Spring 2021

Previvor and High-Risk Breast Cancer Patients' Opinions on A Specialized Management Clinic

Madeleine Nicole Tjoelker

PREVIVOR AND HIGH-RISK BREAST CANCER PATIENTS' OPINIONS ON A SPECIALIZED MANAGEMENT CLINIC

by

Madeleine Nicole Tjoelker

Bachelor of Science

Trinity Christian College, 2018

Submitted in Partial Fulfillment of the Requirements

For the Degree of Master of Science in

Genetic Counseling

School of Medicine

University of South Carolina

2021

Accepted by:

Whitney Dobek, Director of Thesis

Julian Kim, Reader

Gail Stapleton, Reader

LeAnn Perkins, Reader

Tracey L. Weldon, Interim Vice Provost and Dean of the Graduate School

DEDICATION

This project is dedicated to my loved ones who have battled cancer and shown me what true courage looks like. You are an inspiration for all who know you.

ACKNOWLEDGMENTS

This project would not have been possible without the unceasing support of my thesis committee: Whitney Dobek, LeAnn Perkins, Gail Stapleton, and Julian Kim. I am so grateful to each one of you for your insight, your guidance, and your encouragement. I'd also like to thank Amy Wardyn for her leadership over the thesis process and for providing assistance and advice along this journey. I'd especially like to thank Janice Edwards and the faculty of the University of South Carolina Genetic Counseling Program for shaping me as a genetic counselor and igniting my passion for previvorship and cancer genetics. I'd like to thank my classmates and friends for walking alongside me and giving me two years I'll always remember. And a special thank you to Dacia Lipkea and Taylor Kupneski for their friendship and support, both in the classroom and out. Lastly, I'd like to thank my family for their unwavering support and encouragement as I followed my dream of becoming a genetic counselor. I could not have made it to where I am without your love and support.

ABSTRACT

Approximately 5-10% of cancers are thought to be hereditary, caused by pathogenic variants in genes associated with inherited cancer syndromes. *Previvors*, individuals who have a higher predisposition to cancer due to genetic or other risk factors, have specific healthcare and psychological needs that may be better served by a specialized management clinic. This study compared the experiences of previvors who had access to a specialized management clinic with those who did not, in order to better understand the unique needs of previvors. This study utilized a mixed methods design including an online survey (N=26) and semi-structured phone interview (N=6). Overall, previvors with access to a specialized management expressed a reduction in stress level (N=5), expedited necessary medical care (N=2), access to a simplified clinical process (N=5), and provision of information needed to make informed decisions regarding their medical care (N=8). Previvors who did not have access to a specialty clinic described challenges with finding information about their risk (N=4), receiving care from general practitioners (N=3), and having questions unanswered by healthcare providers (N=6), further supporting previous literature that investigated the nuanced care required by previvors. Previvors without access to a management clinic desired a team of specialists familiar with genetics, a forum to ask questions, and a clinic that would ensure their care meets the current recommendations. This study demonstrates the need for specialized management clinics designed with previvors' needs in mind in order to provide these patients with the most appropriate care.

TABLE OF CONTENTS

LIST OF TABLES

LIST OF FIGURES

CHAPTER 1: BACKGROUND AND LITERATURE REVIEW

1.1 Hereditary Cancer

Approximately 5-10% of cancers are thought to be hereditary. Hereditary cancer is caused by mutations in genes associated with hereditary cancer syndromes, and a pathogenic mutation results in a significantly increased risk for cancer development compared to that of the average population (Senter & Hatfield, 2016). Mutations that cause hereditary cancer syndromes are identified through the use of genetic testing, specifically testing that targets oncogenes and tumor suppressor genes, genes associated with regulation of cell growth and hereditary cancer (Dekanek et al., 2019). Once a pathogenic mutation has been identified, it is recommended that the patient pursue management specific to their cancer risks, often as directed by the guidelines established by the National Comprehensive Cancer Network (NCCN), a society recognized in the medical community to be an authority on cancer care (NCCN, 2020).

