any time, just let me know.

There are two main things I am hoping to discuss during the interview. First, I want to find out if you have had genetic testing and/or counseling. Second, I want to ask you about your opinions about genetic testing and counseling for hereditary cancer syndromes.

First, could you tell me a little bit about your cancer experience? While we chat about that, I can draw your family tree (pedigree) to gather information about your personal and family history of cancer.

**Probes:** when/where/how were you diagnosed, what kind of cancer did you have, what kind of treatment did you receive?

At any point during your cancer experience, was genetic testing and counseling (GT/C) offered to you? Did you decide to undergo GT/C?

**IF YES**

Could you tell me a little bit more about your GT/C experience and why you decided to undergo GT/C?

*Probes:* did you just have testing, or did it include counseling? Who ordered testing? Who gave you your results? Do you remember your results?

What did you know about GT/C before undergoing testing? Did you have any family and/or friends who had had GT/C?

*Probes:* Can you tell me about their (friends/family) experience? What they told you to expect?

Was GT/C what you expected it to be like? Why or why not?

*Probes:* what did you hope to learn during your GT/C session? Did you have any expectations of what it was going to be like?

What was the most useful part about your experience?

*Probes:* try to determine specifics — was it the testing portion? The counseling portion? Because of whoever gave results?

What was the least useful part about your experience?

Was there anything you were confused about or unsure of?

Was there anything you wanted to talk about but didn’t during GT/C? If yes, please explain.

Did you share this information with your family? If yes, who? Why did you feel it was important to share (or not)?

Tell me about getting your test results — how did that make you feel? How did you use that information?
What (if anything) was the most valuable to you about genetic testing and/or counseling? If not valuable, why not?

Your provider suggested testing because you were deemed “high-risk” – what does that mean to you? Is that something you think about still today? How does that effect you?

**IF NO**

Why did you decide to not undergo GT/C?

*Probes: who offered you GT/C? Money, insurance issues? Do you remember how they described it?*

What did you know, or what have you heard, about GT/C?

*Probes: Did you have any family and/or friends who had had GT/C? What was their experience?*

What do you think **is** useful about GT/C? What is **not** useful about it?

Many people decide to undergo GT/C to help with their treatment plan and/or to provide information for their family members. Were these things you considered? How did they factor into your decision not to undergo GT/C?

**If testing was not offered...**

Why do you think testing was not offered to you as an option?

What did you know, or what have you heard, about GT/C?

*Probes: Did you have any family and/or friends who had had GT/C? What was their experience?*

What do you think **is** useful/valuable about GT/C? What is **not** useful/valuable about it?

**FOR EVERYONE**

Have you heard about newer testing options for hereditary cancers? *(Explain: now have tests that assess multiple genes [e.g. more than just BRCA1/2. While some genes have clear treatment options to reduce risk of cancer, not all these genes have clear treatment]).* Would that be something you would be interested in? Why or why not?

Great, I’d like to gather some additional information in order to help with my research. Please let me know the following:

- Education (highest degree received)
- Marital status
- Employment status
- Insurance status (at time of GT/C and current)

Before we end, I want to find out what else you would like to share with me today about your experience with or opinions about genetic counseling or genetic testing.

**CLOSING**

Thank you so much for sharing your ideas and time with me today.

**Appendix B: Pedigrees**
1. April (taken April 2017)

2. Katherine
Taken June 2017
3. Lisa (taken June 2017)

- Ovarian cancer
- Breast cancer
- Other cancer

- Ashkenazi Jewish, Russian
  - Male, 50s (dx. late 40s)
  - Male, 50s (dx. late 40s)
  - Male, 82 (dx. prostate cancer 65)
  - Male, 89 (dx. 70, 73, BRCA2+)
  - Female, 50 (BRCA2+)

- German
  - Male, 65 (dx. 55, BRCA2+)
  - Male, 69 (BRCA2+)
  - Male, 34 (BRCA-)
  - Male, 37 (No testing)
  - Male, 40 (BRCA2+)
4. Helen
Taken June 2017

- Breast cancer
- Other cancer

Sicilian/Italian

Hungarian

- dx. prostate ca 30s
- d. late 80s

- 66

- 65

- 50s

- dx. 40
- Panel negative

- 44

- 48

- 44

- 28

- 25

- 14
7_Lillian
Taken July 2017

Breast cancer  Other cancer

Scottish

Italian

dx. pancreatic ca?

d. 70s  d. late 60s

late 60s  d. late 60s

dx. endometrial ca 58

70

dx. 00s

d. 00s

74  d. 60s

75

49  dx. 42  BRCA negative

53

55

54  dx. skin ca 47

18
11_Sissy (Taken March 2018)

Breast cancer □ Other cancer □

Scottish

dx. unknown cancer
d. 1978

dx. skin cancer 1970
dx. prostate cancer 1972
dx. glioblastoma 1978

English

d. 1993

62
dx. 49
BRCA?
questionable colon cancer result?

66
13_ Martha (taken March 2018)

- Ovarian cancer
- Breast cancer
- Other cancer

Scotch-Irish

- d. 70a
- d. 70b
- d. 70s
- d. 78

German

- d. 78
- dx. stomach (?)

- d. 87
- dx. skin cancer

75

- dx. ovarian cancer 63
- dx. skin cancer (NCC) 73
- no genetic testing

75

- dx. 70

77

- 67

43

38

3

- dx. skin cancer (BCC) 40
16_Angie

| Spanish, Portuguese, Sephardic Jew
| d. 61
| d. 70s
| d. 63

| Polish, AJ
| d. 79
| Holocaust, unk

70
dx 66
BRCA neg

40

51
BRCA neg

Taken April 2018
24. Kristina (drawn April 2018)

25. Jill (taken April 2018)
28_Debra (taken April 2018)

- **Ovarian cancer**
- **Breast cancer**
- **Other cancer**

**Italian**
- ○ d. 75
dx. lung cancer
- ○ d. 83
dx. rectal cancer 63

**English**
- ○ d. 78
dx. rectal cancer 70s
- ○ d. 81
dx. breast 73
dx. BCC 60s

- ○ dx. 34
RRCA negative

- ○ 58
dx. leukemia 54

- ○ 31
- ○ 34
30_Gail (taken April 2018)

- Red: Ovarian cancer
- Blue: Breast cancer
- Green: Other cancer
- Yellow: Colon cancer

Irish, German

- D. 80s
- D. 83
  - Dx. breast 70s
  - Dx. colon 80s

Irish, German

- Dx. unknown cancer

- 71
  - Dx. breast 42
  - Dx. ovarias 66
  - BRCA positive

- 45
33_Bridget (taken April 2018)

- Red: Ovarian cancer
- Blue: Breast cancer
- Green: Other cancer
- Yellow: Colon cancer

Spanish, Dominican

- d. 50s
dx. colon cancer

- 68
dx. testicular cancer (young)

- d. 60s
dx. lung cancer

- d. 71
dx. breast cancer 67
dx. pancreatic cancer 71

- 64
dx. 51
BRCA negative

German, Welsh

- d. 65
- d. 86

- d. 85:
- 85
Appendix C. Selected participant excerpts

Appendix C1. Theme: Family

<table>
<thead>
<tr>
<th>Theme</th>
<th>Excerpt</th>
</tr>
</thead>
<tbody>
<tr>
<td>Family</td>
<td>“I was asking her [oncologist] generally speaking without knowing the source of why I had cancer, was there automatically a risk to my daughter. And we got into the conversation about her, and then we started talking a little bit more about my family history similar to what I shared with you, and she said you know what, let’s go ahead and do that kind of testing to understand that.” AND “…what do you do with the information when you get it? So in other words, let’s say if it came back positive for heredity, what do I do with that. Do you tell your daughter that immediately so she can be aware, but what are the implications of telling her that at 23 years old. We talked about, if you tell her, are you going to do anything about it? Are you going to say okay, maybe you should consider having a mastectomy? Things of that nature.” (Marjorie, 56, BRCA negative, breast cancer at 56)</td>
</tr>
<tr>
<td>Breast</td>
<td>“I made sure that my brothers knew that I was doing the testing, too. They thought, initially well, I'm a guy I don't have to worry about ovarian or breast cancer, and I was explaining that the BRCA gene, and this is what I learned from the testing, is that it can get passed down through the sons, too. So if I had the BRCA gene it's possible that they could have it, and so it could be something that gets passed down to their daughters.” (Melanie, 60, BRCA negative, breast cancer at 42, ovarian cancer at 56)</td>
</tr>
<tr>
<td>cancer</td>
<td>“Well what happened was they found mine [cancer] on a mammogram and did a biopsy and came back positive, and so then they looked at my [family] history and said, ooooh, we need to genetic test ya.” AND “They make you, they give you this letter, you need to send this to your whole family! Oh, okay. I shared it with my kids.” (Lisa, 66, BRCA2+, breast cancer at 55)</td>
</tr>
<tr>
<td>N=11</td>
<td>“My mom passed away in 2002 and my mother before she passed away got genetically tested and it was brand new back then. So my mother got tested and then she ended up being positive and she had three daughters and all three of us got tested.” AND “I started with my mother being tested and the 3 of us [siblings] got tested. Two of the 3 of us, had the gene. So my sister ‘M’ and I were positive. Obviously, it's a huge concern for us for our daughters.” (Gwen, 53, BRCA1+, breast cancer at 45)</td>
</tr>
<tr>
<td>Frequency: 63.6%</td>
<td>“And the genetic counselors were explaining if I was positive that meant that, it had implications for my daughter and my granddaughter and nieces in terms of how often they were monitored for this type of disease.” (Melanie, 60, BRCA negative, breast cancer at 42, ovarian cancer at 56)</td>
</tr>
<tr>
<td>n= 7</td>
<td>“I think we talked about the genetic testing because I have three daughters, and I have three grandchildren, and I just really needed to know, ya know.” (Helen, 43, BRCA negative, breast cancer at 40)</td>
</tr>
<tr>
<td></td>
<td>“But she [daughter] knows that she’s at a greater risk because of my testing. But what she chooses to do with that is out of my control. Dr. K [gynecological-oncologist] has talked to her about it and told her to get tested.”</td>
</tr>
<tr>
<td>Family</td>
<td>Ovarian cancer</td>
</tr>
<tr>
<td>--------</td>
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<tr>
<td>N=16</td>
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“T’m really interested in having that testing because I have two sons…Since I don’t have daughters, but I do have a granddaughter, might be helpful for her, too.”