There are a large number of genes associated with hereditary cancer. This study encompassed individuals with mutations in *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN*, and *TP53.* While these genes are mainly associated with breast cancer, mutations in them also carry other cancer risks. In addition, these genes have specific management recommendations established by NCCN that aid in patient care. The cancer risks for each gene are shown in Table 1.1 below (adapted from GeneReviews (2016); National Comprehensive Cancer Network (NCCN), 2020).

Table 1.1 High-Risk Breast Cancer Genes and Associated Cancer Risks

Gene	Cancer Risks
ATM	Breast (15-40%) Pancreatic (5-10%) Ovarian (<3%) Prostate
BRCA1	Breast, with predisposition to triple negative disease (>60%) Ovarian (39-58%) Male breast Prostate Pancreatic (≤ 5%)
BRCA2	Breast, with predisposition to ER+ disease (>60%) Ovarian (13-29%) Male breast Prostate Pancreatic (5-10%) Melanoma (elevated)
CDH1	Breast, with predisposition to lobular disease (41-60%) Diffuse gastric
CHEK2	Breast, with predisposition to ER+ disease (15-40%) Colon Prostate Stomach Sarcoma Kidney
PALB2	Breast (41-60%) Ovarian (3-5%) Male breast Pancreatic (5-10%)
PTEN	Breast (40-60%, may be >60%) Thyroid Renal cell Endometrial Colorectal
TP53	Breast (>60%) Pancreatic (5-10%) Soft tissue sarcoma Osteosarcoma Brain tumors Adrenocortical carcinoma Leukemia

1.2 Genetic Testing Recommendations for Hereditary Cancer

Genetic testing for hereditary cancer syndromes is recommended for individuals who meet certain criteria as determined by NCCN. One example of this criteria is the NCCN Testing Criteria for High-Penetrance Breast and/or Ovarian Cancer Susceptibility Genes. If criteria are met and no known familial mutation has been identified, providers should consider comprehensive testing for the patient with a multi-gene panel (NCCN, 2020). NCCN (2020) recommends genetic testing for hereditary breast and ovarian cancer for the following indications, listed in Table 1.2 below (adapted from NCCN, 2020).

Table 1.2 NCCN Testing Criteria for High-Penetrance Breast/Ovarian Cancer Susceptibility Genes

NCCN Testing Criteria for High-Penetrance Breast/Ovarian Cancer Susceptibility Genes
1. Any blood relative with a pathogenic/likely pathogenic variant in a cancer susceptibility gene
2. Personal history of breast cancer under age 45 or age 46-50 with a second breast cancer at any age or at least one close relative with breast, ovarian, pancreatic, or prostate cancer at any age
3. Personal history of triple negative breast cancer at age 60 or younger
4. Personal history of breast cancer at any age with Ashkenazi Jewish ancestry or a close relative with breast cancer under age 50, ovarian, pancreatic, or metastatic prostate cancer at any age
5. Diagnosis of male breast cancer at any age
6. Diagnosis of epithelial ovarian cancer (including fallopian tube cancer or peritoneal cancer) at any age
7. Diagnosis of exocrine pancreatic cancer at any age
8. Diagnosis of prostate cancer at any age with metastatic, intraductal/cribriform histology, or high- or very-high-risk group
9. Diagnosis of prostate cancer of any NCCN risk group with Ashkenazi Jewish ancestry, one or more close relatives with breast cancer under age 50, ovarian, pancreatic, or metastatic, or intraductal/cribriform prostate cancer at any age, or 2 or more close relatives with either breast or prostate cancer (any grade) at any age
10. A mutation was identified on tumor genomic testing that has clinical implications if identified in the germline
11. Meets Li-Fraumeni Syndrome testing criteria, Cowden syndrome/PTEN hamartoma syndrome testing criteria