(Martha, 73, no testing, ovarian cancer at 63)

“Just to know whether or not I carried that gene and thus if there was a possibility of my sons carrying that gene. And the real possibility of them developing cancer down the road.”

(Megan, 59, BRCA VUS, ovarian cancer at 39)

“...it [genetic test] came back and my daughter was in town so we both went because it would affect her as well, and it came back negative for BRCA1 and BRCA2.”

(Angie, 70, BRCA negative, ovarian cancer at 66)

“My second cousin Sally, who had ovarian and then had breast, she was the one, cuz I had had ovarian already the first time, when she was diagnosed with breast. We kind of corresponded and she said, I've been genetically tested I suggest that you do the same.”

(Roxanne, 61, BRCA1+, ovarian cancer at 52)

“Well then I knew that my sisters and any children and grandchildren were not at as great a risk for breast and ovarian.”

(Stacey, 77, BRCA negative, ovarian cancer at 67)

“Well because my mother had it [cancer] and I had it, so there was a very strong correlation. And he [doctor] was thinking, he knew I had a sister, he knew I had a niece, he knew I had a daughter…”

(Lynne, 60, BRCA2+, ovarian cancer at 54)

“Well I wasn’t offered [laughing, genetic testing]. I said, are you [oncologist] going to do it? Well, we’ll do it when frontline chemo is over. But you probably really don’t need it. I don’t know why he thought that, but they didn’t think I needed it. See the kicker to me of the whole thing is my normal gynecologist who I saw every year for 20 years, I told him every year, don’t forget: mother had breast cancer, her sister had ovarian cancer. Because to me, regardless of genetics that was a big red flag. And every year the gynecologist would say, were they diagnosed before the age of 50, and I would say no, and he’d say, then you don’t have anything to worry about. Well we all now know that that’s not accurate science and he should have known better, long before I was diagnosed. So that’s why I insisted through the oncologist that I get the genetic testing done.

(Joyce, 60, BRCA2+ and NBN+, ovarian cancer at 57)

“I would say finding out that I don’t have BRCA genes because I have daughters, so knowing that at least those particular things are not risks for my offspring. That was the most important thing for me. It was like okay, well they’re not BRCA so at least they don’t have to worry about that.”

(Clara, 57, BRCA negative, ovarian cancer at 54)

“I know I did the test because I wanted to make sure that my daughter wasn’t a risk factor [at increased risk for cancer]. And she’s not.”

(Marie, 57, BRCA negative, ovarian cancer at 51)

“I wasn’t genetic tested until my mother was diagnosed with breast cancer…they were asking her history and she didn’t say anything about me and I’m like…mom. So when I said I had ovarian cancer they all looked at me and said okay, stop, let’s start again…has anybody been genetic tested?”

(Debra, 62, BRCA negative, ovarian cancer at 34)

“Because I have children and grandchildren I really wanted to do it.”

(Kristy, 71, BRCA negative, ovarian cancer at 69)
“My primary reason for wanting to know obviously for myself was because I have 2 children so I have to think about them and then when they’re 18 I have to make sure that they get tested or if I’m not here to make sure that people know that they need to get tested.”

AND

“They knew that I had the BRCA gene and that was most likely the causation of my diagnosis so I told them [her brothers], you guys need to be tested because all of them have children. And I said the males, and one doctor told my brother that oh you don’t have to be tested because you’re a guy, and I said Kevin, that’s wrong.”

(Yvonne, 49, BRCA2+, ovarian cancer at 46)

“But now I feel like it’s more important for my family. It became more important for the family, to let them know, and gave me forms to pass on, telling them the importance and just saying what the percentages of chance of developing cancer and how to monitor it.”

(Bonnie, 61, BRCA2+, ovarian cancer at 59)

“And then I explained to the doctor, [sister] she had all this cancer, and this and that, and they just kind of put the pieces together and did this BRCA thing. The breast surgeon, right away he knew, he’s like let’s do this BRCA thing.”

(Joann, 47, BRCA1+, ovarian/fallopian tube cancer at 44)

<table>
<thead>
<tr>
<th>Family</th>
<th>No cancer</th>
<th>N=5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Frequency: 100%</td>
<td>n=5</td>
<td></td>
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</tbody>
</table>

“I said, mother’s test…she decided to do it after she was diagnosed because I have girls and I have granddaughters, and did it, and it came out negative.”

AND

“I do have three granddaughters and just for my own sense of, if I was at risk for it, and also for their sake.”

(Diane, 71, BRCA negative, no cancer)

“When she [mother] got diagnosed and we found out she was BRCA1, the doctor did the whole family tree and said, the girls need to get tested. So I got tested, I think right away.”

(Becky, 30, BRCA1+, no cancer)

“My half-sister was diagnosed with breast cancer and then went through genetic testing and discovered that she had this genetic mutation. My other half-sister, the one who did not have cancer, insisted that I go through this genetic counseling as she was doing as well.”

(Courtney, 37, unknown VUS, no cancer)

“I think I kind of resisted it at first because it was something my dad kind of kept pushing for after my mom passed, he would kind of push me and say oh you now, your next doctor’s visit ask them about this cuz your mom had all these indicators and your grandma had this too so you have a family history of it. And I think part of me didn’t want to do it because I kind of didn’t want to know, kind of part of my grieving process with my mom is like, I just didn’t really want to talk about anything that had to do breast cancer or anything related to it.”

(Leigh, 27, BRCA negative, no cancer)

“I was the lucky one. I never really considered testing because the gene had really, there was just talk about it. And back then they said, if your parent had cancer late, you wouldn’t have the gene. Well of course that’s not true, we found out later. And so, I wasn’t tested for a long time and then finally a doctor said, you know maybe you should be tested and so I didn’t have genetic counseling which was a huge mistake. My doctor just did the test and then I never expected it to be positive, really did not think it was going to be positive. Cuz my mother was the only one that had it, and we didn’t know if she had the gene or not, it was just, it was way back.”

AND

“My son was 28 and my girls were 25. Back then, I don’t know if it’s changed, they recommend testing at age 25, so they were just at the age to be tested. [she spoke to them about her results].”
### Appendix C2. Theme: Treatment

<table>
<thead>
<tr>
<th>Theme</th>
<th>Excerpt</th>
</tr>
</thead>
<tbody>
<tr>
<td>Treatment</td>
<td>“I just wanted to know if I should take my ovaries out.” (Katherine, 49, BRCA negative, breast cancer at 35)</td>
</tr>
<tr>
<td>Breast cancer</td>
<td>“Of course when they first diagnosed me with breast cancer the wanted to know if it was BRCA because of the treatment options and increased risk of ovarian cancer.” AND “At that time I did decide I was going to get a complete bilateral mastectomy because I felt like the less tissue the less chances of it coming back anyways because I was young. I was going to go through the whole radical mastectomy but because I wasn’t BRCA positive my surgeon said we could do nipple sparing because I don’t see it coming back inside of your nipples, that’s just rare. So that was one thing. She said if you were BRCA we would just be really aggressive and take everything anyway, like they did skin sparing at least.” (Gretta, 42, BRCA negative, breast cancer at 38)</td>
</tr>
<tr>
<td>N=11</td>
<td>“I was going to do a lumpectomy and radiation like my mom did. She did okay. I had to wait a month, so they said we’ll test you and then get you in. So then they did a lumpectomy, she said yea everything is fine. Go back two weeks later, I got the gene, I’m just having a mastectomy. And that way I didn’t need radiation which I was glad I didn’t have. I just had chemo.” (Lisa, 66, BRCA2+, breast cancer at 55)</td>
</tr>
<tr>
<td>Frequency: 72.7% n=8</td>
<td>“I went down there, and I’m pretty sure it was the nurse that did the original intake part of it, you know where they discuss everything with you, that just gave me the result but uhm…I think they were basing my chemo, and if they needed to do a double mastectomy off of the basis of those results. He [doctor] talked to me about, well what if they found out that I do have the BRCA1 or 2 gene, I had to say, I would like to know, because I could maybe be more proactive and take my ovaries out.” (Helen, 43, BRCA negative, breast cancer at 40)</td>
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<td></td>
<td>“I know some people have mixed feelings about genetic testing and, I always explain to people well on the one hand, oh it’s bad to have a genetic mutation, but for my understanding in the case of a reoccurrence, they know better how to treat people with a reoccurrence that had the genetic mutation. So it’s like, well if it comes back…they know more about how to treat that, at least in the case of ovarian cancer.” (Melanie, 60, BRCA negative, breast cancer at 42, ovarian cancer at 56)</td>
</tr>
<tr>
<td></td>
<td>“It would determine what type of surgery I was gonna have, mastectomy or lumpectomy vs. what type of treatment might be needed, so I was very aware of what the results, what the final results presented would determine in terms of treatment and surgery options. Options. The two of the three consultations said you need to have a bilateral mastectomy, and I already went into the initial diagnosis very stubborn saying I wasn’t having a mastectomy and I wasn’t having chemo, and I didn’t. And I didn’t need to.” AND “I’m driven and at that moment I said I’m doing this because this is going to determine the next steps on my journey. I didn’t know necessarily specifically what that meant, but I just knew that doing this would help me make the decisions with the OBGYN, and then with the oncologist as well as with who my breast surgeon would be, er my breast oncologist.” (Lillian, 48, BRCA negative, breast cancer at 42)</td>
</tr>
</tbody>
</table>
|               | “We [oncologist and her] had conversations about genetic testing if it does come back as positive,
when I was 35 I was planning to have my ovaries removed if it came back as positive. If it was negative they said I wouldn’t be at risk, but I did want to do that.”