12. To aid in systemic therapy decision-making, such as for HER2-negative breast cancer
13. An affected or unaffected individual with a first- or second- degree relative meeting any of the above
14. An affected or unaffected individual who otherwise does not meet the criteria above but has a probability >5% of a *BRCA1/2* pathogenic variant based on prior probability models (e.g., Tyrer-Cuzick, BRCAPro, Can Risk)

1.3 Previvorship

By meeting the first or thirteenth criterion described above, and pursing testing for a known familial variant or because of a family history of cancer, individuals are learning of their *previvor* status (NCCN, 2020). The term *previvor* was initially coined by the advocacy group, Facing Our Risk of Cancer Empowered (FORCE), and is used to describe individuals "who have a much greater predisposition to cancer than individuals in the general population but who have not yet developed the disease" (Carvalho et al., 2019, p. 1). Increase in the identification of previvors can be attributed to increased interest in genetic testing, increased testing of ovarian cancer patients who may be candidates for PARP (poly ADP-ribose polymerase) inhibitor therapies, and the identification of relatives of these patients who may be at risk for carrying these mutations (Carvalho et al., 2019). This greater susceptibility may be due to the presence of a pathogenic mutation in a hereditary cancer gene, increasing the risk for cancer, or could be caused by other factors including family history (Getachew-Smith et al., 2019). It has been recognized that individuals falling within the previvor category have "specific psychosocial and healthcare needs…to help them decide how to manage this substantial risk" (Mahon, 2014, p. 21).

Getachew-Smith et al. (2019) studied patients' perceptions of the term *previvor* and whether or not that identity resonated with them. Although FORCE established a

definition for what constitutes a previvor, individuals who fall into that category had two main distinctions in their definition. One group identified previvors as those with a positive genetic test result, while the other group only considered previvors to be those that have undergone some form of risk reducing surgery, either mastectomy or oophorectomy. When assessing whether or not individuals accepted the term, they found that the majority accepted the label, claiming that the term previvor gave them a sense of community and validated their experience. However, some rejected the term due to its similarity to the word "survivor," and felt that perhaps the label diminished the experience of cancer survivors. Some also felt that the term invoked fear, making it sound as if cancer was inevitable (Getachew-Smith et al., 2019). Although there is no clear consensus on the use of the term *previvor*, it is clear individuals within this category have unique challenges.

1.4 Genetics-Based Management

Individuals with mutations in hereditary cancer genes have specific management recommendations established by NCCN, but recommendations differ slightly from gene to gene. In general, breast cancer screenings begin earlier than for women of average risk, and for some mutations, a risk-reducing surgery, such as a mastectomy or salpingo-oophorectomy, may be considered. Because cancer risks vary from gene to gene, it is important to know the patient's carrier status when determining a management plan. Table 1.3 outlines recommendations for each gene, adapted from the NCCN guidelines (NCCN, 2019).