(April, 39, BRCA negative, breast cancer 31)

“We [oncologist and her] talked about if its hereditary, the risk of dealing with this again is pretty high so you could take the route of having a mastectomy or double mastectomy, and then we talked about removing ovaries and uterus and all that.”

(Marjorie, 56, BRCA negative, breast cancer at 56)

<table>
<thead>
<tr>
<th>Treatment</th>
<th>Ovarian cancer N=16</th>
<th>Frequency: 62.5% n=10</th>
</tr>
</thead>
</table>
| “And I also knew at that time that if I tested positive there were trials for PARP inhibitors that I could then participate in. Well, if I did not have it then I was relieved for my children and if I did have the gene mutation then I understood I would also have to consider a prophylactic mastectomy. And uh, then I also knew that there were different protocols for people who were BRCA versus people who were not.”

(Angie, 70, BRCA negative, ovarian cancer at 66)

“At the time, I think my thinking was, that would make up my mind for treatment. If I have the genetic defect then for sure a double mastectomy.”

(Kristina, 60, BRCA negative, ovarian cancer at 21, breast cancer at 55)

“I don’t know, just to find out and it helps with treatments and the PARP inhibitors were just coming out but she [doctor] doesn’t do those unless you have a recurrence. It wasn’t the main reason why you should do it, like for your family, but I thought it was more to direct what chemo you might need.”

(Bonnie, 61, BRCA2+, ovarian cancer at 59)

“I mean he [doctor] was very forthright with me. He said you need to get everything removed. He said you need to get a mastectomy, and you need to...he said you don’t need your ovaries, he said you don’t need these things anymore.”

(Joann, 47, BRCA1+, ovarian/fallopian tube cancer 44)

“I think most hospitals are trying to get insurances to cover gyn-one patients genetic testing because they feel like it helps them treat. Cuz there’s a difference between treating BRCA1 positive versus negative.”

(Clara, 57, BRCA negative, ovarian cancer at 54)

“...it was more they were going over whether they were positive or negative and those that were positive, why they figured they were going to be found positive and, the repercussions of that, whether or not to discuss it with their kids if they had kids, and to suggest that they have the testing themselves and then the decisions for them to have to make as far as doing things ahead of time as far as having mastectomy or having hysterectomy et cetera.”

(Megan, 59, BRCA VUS, ovarian cancer at 39)

“I’m hoping it can help if the cancer comes back or if I get another cancer, that it will help maybe target my therapy better? Than if I had not gotten genetically tested?”

(Roxanne, 61, BRCA1+, ovarian cancer at 52)

“And no, I’m not going to do prophylactic mastectomies because guess what? Medicare doesn’t pay for prophylactic mastectomies, even with the test results. They don’t even pay for, if you got breast cancer in one breast, they don’t pay for removing the other one as a prophylactic measure. And since I went on disability from the cancer then I’m now on Medicare, so yeah.”

(Joyce, 60, BRCA2+ and NBN+, ovarian cancer at 57)

“...also it’s my understanding that some chemos work better if you’re a BRCA carrier than if you’re not.”

(Kristy, 71, BRCA negative, ovarian cancer at 69)

“I scheduled my prophylactic mastectomy and in that process of the pre-op work they needed a
breast MRI within 12 months before they would do surgery. And because I was in the study and I was on top of all my appointments, I said my breast MRI is 13 months, are you really going to make me do another one? And they said yes, it has to be within 12 months. I said okay fine. So a few months before the surgery I did the breast MRI and they found my breast cancer. So my sister’s prophylactic surgery was not prophylactic, and my prophylactic breast surgery was not prophylactic.”

(Gwen, 53, BRCA1+, breast cancer at 45)

<table>
<thead>
<tr>
<th>Treatment</th>
<th>“Immediately I said I want surgery. I don’t want to live with this. I want a mastectomy. And almost immediately I started looking into different options.”</th>
</tr>
</thead>
<tbody>
<tr>
<td>No cancer</td>
<td>(Julie 71, BRCA1 positive, no cancer)</td>
</tr>
<tr>
<td>Frequency: 40%</td>
<td>“I want to have the surgeries, but I want to have kids.”</td>
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<tr>
<td>n=2</td>
<td>(Becky 30, BRCA1+, no cancer)</td>
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Appendix C3. Theme: Surveillance

<table>
<thead>
<tr>
<th>Theme</th>
<th>Excerpt</th>
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<tbody>
<tr>
<td>Surveillance</td>
<td>“They had asked me if I wanted to do a drug trial that I turned down but I did want to do the DNA, or the genetic testing. Because my next concern was ovarian cancer. And if I have this cancer that came out of nowhere, when is that cancer gonna come.”</td>
</tr>
<tr>
<td>Breast cancer</td>
<td>(April, 39, BRCA negative, breast cancer 31)</td>
</tr>
<tr>
<td>N=11</td>
<td>“That is what I was thinking of, if I have it, I’m going to get my ovaries taken out, and now I have to have my kids having earlier surveillance. You know they already have to have early surveillance because of me, because of getting it so young.”</td>
</tr>
<tr>
<td>Frequency: 63.6% (n=7)</td>
<td>(Katherine, 49, BRCA negative, breast cancer at 35)</td>
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<tr>
<td></td>
<td>“I had options to hopefully prevent future cancers was a good outcome that I would have never known otherwise.”</td>
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<tr>
<td></td>
<td>(Lisa, 66, BRCA2+, breast cancer at 55)</td>
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<tr>
<td></td>
<td>“I do remember them [genetic counselor] talking about well, here’s knowledge that you have this gene and you have these increased chances of getting these cancers.”</td>
</tr>
<tr>
<td>AND</td>
<td>“So my sister J and I were positive. So J and I joined a study, a local study, in St. Pete with a doctor that one arm of the study was to get your CA125 checked, we used to get blood work done every 3 or 6 months, I can’t remember, we also had ultrasounds of our ovaries every 6 months or whatever. We were in this study that did a lot of checking, so it was kind of overly checking and it was one of the things they wanted to know is if your CA125 would predict ovarian cancer, etc.”</td>
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<td></td>
<td>(Gwen, 53, BRCA1+, breast cancer at 45)</td>
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<tr>
<td></td>
<td>“I think that right about the time that was happening [Lynch testing in her relatives] my dad had colon cancer, so they went from saying it was a coincidence with me, because no one in my immediate family had cancer, so then finding out my dad had cancer, and I started looking up connections between breast and colon cancer, and I just wanted to make sure…that either there wasn’t a connection between my dad and mine’s cancer or especially the Lynch. Like I said, the pancreatic, by the time you know is too late, and I just was kind of in survival mode. I didn’t want any surprises, anymore cancer after I just finished treatment. I would definitely say that I think genetic testing should be pretty much given for any cancer diagnosis of anybody, but I think it should also be included if your relatives have it. You shouldn’t have to worry about paying for it, it’s proactive, it’s preventative. It could save a lot of lives. If my uncle knew he was Lynch positive and what that meant, they might have been able to scan if he wanted to or treat him earlier. Even my niece, she’s 26 years old she would never be thinking about the fact that she, not just the pancreatic cancer but the ovarian cancer, they’re recommending...”</td>
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</table>
she have a hysterectomy when she’s 30. She’s been diagnosed for 2 years with the Lynch so she can at least be proactive and make some decisions about not just her cancer health but her family choices and all that, instead of my uncle being 60 being given 2 months to live.”

(Gretta, 42, BRCA negative, breast cancer at 38)

“It was just the one on the colon which was interesting because I did have a polyp. And so they were gonna do me like every 10 years and now I’m taking that into him, so hopefully I’ll do it every year, I hope. I hope that will give him the okay and insurance will say okay.”

(Sissy, 62, BRCA negative/Lynch VUS, breast cancer at 49)

“…it was just helpful to know and to be able to share with my daughter the recommendations they had about how often my daughter should be checked for breast cancer and stuff like that, to give her that type of feedback.”

(Melanie, 60, BRCA negative breast cancer at 42, ovarian cancer at 56)

<table>
<thead>
<tr>
<th>Surveillance</th>
<th>Ovarian cancer</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N=16</td>
</tr>
<tr>
<td>Frequency:</td>
<td>31.3% (n=5)</td>
</tr>
</tbody>
</table>

“...And I remember, she [genetic counselor] said because there has been other cancers in your family I would recommend the whole panel, like pancreatic in particular. She said, you can just do the BRCA but she said, you’ve got pancreatic with the female relatives on your father’s side and she said if I were you I would go ahead and get that done. And as soon as she said that I was like oh, I didn’t even know that there could be testing for other things at the same time. I just had no idea. And since then, in the last two years, I have read something of the possibility of some connection between the pancreatic mutation and the ovarian.”

(Jill, 64, BRCA negative, ovarian cancer at 51)

“The knowledge that I was also at risk for additional cancer, for breast cancer. I wasn't going to fight ovarian cancer and then turn around and get breast cancer if I could help it.”

(Lynne, 60, BRCA2+, ovarian cancer at 64)

“Well I’m the one basically that has had to say to the provider, you know, I’m BRCA2 positive, you know that puts me at a higher risk for…depends on the provider, what disease we’re talking…and they all look at me and go, okay, they just, it’s almost as if the dermatologist and the GI doctor in particular are less than aware of the issue. They don’t seem to care if you’re not bleeding to death. There are actually the University of Chicago’s protocol for screening for pancreatic cancer that one of my brother’s actually received and when I tried to talk to 2 different GI doctors they were like, nah we don't need to do that. Although I’m not at humongous risk, I think the risk is 7% greater than the average person’s risk of pancreatic cancer, but considering pancreatic cancer is so incredibly deadly, why would we not screen?”

(Joyce, 60, BRCA2+ and NBN+, ovarian cancer at 57)

“I saw a genetic counselor and she took blood and she sent it out, they sent it to the same lab. And then mine came back positive. And we knew, my doctor was positive it would come back positive. So when the results came back I went in for the appointment and she talked all about the chances…see even though I’m BRCA positive, I really should’ve theoretically speaking developed breast cancer not ovarian cancer, because with BRCA2 you’re more predisposed, your percentages of developing breast cancer are higher than ovarian cancer even though your risk increases for ovarian cancer too. So they did suggest that I be monitored closely for breast cancer. I’m 80% more likely to get breast cancer with this gene.”

(Yvonne, 49, BRCA2+, ovarian cancer at 49)

“I feel like the genetic testing is important. And especially if you know that, now you know, we’ve got this thing in the family, you need to be on top of it. You’ve got this gene, it’s like anything else. Now you [family members] do your homework and you stay on top of it.”

(Joann, 47, BRCA1+, ovarian/fallopian tube cancer at 44)

<table>
<thead>
<tr>
<th>Surveillance</th>
<th>No cancer</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>N=5</td>
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“I wanna be proactive, but also at the same time feel like with this BRCA mutation I have a bomb attached to me and I do the self-exams and I have fibrous breasts, so a million lumps but anytime I feel anything I’m always like, what is this what is this what is this, and I think about it at night. I know I’m only 30 but, younger girls than me have gotten cancer. I feel like it’s a bomb waiting to go off.”

(Becky, 30, BRCA1+, no cancer)
“She [OBGYN] did talk to me about it, I don’t remember the exact ages she said, but she said typically people start doing screenings at this age but we’re gonna start you off earlier, I don’t remember what the typical age was...is it like 35 or something? I think it was whatever the recommended age she said something like 5 years before that, I dunno if she said around age 30 they want to start with screenings...she did talk about that part when I asked about the testing.”

(Leigh, 27, BRCA negative, no cancer)

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<tr>
<th>Theme</th>
<th>Excerpt</th>
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<tbody>
<tr>
<td>Information/research</td>
<td>“With all the things that she (GC) said, and I felt like...I was thrilled that she was telling me all this information, the more information the better, but genetics, like any science or medicine, is super complicated, and I think it’s very difficult to explain to the average person who doesn’t know much about it, and I don’t know much about science, the only time I was interested in science was when we were talking about genetics. So, I was fascinated with what she was saying, but so much of it was, there was no context for it really. I don’t have a box to put this in, I don’t have filter to run this through to translate it into my words, or how to make it apply to my life or my body. And that was where I would like to have, in retrospect, gone through that meeting, listened to her, and then just had another meeting scheduled, not me having...I’m sure she said, let me know if you have any questions, here’s my card, let me know if you have any follow up questions. Well, I don’t even know! You know, I don’t even know what to ask, I don’t even know if this is a legitimate question, did you answer this the first time? I don’t even know. So, I almost wish that it included two appointments, one where she goes through everything and then another one where you come back and, okay now I’ve thought about this, I’ve done a little bit of research, I’ve talked to a few people, I have a couple questions, what about this...I think that would have been helpful.” (April, 39, BRCA negative, breast cancer 31)</td>
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<tr>
<td>Breast cancer</td>
<td>“...the process [genetic testing] itself was very easy, to me it was empowering because it’s given me more education. I would do all the time if they have more advancements, absolutely. Like figure this out. And then the second time [having genetic testing done] is just kind like, trying to resolve this mystery. Because, my only treatment, to this day, is chemotherapy. If there’s anything out there that can help people, researchers, oncologists, figure out what’s driving it, maybe they can help treat it better. Because right now it’s hard to treat, if they don’t know about it...it’s like for triple negative they need to find out as much as they can so they know what can work besides the thing that’s old.” (Katherine, 49, BRCA negative, breast cancer at 35)</td>
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<td>“I mean, it’s information you can use to help your family, I don’t understand why anyone would say no unless they had something to hide, I dunno. You need to know if you’re positive to see what drives your cancer so they can critique the treatment just for you. Like your long-term treatment, they put me on medicine for 10 years because I was so young. Otherwise how would they know what to do for you?” (Helen, 43, BRCA negative, breast cancer at 40)</td>
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<td>“Knowledge is a good thing. I know some people have mixed feelings about genetic testing and, I always explain to people well on the one hand, oh it’s bad you have a genetic mutation, but for my understanding in the case of a reoccurrence, they know better how to treat people with a reoccurrence that had the genetic mutation. So it’s like, well if it comes back...they know more about how to treat that, at least in the case of ovarian cancer.” (Melanie, 60, BRCA negative breast cancer at 42, ovarian cancer at 56)</td>
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<td>“And it was kind of new, and so I thought, if I could help with anything, it didn’t matter that my name was out there.”</td>
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<tr>
<td>Information/research</td>
<td>No cancer</td>
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<tr>
<td>Ovarian cancer</td>
<td>N=5</td>
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<tr>
<td>N=16</td>
<td>Frequency: 20%</td>
</tr>
<tr>
<td>n=1</td>
<td>Frequency: 20%</td>
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“'I feel like it's research...so because it's research I think, we don’t know what we don't know. I look at research and say okay, collect all the information you want. You're not gonna get a chance to talk to me again you want to do it now, fine. So I didn't feel like any of it was a waste. Because I'm in a university setting, b/c exciting new things happen all the time and I know that, there wasn't any part of it that felt wasted.’”

(Clara, 57, BRCA negative, ovarian cancer at 54)

“'I know I had to sign a permission slip and I knew it would be used for research and I really feel an obligation to further research so that, maybe a generation or two from now, something that I have or don't have will help others.’”

(Kristy, 71, BRCA negative, ovarian cancer at 69)
### Appendix C5. Theme: Ashkenazi Jewish heritage

<table>
<thead>
<tr>
<th>Theme</th>
<th>Excerpt</th>
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<tbody>
<tr>
<td>Ashkenazi Jewish heritage</td>
<td>“They [doctors] said, well ya know your heritage because your Jewish you’re from the Ashkenazi cuz where your parents came from, there were 40 of them and they interbred and that’s where this gene came from.” (Lisa, 66, BRCA2+, breast cancer at 55)</td>
</tr>
</tbody>
</table>
| Breast cancer | N=11  
Frequency: 9.1%  
n= 1 |
| Ashkenazi Jewish heritage | “It was from the minute I changed physicians and hospitals that was the first thing he said, you’re Ashkenazi, you better have testing.” (Angie, 70, BRCA negative, ovarian cancer at 66) |
| Ovarian cancer | N=16  
Frequency: 18.8%  
n=3 |
| Ashkenazi Jewish heritage | “I don’t know if you’re familiar with Hadassah hospital in Israel? I belong to Hadassah here where we, obviously its educational and activism for women health issues, but we also raise money to support Hadassah Hospital in Israel and all of their functions. I don’t know why but I somehow mentioned Hadassah in my very first visit and the doctor right then and there boy she was really tuned in, I guess she couldn’t ask my religion? So she [doctor] asked me if I knew any Ashkenazi Jews and I told her I was one and she said well once we get you started let’s talk about genetic counseling if you are interested.” (Kristy, 71, BRCA negative, ovarian cancer at 69) |
| No cancer | N=5  
Frequency: 20%  
n=1 |
| Ashkenazi Jewish heritage | “[Was your AJ heritage an indicator to your doctor?] Yes. But in 2002, really there wasn’t a whole lot known, the gene was only discovered I think in 1994, so there wasn’t a lot of progress made. I mean, there’s so much more now from 2002 from when I found out to now. It was the early days of information.” (Julie, 71, BRCA1+, no cancer) |

### Appendix C6. Reactions to test results: positive, NGP

<table>
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<tr>
<th>Category</th>
<th>Excerpt</th>
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<tbody>
<tr>
<td>Test results (POSITIVE, NGP) (n=5)</td>
<td>“I don't know. I just remember Dr. K told me I was BRCA positive.” (Gail, 71, BRCA+, breast cancer at 42, ovarian cancer at 67)</td>
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<td>“It just sort of knocked the wind out of me. He said, I got the results, and there positive. And it’s like, whoa. I sort of didn’t believe it. So, you know, basically there was no preparation. If you were having genetic counseling, you have your appointment first, and they explain what it’s all about, and then you go back to get your results. So you kind of prepared like, oh like I’m going to be positive or, hopefully I’m negative but at least you’re prepared for it. This hit me like a ton of bricks.” (Julie, 71, BRCA1+, no cancer)</td>
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<td>“Well I was pretty shocked because I was the first one in the entire family to have any kind of genetic testing so I just really was surprised. But then a little bit scared because of the increased risk for other cancers.” (Joyce, 60, BRCA2+ and NBN+, ovarian cancer at 57)</td>
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<td>“It made me feel angry with myself that I hadn't taken advantage of the free testing that had been offered to me about 4 months earlier. Four or 5 months earlier I was offered free...” (Julie, 71, BRCA1+, no cancer)</td>
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testing and I started the process and I didn't finish. My life would have been so much different. Can you imagine? I was offered free BRCA testing and it would have come back, hey by the way you're positive, I would have rushed right in for a hysterectomy. But no, for whatever reason I delayed it, I wasn't worried about it. My gynecologist kept saying, when are we going to take out those ovaries? She didn't want me to have them, she'd been telling me for 2 years you need to get those over is out they're not doing you any good and they're just a risk. She knew my mother had had a risk, she knew my mother, but there was no evidence for me. But BRCA testing was expensive, I think at the time it was three or four thousand dollars yes. Guess what I couldn't afford that at the time. I have financially struggled since, I lost a lot of income and I spent a lot of money, not as much as my insurance company, but it's cost me a lot and I know they're not supposed to hold past circumstances against you but my insurance is horrible, mine is terrible. So yeah I was very angry at myself.”

(Lynne, 60, BRCA2+, ovarian cancer at 64)

“And then I explained to the doctor, [my sister, deceased] she had all this cancer, and this and that, and they just kind of put the pieces together and did this BRCA thing. The breast surgeon, right away he knew, he's like let's do this BRCA thing. After chemo and radiation and all that, I'm going with her [sister, diagnosed breast cancer] to her final visit to remove the bandages and she's just flat like a boy! Loving it. That's when Dr. [breast surgeon] looked at me and said let me just test you. Then when I was positive and I saw him because he wanted to discuss you know, what we're going to do now, he said to me, I knew you would be positive. And I'm looking at him going, I still don't know what BRCA is and why did you know?”

(Joann, 47, BRCA1+, ovarian/fallopian tube cancer 44)

Appendix C7. Reactions to test results: positive, GP

<table>
<thead>
<tr>
<th>Category</th>
<th>Excerpt</th>
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| Test results (POSITIVE, GP) (n=6) | “I was upset, but at the same time I knew it. I always had this gut feeling that I had it and then, of course, the guilt of why didn't I push harder 10 years ago, she [mother] wouldn't have had to go through that [getting diagnosed with breast cancer].”
(Becky 30, BRCA1+, no cancer) |
| | “How did we [siblings] feel…disturbed? Upset? I can tell you how my older sister, the one who did not have it, took it. Of course, she was incredibly relieved, but she had that guilty feeling that the two of us both had it. I should probably say, we are an incredibly close family. My kids and their cousins are like all siblings, we are incredibly close, so my older sister felt very guilty. Not guilty that she didn't have it but guilty that we had it.”
(Gwen, 53, BRCA1+, breast cancer at 45) |
| | “It made me feel good, like it wasn’t my fault. It wasn’t what I ate or what I…it gave me a reason why I got it, cancer.”
(Bonnie, 61, BRCA2+, ovarian cancer at 59) |
| | “I wasn't surprised.”
(Roxanne, 61, BRCA1+, ovarian cancer at 52) |
| | “I honestly didn’t…I already knew it. It was just confirmation of something I already knew, so it was like okay. I hate to sound so blasé about it but that’s really what’s gotten me through this whole thing, it’s just to have a very pragmatic kind of, let’s just get this done type of attitude. I’ve never said oh why me and why is this happening to me and my life is terrible. It’s just, it is what it is and what do we need to do to fix it. And what do we need to do going forward as far as being proactive about it and I feel my medical team is very proactive so I just take each day as it comes. I have the gene, okay, there’s nothing I can do about it. I’m having recurrences, there’s
nothing I can do about it. Not to be all Christian-γ about it but it’s in god’s hands and his will be done and, He is working through my medical team and that’s all I can do.”
(Yvonne, 49, BRCA2+, ovarian cancer at 46)

Appendix C8. Reactions to test results: negative/VUS, NGP

<table>
<thead>
<tr>
<th>Category</th>
<th>Excerpt</th>
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<tbody>
<tr>
<td>Test results (NEGATIVE/VUS, NGP)</td>
<td>“I know I was relieved when the results came.”</td>
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<tr>
<td>(n=7 negative, n=2 VUS)</td>
<td>(Stacey, 77, BRCA negative, ovarian cancer at 67)</td>
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<td>“For whatever reason that I don’t know I have a label to attach to it, I was thrilled! And</td>
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<td>I don’t know why at that time I was thrilled that it was negative. Other than…I dunno, I</td>
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<td>guess because my two aunts, their procedures, and both of them went through Yale, which is</td>
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<td>where I was gonna go, so they had really quality medical care and quality doctors…I just,</td>
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<td>knew of their story and experience and it was just so damn miserable. The way they</td>
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<td>approached it and their procedure and recovery and…I recall Aunt I did a double</td>
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<td>mastectomy, but she was also ducal carcinoma, I think, and Aunt D, they both</td>
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<td>seemed to be so…miserable, or angry. I’m not saying I wasn’t angry…I don’t know. There</td>
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<td>was something. I still felt a lightness about it even though I was angry and uhm…but</td>
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<td>hearing negative was like yahhhhh whoo hoo!! In my mind a negative was really good! And</td>
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<td>positive was bad.”</td>
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<td>(Lillian, 48, BRCA negative, breast cancer at 42)</td>
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<td>“It felt good to know but it was also kind of like…I know that the odds are better</td>
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<td>not to have it, but still, I don’t remember the last time I looked at what the risk is, is</td>
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<td>it like I out of 5 women will have a breast cancer diagnosis? I don’t remember, cuz I</td>
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<td>guess I’m still just afraid I’ll have it anyways, so it was a little bit of a relief. It</td>
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<td>was like okay, at least I might not have, cuz my mom tested positive for it and hers was</td>
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<td>triple negative, it didn’t show up on a mammogram or an x-ray, maybe it was that it didn’t</td>
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<td>show up on a mammogram they had to do an x-ray to see it. So by the time they found it</td>
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<td>it was stage 4 and it had metastasized and it was just a really aggressive form, and so</td>
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<td>I guess I feel relieved that since I don’t have the genetic indicators I won’t have as</td>
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<td>severe as her, if it does happen, but I guess, I wasn’t as relieved as I thought I would</td>
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<td>be getting the test results, I still kind of freaked out about it just because she had it.”</td>
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<td>(Leigh, 27, BRCA negative, no cancer)</td>
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<td>“And it was really weird to hear back that no, I didn’t have those markers. And the breast</td>
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<td>specialist said well it’s probably just some kind of a genetic flaw but they haven’t</td>
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<td>found that particular gene, they’re not testing that particular gene. I was just stunned!</td>
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<td>It was sort of like, should I do that test again? Could we do a redo? I was so expecting it</td>
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<td>to say yes, and when it was no it was, I thought that was gonna be the thing that made my</td>
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<td>decision for me. So I don’t know if you could say I was disappointed but I was</td>
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<td>surprised, I was really surprised.”</td>
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<td>(Kristina, 60, BRCA negative, ovarian cancer at 21, breast cancer at 55)</td>
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<td>“Ironically enough, I had mixed feelings about the results and that probably sounds</td>
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<td>crazy…but a part of me did not want it to be hereditary, obviously for my daughter’s</td>
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<td>sake and her future children’s sake but there was a part of me that wanted to know the</td>
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<td>source, so it ruled out heredity but it didn’t tell me the source, so there’s a little bit</td>
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<td>of frustration in not being able understand how this happened.”</td>
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<td>(Marjorie, 56, BRCA negative, breast cancer at 56)</td>
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<td>“Well it made me feel a little bit less stressed about getting it but then not really</td>
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<td>because I said mother didn’t have it and she had ovarian, know what I mean? It’s sort of</td>
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<td>like you think, okay well I’m in the clear but I’m not.”</td>
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“Well the doctor explained to me that it was good and bad. Good for my family and not bad but that it maybe limited some of the avenues that were available for treatment for me. It just is and move on with what you can do. I don’t know if I was expecting it. Before I became ill I never really thought about it because of my family history I never thought about being a carrier. So I don’t know that I was surprised, I think maybe I was relieved because of my children. But I don’t think I would have been surprised had I been a carrier. I don’t think I would have been surprised either way.”

(Kristy, 71, BRCA negative, ovarian cancer at 69)
“I was kind of hoping that they were gonna find…something? A reason, because it’s all a mystery for me, at least, with my sub-class [triple negative]. Just the unknown of everything is scary, not knowing what’s feeding or driving this cancer. I mean I was glad that I didn't have anything that I passed on to my kids, that was my main concern, but the process itself was very easy, to me it was empowering because it’s given me more education. I would do all the time if they have more advancements, absolutely. Like figure this out.”

(Katherine, 49, BRCA negative, breast cancer at 35)

“There was definitely some sense of relief and there was a little bit of, huh, like a little disappointment, I’m surprised, I thought that was gonna help us there, right? If I was 40% more likely, to be carrying this gene which gives me a predisposition cuz of course now…not that it matters that much, but there’s not really a cause. What is the cause of my ovarian cancer? I don’t know. It’s not my genetics, or it might be my genetics and we just don’t know it yet, or it might be environmental. So in some ways having the positive result would at least be like, oh okay, it gives you a place to put the blame. Life’s just not that clean, I get it. But people like to have answers in that way.”

(Clara, 57, BRCA negative, ovarian cancer at 54)

Appendix C10. Women’s experiences with NGPs

<table>
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<tr>
<th>Code</th>
<th>Excerpt</th>
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<tbody>
<tr>
<td>Genetic testing (NGP)</td>
<td>[Explained her sister’s ovarian cancer diagnosis] “And then I explained to the doctor, Marianne she had all this cancer, and this and that, and they just kind of put the pieces together and did this BRCA thing. The breast surgeon, right away he knew, he’s like let’s do this BRCA thing. That's when Dr. C looked at me and said let me just test you. Then when I was positive and I saw him because he wanted to discuss you know, what we’re going to do now, he said to me, I knew you would be positive. And I’m looking at him going, I still don’t know what BRCA is and why did you know? [asked her if the surgeon clarified BRCA to her] Do you know what, I still really don't know? When there is so much going on, now I know that’s why it’s important to take people with you or take someone with you, because I’m not listening to half the stuff. My brain like shut down, I’m like surgeries? Double mastectomy? Remove ovaries, fallopian tubes, all that why? I’m hearing, menopause, and I'm like what? I don’t want to go into menopause! My understanding is just that it's this horrible gene, he [the doctor] said it's good I never had children…and he said to me, this is not something that you want to pass down. I just know it's not a good thing to have.” (Joann, 47, BRCA1+, ovarian/fallopian tube cancer 44)</td>
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<td>“It would have been after my radiation/chemotherapy I was having a conversation with a doctor [oncologist], just a general conversation, and that led us to the path of maybe we should do the genetic testing. I was asking her generally speaking without knowing the source of why I had cancer, was there automatically a risk to my daughter. And we got into the conversation about her, and then we started talking a little bit more about my family history similar to what I shared with you, and she said you know what, let’s go ahead and do that kind of testing to understand that. I asked about repercussions of the test, cuz what I was concerned about if I take this test and it comes back hereditary, is there anyway shape or form that could impact me or my daughter from an insurance perspective. She basically talked about the privacy of information, and insurance company couldn’t just...let me say it differently. If I was looking to get insurance with a different company they couldn’t automatically request my medical records and see that. She said this insurance company would have any idea because obviously they’re paying for the testing, but she didn’t really think there were any repercussions to doing that. [any other issues you brought up before testing that you were curious about?] No, she had given me before I actually took the test a bunch of information, you know contained</td>
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in brochures and stuff like that to let me read it because initially I was not comfortable with it. I wanted to get it done, but even when she told me she didn’t think there was any insurance risk I wanted to do a little bit of digging myself, so I didn’t take the test immediately, so I read a lot of stuff in the brochures on my own, and then I subsequently took the test.

(Marjorie, 56, BRCA negative, breast cancer at 56)

“I went and did the biopsy and then I waited for the results and as soon as she [OBGYN] called me with the results that day, when I hung up with her I said I’ll be in your office in a few minutes. And that’s what I did. I called my brother and told him to tell my family, that I was going to do the bloodwork for genetic testing before I freaked out even more per say, but I was actually very calm. I handle things very well, for the most part. So I was on a mission, and I went into her office and did the genetic testing. I think she had mentioned the testing again and I said I’m coming in and doing it. There’s no questions, I didn’t care about the cost, I just was doing it.”

(Lillian, 48, BRCA negative, breast cancer at 42)

“I wasn’t tested for a long time and then finally a doctor [surgeon] said, you know maybe you should be tested and so I didn’t have genetic counseling which was a huge mistake. My doctor just did the test and then I never expected it to be positive, really did not think it was going to be positive. Cuz my mother was the only one that had it, and we didn’t know if she had the gene or not, it was just, it was way back. So, I was tested and I found out, actually my husband is the one who told me because the doctor called and talked to him. Well my husband is a doctor and they were colleagues, but it’s not the way to find out. So, I wasn’t really prepared. So that was the extent of my genetic counseling, which was none.”

(Julie 71, BRCA1 positive, no cancer)

“I wasn’t tested for a long time and then finally a doctor [surgeon] said, you know maybe you should be tested and so I didn’t have genetic counseling which was a huge mistake. My doctor just did the test and then I never expected it to be positive, really did not think it was going to be positive. Cuz my mother was the only one that had it, and we didn’t know if she had the gene or not, it was just, it was way back. So, I was tested and I found out, actually my husband is the one who told me because the doctor called and talked to him. Well my husband is a doctor and they were colleagues, but it’s not the way to find out. So, I wasn’t really prepared. So that was the extent of my genetic counseling, which was none.”

(Lillian, 48, BRCA negative, breast cancer at 42)

“I was offered [genetic testing] when I was first diagnosed in 2008. She [gynecological oncologist] asked me some questions like you did about my family history but that was the extent of it and it came back negative.”

(Stacey, 77, BRCA negative, ovarian cancer at 67)

“I think that my doctor did it as part of my first surgery testing. In other words, I think he took blood during my, they didn't take blood in the office. I always bad to go to a lab for my CA125, I'm sorry I honestly don't remember. I know my oncologist, my gynecologist oncologist I said, he wanted to do it and it may have been when they were drawing blood to test me before chemo, or he may have taken blood at the hospital I don't remember. I remember him mentioning it to me as part of surgery. I only had BRCA1 and 2 testing, and they found a specific anomaly.”

(Lynne, 60, BRCA2+, ovarian cancer at 64)

“I knew that I needed to do it [genetic testing] just because of the type of breast cancer she [mother] had, but I think it was around the time that my aunt M got diagnosed, I started thinking about it again and I called the doctor at one point, not meaning to bring it up again, I kind of called just because I wanted to keep in touch with her and at least let her know I was thinking about her, but she kind of brought it up. She told me that they had done the test for her and she was negative, even though she had it, she was negative for the genetic indicators. But that kind of gave me more of the push to do it, talking to her. So I wanna say it was probably within a month or two of talking to her after her diagnosis that I just brought it up at my next OBGYN appointment. I just had a checkup and I mentioned it, that it was something I wanted to talk about and find out what the next steps were, if I wanted to pursue it. And they were like we can do here, right now today, and just take your blood, we'll send it off to this lab, we'll have it back to you in like 30 days. So, it was really easy. I didn't have to make a separate appointment for it or anything.

[did they talk to you about anything specific?]
I don’t know if they take a different approach to it based on my age and the fact that a lot of people who do the genetic testing have a current diagnosis, so I think they were a little more, I don’t want to say light-hearted about it, but they [OBGYN] were a little more like, oh we’ll send it off to this lab, they’ll do XYX testing, and you’ll get your results in a month. They didn’t really talk too much about what happens if its positive. I don’t want to say they weren’t concerned about it but I think it…I’m not sure, they didn’t really cover too much of what the details, they said I would get a packet from the lab that
was doing the test explaining the results and everything, so. It was kind of just an in and out thing with that part of it, and I don't know if it's because I tacked it onto a checkup and they hadn't planned on doing it but, if they mentioned anything about if it were positive, then they were just kind of like oh well if it's positive we'll talk about options then. I think if they said anything that was probably the extent of it because I don't remember a particularly long discussion about the test. [She received a packet in the mail in conjunction with the discussion of her results]

The packet just has information on what the test results mean, just kind of said what the indicators were and why you should get tested which I thought was a little after the fact, like well I already did the test that's why I got this packet which doesn't make sense. I think it was one of the nurses at my OB/GYN office that called and told me that my test results were back and that I was negative and asked me if I had received the packet and I was like yeah, and she was like okay. I was kind of, I don't want to say they were cavalier about it, but it was all very like…I don't know. It all just seemed kind of quick and just like, okay well we're done with this. And again, I don't know if it's that different attitude because I requested the testing and maybe they're not used to that, I don't know if it's just procedural for them.”

(Leigh, 27, BRCA negative, no cancer)

“I want to say there was paperwork I think that came along with it that talked about what genetic testing was, like she [breast surgeon] gave me the information to read. She talked to me about it, gave me the information to read and then she left the room while I read it so I could make the decision. She made it clear to me that I didn't have to make that choice but it was confidential, and I think I did have that little bit of fear of, would this be used against me with my insurance? And I believe the paperwork talked about how that couldn't be shared with insurance companies or it couldn't deny me insurance coverage. I wanna say that was in the paperwork but it was such a mind-blowing day. And then, I don't remember waffling, I just remember yeah, I'm gonna do this. And I wanna say the charge, I can't remember if my insurance covered it or not, I think I had to fight to have them cover it, I want to say it was 3-4k dollars. I think I ended up paying the 80% of it but I had to fight them to do it.”

(Kristina, 60, BRCA negative, ovarian cancer at 21, breast cancer at 55)

“It was in his [gynecologist's] office and they agreed to do it, or have it done there and like I said, it was a saliva test and then the results came back to me that everything was normal, that I didn't carry the BRCA gene. And I don't know if that's all they were testing for? I guess, I'm not up on all, I should be, but I'm not. I was tested probably four years ago, 2014. They just mailed the results. It was just information, and then I think on my next visit my gynecologist mentioned it to me that the testing came back negative.”

(Diane, 71, BRCA negative, no cancer)

“My doctor [gynecological oncologist] that operated on me at the cancer institute, he did it then. They did it, I guess when they operated on me, I didn't even know they did it until after I had the surgery. I just remember Dr. K told me I was BRCA positive. He said that because I had a daughter it would be important to her.”

(Gail, 71, BRCA+, breast cancer at 42, ovarian cancer at 67)

“My first visit with the gyn-onc I mentioned, I don't know if you're familiar with Hadassah hospital in Israel? I belong to Hadassah here where we, obviously its educational and activism for women health issues, but we also raise money to support Hadassah Hospital in Israel and all of their functions. I don't know why but I somehow mentioned Hadassah in my very first visit and the doctor right then and there boy she was really tuned in, I guess she couldn't ask my religion? So she asked me if I knew any Ashkenazi Jews and I told her I was one and she said well once we get you started let's talk about genetic counseling if you are interested. And of course I was so. I think like my second or third visit with her we filled out the paperwork. And I'm sure within a month I knew that I was not a BRCA carrier."

(Kristy, 71, BRCA negative, ovarian cancer at 69)
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<th>Code</th>
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<tr>
<td>Genetic counseling</td>
<td>“I guess I’m kind of a person that just kind of accepts what’s going on, like I really didn’t know that, you know I knew people have BRCA genes, but I didn’t know it would be my heritage and so…I forget the woman’s first name, something Silverman, and she was very good and said you know, if you’re worried or you’re depressed, and I’m like I just want this over with just get everything out and I’m done.” AND “I think they wanted me to meet with them more than I felt I needed it, so I said ehhh I’m really pretty good, I don’t need counseling, like mental counseling, cuz I’m okay with it, I got it, I’ll deal with it.” AND “I didn’t really need much because I wasn’t depressed, I was just ready to do whatever I had to do.” (Lisa, 66, BRCA2+, breast cancer at 55)</td>
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<td>(with GP)</td>
<td>“Me, my mother, and my dad went [to the genetic counseling appointment]. They were really freaking out about it, but I wasn’t at all. I thought, there’s no way we’re positive. I understood why they wanted to do it and I thought it was a good idea. And at the time it was only one group doing the testing and they were really expensive, it was like $3000 for the test at the time.” (Debra, 62, BRCA negative, ovarian cancer at 34)</td>
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<td>“I went in and I guess, the actual experience of it was… I guess it was surprisingly uninvasive and simple, I don’t even really recall the details of that day. There was like a minor, physical exam and then whatever else happened after that I don’t know. I was surprised by the nature of the follow up. Going into discuss the results, it struck me as very, call-centerish? The person was taking me through this flip notebook with all these tabs, and it seemed as though, he was trying to be very delicate in terms of talking about probability and what various things could possible mean and that nothing is a guarantee, and it seemed like he was gonna say the same thing to anybody who came back with my particular result regardless of who I was. And it felt very impersonal, and very scripted. I was surprised by that. I don’t know why I was surprised by that. But I was moved by that. There didn’t seem to be any recognition of what I already knew, or what I was capable of knowing, there was just a lack of recognition of me. And I was pretty surprised that anything that was revealed in that follow up I could have read online, or been emailed, or…there was really no necessity to that follow up, I didn’t learn anything there other than the DNA sequencing or whatever suggested I hadn’t been a recipient of that genetic mutation, and I was like well that probably could have been handled in a very different way just in terms of like, what they were considering the patient education. It didn’t really feel to me like, educational.” [more in GC code excerpts] (Courtney 37, unknown VUS, no cancer)</td>
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<td>“She [genetic counselor] explained what the BRCA meant, and the HER2, and likelihood of recurrence if you’re positive and that kind of stuff. I talked more with Dr. [surgeon] about that stuff and Dr. [oncologist] I talked to her about it. But I think Dr. [surgeon] explained most of it to me because I was like, who knows what they’re talking about.” AND “Yeah, I have no idea how genetics works, it doesn’t matter to me, I’m a very simple person, ya know, so it’s like…we’d like you to do this, and I just agreed. They didn’t explain but they did say it would be higher risk if you’re certain ethnicities or that kind of stuff but that’s about all we discussed. I didn’t ask them any questions, because I know they were just drawing my blood and testing it from there ya know. And they’re usually rushing you from one appointment to another so its like, holy cow. I think I had a bone density scan that day too so it was just like chaos. I used to be there about 12 hours and I was like get me out of this hospital.” (Helen, 43, BRCA negative, breast cancer at 40)</td>
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<td>“At some point I thought I really ought to get the genetic testing but I also thought, I’m really doing fine. And they are looking closely at my breasts and my sisters and I have dense breasts and we’re always...”</td>
<td>(Name, age, genetic testing status, cancer diagnosis)</td>
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laughing about that, sometimes they do extra imaging on us, or extra feeling, because they have dense breasts. So, I didn't really think that I needed the BRCA testing but then as I was in touch with, in these advocacy groups with these other women I realized that it's kind of more standard now, so I just asked my doctor and he said if you want it go ahead and get it. He said, if you have any doubts or concerns at all you need to get them settled and go ahead. And he set it up for me, at Moffitt. I just went and had it done, it was no big deal and I didn't expect it to come back positive and it didn't. I didn't have that genetic testing until maybe 2015?”

(Jill, 64, BRCA negative, ovarian cancer at 51)

“I mean it was, it was a lot of questioning, it was probably hour and a half, two hours I spent out there going through all that, was told even after I’d had it done that I should have actually taken those results and gone back out there and had them review it with me but, that didn't happen.”

(Megan, 59, BRCA VUS, ovarian cancer at 39)

“The woman that I spoke to was lovely and very helpful and if I had had a bunch more questions I'm sure she would have, it was very informative, I just don't remember thinking it was anything in particular that I hadn't already sort of been prepared for.”

(Jill, 64, BRCA negative, ovarian cancer at 51)

“I thought it was real interesting, she gave us some information. The one thing that sticks out in my mind is the German Jews, Ashkenazi Jews, I found that interesting, I don't think that’s me but. I dunno, it was just nice to have a professional tell me instead of the internet.”

(Lisa, 66, BRCA2+, breast cancer at 55)

“The genetic counselors were explaining if I was positive that meant that, it had implications for my daughter and my granddaughter and nieces in terms of how often they were monitored for this type of disease. And later I found out that also knowing whether you’re BRCA1 and BRCA2 is helpful to know if you have a reoccurrence of ovarian cancer and what your treatment would be, so that's just good information to know since ovarian cancer often does reoccur. I think it was just helpful to know and to be able to share with my daughter the recommendations they had about how often my daughter should be checked for breast cancer and stuff like that, to give her that type of feedback. I was doing this testing in the midst of doing chemo, so I was more focused on getting through chemo and staying alive! I was pretty humbled by the statistic that 20% of women who are diagnosed with ovarian cancer die within a year.

(Melanie, 60, BRCA negative, breast cancer at 42, ovarian cancer at 56)

“They discussed family history and again since mine was incomplete they felt it was even more important that I have the testing. And I also knew at that time that if I tested positive there were trails for PARP inhibitors that I could then participate in. I went and I spoke to them and they gave me vials I had to spit, and then it takes a while to come back, and it came back and my daughter was in town so we both went because it would affect her as well, and it came back negative for BRCA1 and BRCA2 and they said there was some more tests being run but they would take longer, so I would say maybe 3 or 4 weeks later I went back and they said everything came back and they said everything came back, that there was nothing that they could see. I found it interesting and I found it informative, I found it, I think it’s a really, really important thing. So, I think just, when I went there initially and before, when they explained everything to me, I thought that was very helpful. Maybe not for me but I understand it better than I… it was just very informative, that’s all.”

(Angie, 70, BRCA negative, ovarian cancer at 66)

“The thing I remember the most about the genetic counselor was she was very nice, I remember the room that we were sitting in, it was like a small conference room, a small table, it was just the two of us. And she had the papers in front of her and she had handed me a few things and I was reading. I was trying to listen as much as I could to understand, and even really understand what questions to ask, but what I ended up hearing was that, both of these are negative, and then she…it sort of felt like…I mean genetics is a really complex topic, I mean there’s a lot, its science, I mean it’s very complex and much more than the average layperson would understand, so I do understand why, I think there’s certain things she had to say, and there’s certain ways she had to explain things, but it was… I wish I could
have been able to digest it and then go back for a second appointment and ask more questions. Cuz I know that they were negative, I know that I was negative for BRCA 1 and 2, and my risk was very low for ovarian cancer, especially without family history, but past that, for that half hour, 45 min or hour or I don't even remember, whatever we were in there…I don't know that I took much more out of it.’”

AND

“With all the things that she [genetic counselor] said, and I felt like…I was thrilled that she was telling me all this information, the more information the better, but genetics, like any science or medicine, is super complicated, and I think it’s very difficult to explain to the average person who doesn’t know much about it, and I don’t know much about science, the only time I was interested in science was when we were talking about genetics. That part actually interested me, but not enough for me to actually go into science.

So, I was fascinated with what she was saying, but so much of it was, there was no context for it really. I don't have a box to put this in, I don't have filter to run this through to translate it into my words, or how to make it apply to my life or my body. And that was where I would like to have, in retrospect, gone through that meeting, listened to her, and then just had another meeting scheduled, not me having…I'm sure she said, let me know if you have any questions, here’s my card, let me know if you have any follow up questions. Well, I don’t even know! You know, I don’t even know what to ask, I don’t even know if this is a legitimate question, did you answer this the first time? I don’t even know. So, I almost wish that it included two appointments, one where she goes through everything and then another one where you come back and, okay now I’ve thought about this, I've done a little bit of research, I’ve talked to a few people, I have a couple questions, what about this…I think that would have been helpful.

(April, 39, BRCA negative, breast cancer 31)

“From what I remember, I think it was statistically when you have this gene your chances of getting cancer by this age have increased by whatever percentage. I’m trying to remember…I honestly don’t remember, and I'll have to think about this, and of course I can’t ask my sister because she’s gone, I can’t remember if they were…not offering but perhaps suggesting prophylactic surgeries or giving it out there as an option, because this was so early that I don’t even know that insurance companies were necessarily paying for it at that point. I think now it’s much more common that insurance would realize hey, it’s a lot surgery for me to do this surgery than to pay for chemo. I don’t remember them pushing those kind of options, I do remember them talking about well, here’s knowledge that you have this gene and you have these increased chances of getting these cancers. When I had my ovaries out, what was explained to me with that prophylactic surgery was that once you were decreasing the amount of estrogen when you remove your ovaries, although I was doing a bioidentical hormone replaces, but because I had so much less estrogen that would decrease my risk for breast cancer because my breasts were not exposed to as much estrogen. So prophylactically removing my ovaries was supposed to reduce my risk of breast cancer, which didn’t seem to really help but okay.”

AND

“And they had statistics, but I don’t know that it's the same as what they have today because obviously today they could tell you people who have breast cancer what percent are genetic whereas back then it was only a certain number of people were even getting genetic testing. So I think there weren’t as many figures, it wasn't as clear cut maybe? You were told of an increased chance and you were given numbers but…I don't know.”

(Gwen, 53, BRCA1+, breast cancer at 45)

“Yes she [mother] went to the same genetic counselor that my aunt went to, her sister who is now deceased. My parents live in Connecticut, I live in North Carolina. So my mom came down and she agreed to be tested so I brought her to where my aunt was receiving care which is the other healthcare system here in Charlotte, and she was tested and they did the family tree while we were sitting there, I went to the appointment with her, and they took blood and then a couple weeks later the test came back that she was positive for it. And the geneticist was, when I talked to the geneticist because I had permission to talk to her, was shocked that my mom was positive because they don’t have any research on people like my mother who have reached her age and have not developed any type of cancer.”

AND

“I saw a genetic counselor and she took blood and she sent it out, they sent it to the same lab. And then mine came back positive. And we knew, my doctor was positive it would come back positive. So when the results came back I went in for the appointment and she talked all about the chances…see

(Gwen, 53, BRCA1+, breast cancer at 45)
even though I’m BRCA positive, I really should’ve theoretically speaking developed breast cancer not ovarian cancer, because with BRCA2 you’re more predisposed, your percentages of developing breast cancer are higher than ovarian cancer even though your risk increases for ovarian cancer too. So they did suggest that I be monitored closely for breast cancer. I’m 80% more likely to get breast cancer with this gene.”

AND

“The information was useful, but I already knew it because I’d been through the counseling with my mother, so nothing was a surprise to me. And I had researched it on my own as well, so nothing was surprising or new.”

(Yvonne, 49, BRCA2+, ovarian cancer at 46)

“I feel like it’s research…so because it’s research I think, we don’t know what we don’t know. I look at research and say okay, collect all the information you want. You’re not gonna get a chance to talk to me again you want to do it now, fine. So I didn’t feel like any of it was a waste. Because I’m in a university setting, because exciting new things happen all the time and I know that, there wasn’t any part of it that felt wasted.”

(Clara, 57, BRCA negative, ovarian cancer at 54)

“So we have a geneticist here at the clinic, she was just like in her 2nd year on staff and so I went to talk to her and we did, like you did, the family tree and she told me about all the…the spiel was, to me, it was the spiel you would give to someone who had never had, or never battled cancer. To me it was kind of…she had to tell me all the facts and, you know, she gave me the sheet with all those little DNA markers that are flawed, you know? I looked at it and we talked about it, and it was kind of…okay, now I’m gonna go on with living.”

(Roxanne, 61, BRCA1+, ovarian cancer at 52)
Appendix D. Institutional Review Board documents.

D1. Institutional Review Board approval.

April 25, 2017

Dana Ketcher
H Lee Moffitt Cancer Center
12902 Magnolia Drive
MRC-CANCONT-KETCHER
Tampa, FL 33612

RE: Expedited Approval for Initial Review
IRB#: Pro00030472
Title: Attitudes, experiences, and perceptions of cancer genetic testing: Assessing women at high genetic risk

Study Approval Period: 4/25/2017 to 4/25/2018

Dear Ms. Ketcher:

On 4/25/2017, the Institutional Review Board (IRB) reviewed and APPROVED the above application and all documents contained within, including those outlined below.

Approved Item(s):
Protocol Document(s):
IRB study protocol 4.4.2017 Version 1.docx

Consent/Assent Document(s)*:
Adult Min Risk ICF.docx.pdf

*Please use only the official IRB stamped informed consent/assent document(s) found under the "Attachments" tab. Please note, these consent/assent documents are valid until the consent document is amended and approved.

It was the determination of the IRB that your study qualified for expedited review which includes activities that (1) present no more than minimal risk to human subjects, and (2) involve only procedures listed in one or more of the categories outlined below. The IRB may review
research through the expedited review procedure authorized by 45CFR46.110. The research proposed in this study is categorized under the following expedited review category:

(6) Collection of data from voice, video, digital, or image recordings made for research purposes.

(7) Research on individual or group characteristics or behavior (including, but not limited to, research on perception, cognition, motivation, identity, language, communication, cultural beliefs or practices, and social behavior) or research employing survey, interview, oral history, focus group, program evaluation, human factors evaluation, or quality assurance methodologies.

As the principal investigator of this study, it is your responsibility to conduct this study in accordance with IRB policies and procedures and as approved by the IRB. Any changes to the approved research must be submitted to the IRB for review and approval via an amendment. Additionally, all unanticipated problems must be reported to the USF IRB within five (5) calendar days.

We appreciate your dedication to the ethical conduct of human subject research at the University of South Florida and your continued commitment to human research protections. If you have any questions regarding this matter, please call 813-974-5638.

Sincerely,

[Signature]

Kristen Salomon, Ph.D., Vice Chairperson
USF Institutional Review Board
Informed Consent to Participate in Research Involving Minimal Risk

Pro # Pro00030472

You are being asked to take part in a research study. Research studies include only people who choose to take part. This document is called an informed consent form. Please read this information carefully and take your time making your decision. Ask the researcher or study staff to discuss this consent form with you, please ask him/her to explain any words or information you do not clearly understand. The nature of the study, risks, inconveniences, discomforts, and other important information about the study are listed below.

We are asking you to take part in a research study called:

**Attitudes, experiences, and perceptions of cancer genetic testing: Assessing women at high genetic risk**

The person who is in charge of this research study is Dana Ketcher. This person is called the Principal Investigator. However, other research staff may be involved and can act on behalf of the person in charge. She is being guided in this research by Dr. Linda Whiteford, her faculty advisor.

**Purpose of the study**

The purpose of this study is to understand how women at high genetic risk for cancer view and/or have experienced genetic testing and counseling for cancer syndromes.

**Why are you being asked to take part?**

We are asking you to take part in this research study because you meet criteria for genetic high risk individuals.

**Study Procedures:**

If you take part in this study, you will be asked to:

- Participate in one audio-recorded interview, at a place and time you deem convenient, that will ask for your opinions and knowledge regarding genetic testing and counseling for cancer syndromes. Recording is voluntary;
- Participation is expected to last about 30-45 minutes;
- Audio-recorded interviews will only be available to the principal investigator and faculty advisor, and kept on a password protected laptop. Recordings are kept for 5 years after the
Final Report is submitted to IRB, per IRB guidelines. After this time, recordings will be deleted.

**Total Number of Participants**
About 25 individuals will take part in this study.

**Alternatives / Voluntary Participation / Withdrawal**
You do not have to participate in this research study.
You should only take part in this study if you want to volunteer. You should not feel that there is any pressure to take part in the study. You are free to participate in this research or withdraw at any time. There will be no penalty or loss of benefits you are entitled to receive if you stop taking part in this study.

**Benefits**
We are unsure if you will receive any benefits by taking part in this research study. However, your responses, combined with those of other participants, may help to improve our overall understanding of genetic testing and counseling for hereditary cancer syndromes.

**Risks or Discomfort**
This research is considered to be minimal risk. That means that the risks associated with this study are the same as what you face every day. There are no known additional risks to those who take part in this study.

**Compensation**
You will receive no payment or other compensation for taking part in this study.

**Costs**
It will not cost you anything to take part in the study.

**Conflict of Interest Statement**
There are no conflicts of interest.

**Privacy and Confidentiality**
We will keep your study records private and confidential. Certain people may need to see your study records. Anyone who looks at your records must keep them confidential. These individuals include:
- The research team, including the Principal Investigator, and faculty advisor.
- Certain government and university people who need to know more about the study, and individuals who provide oversight to ensure that we are doing the study in the right way.
- Any agency of the federal, state, or local government that regulates this research.
- The USF Institutional Review Board (IRB) and related staff who have oversight responsibilities for this study, including staff in USF Research Integrity and Compliance.
We may publish what we learn from this study. If we do, we will not include your name. We will not publish anything that would let people know who you are.

**You can get the answers to your questions, concerns, or complaints**

If you have any questions, concerns or complaints about this study, or experience an unanticipated problem, call Dana Ketcher at 515-988-0224 or email at d.ketcher@mail.usf.edu.

If you have questions about your rights as a participant in this study, or have complaints, concerns or issues you want to discuss with someone outside the research, call the USF IRB at (813) 974-5638 or contact by email at RSCH-IRB@usf.edu.

**Consent to Take Part in this Research Study**

I freely give my consent to take part in this study. I understand that by signing this form I am agreeing to take part in research. I have received a copy of this form to take with me.

Signature of Person Taking Part in Study ___________ Date ___________

Printed Name of Person Taking Part in Study ______________________

**Statement of Person Obtaining Informed Consent**

I have carefully explained to the person taking part in the study what he or she can expect from their participation. I confirm that this research subject speaks the language that was used to explain this research and is receiving an informed consent form in their primary language. This research subject has provided legally effective informed consent.

Signature of Person obtaining Informed Consent ___________ Date ___________

Printed Name of Person Obtaining Informed Consent _______________________