Table 1.3 NCCN Management Recommendations for Hereditary Cancer Predisposition

Gene	Management Recommendations
ATM	Annual mammogram, consider breast MRI at age 40 Risk-reducing mastectomy or salpingo-oophorectomy based on family history Pancreatic cancer screening (MRCP and EUS) starting at age 50 if family history of pancreatic cancer
BRCA1	Breast awareness at age 18 Clinical breast exam every 6-12 months, beginning at age 25 Annual breast MRI from age 25 to 29 Annual mammogram with or without tomosynthesis from age 30 to 75 Consider risk-reducing mastectomy Recommend salpingo-oophorectomy, typically between age 35-40
BRCA2	Breast awareness at age 18 Clinical breast exam every 6-12 months, beginning at age 25 Annual breast MRI from age 25 to 29 Annual mammogram with or without tomosynthesis from age 30 to 75 Consider risk-reducing mastectomy Recommend salpingo-oophorectomy, no later than age 40-45
CDH1	Annual mammogram, consider breast MRI at age 30 Prophylactic gastrectomy between ages 18 and 40 and baseline endoscopy Risk-reducing mastectomy based on family history
CHEK2	Annual mammogram, consider breast MRI at age 40 Colonoscopy every 5 years, beginning at age 40, or 10 years prior to first-degree relative's age at diagnosis
PALB2	Annual mammogram, consider breast MRI at age 30 Discuss option of risk-reducing mastectomy Risk-reducing salpingo-oophorectomy based on family history Pancreatic cancer screening (MRCP and EUS) starting at age 50 if family history of pancreatic cancer
PTEN	Breast awareness at age 18 Clinical breast exam every 6-12 months, starting at age 25 or 10 years before diagnosis of breast cancer in family (whichever comes first) Annual mammogram, consider breast MRI at age 30-35 (or 10 years before diagnosis in family) until age 75 Discuss option of risk-reducing mastectomy Consider endometrial biopsy every 1-2 years, starting age 35 Consider hysterectomy upon completion of childbearing Annual physical exam, starting age 18 (or 5 years prior to first cancer diagnosis) Annual thyroid ultrasound, starting age 7 Colonoscopy starting age 35, every 5 years if negative (start earlier if family history) Consider renal ultrasound at age 40, every 1-2 years Routine dermatology evaluation Consider psychomotor assessment and brain MRI if symptoms present

TP53	Breast awareness at age 18 Clinical breast exam every 6-12 months, starting age 20 Breast MRI age 20-29, MRI and mammogram age 30-75 Discuss risk-reducing mastectomy Physical exam and neurological evaluation in cancer survivors every 6-12 months Colonoscopy and upper endoscopy every 2-5 years, starting age 25 (or earlier if family history) Annual dermatologic evaluation, starting age 18 Annual whole-body MRI Annual brain MRI Pancreatic cancer screening (MRCP and EUS) starting at age 50 if family history of pancreatic cancer

1.5 Management for Patients at High Risk of Breast Cancer

Some previvors may have a predisposition to cancer not due to a genetic mutation, but rather due to family history or other factors (Getachew-Smith et al., 2019). Several verified risk models exist to determine lifetime risk for breast cancer, including Tyrer Cuzick, Gail, BRCAPRO, Claus, and BOADICEA. These models take into account family history of breast and/or ovarian cancer, age, breast density, reproductive history, hormonal history, genetic testing, and history of abnormal breast imaging or pathology to calculate a lifetime risk for breast cancer (Monticciolo et al., 2018).

Women with a lifetime risk of breast cancer greater than or equal to 20% as determined by these models are recommended to have additional screening, similar to that of individuals with a genetic mutation that increases breast cancer risk (Monticciolo et al., 2018). Table 1.4, below, adapted from NCCN (2021), outlines the recommended management strategies for high-risk individuals:

Table 1.4 NCCN High-Risk Breast Cancer Screening Guidelines

High-Risk Management
Breast awareness
Clinical encounter every 6-12 months (not to be started before age 21)
Annual screening mammogram • 10 years prior to age of youngest breast cancer diagnosis in family • Not to be started before age 30 • Consider tomosynthesis
Recommend annual breast MRI • 10 years prior to age of youngest breast cancer diagnosis in family • Not to be started before age 25
Recommend risk-reducing strategies • Limit alcohol consumption • Increase physical activity • Weight control • Breastfeeding • Consider risk-reducing agents (tamoxifen, raloxifene, aromatase inhibitors)

Tamoxifen is a risk-reducing agent recommended for individuals at an increased risk for breast cancer. NCCN currently recommends women age 35 or older may take 20 milligrams per day for five years and doing so can reduce breast cancer risk up to 49% (NCCN, 2020). For individuals who have a history of atypical hyperplasia, taking tamoxifen may reduce breast cancer risk by up to 89% (NCCN, 2020). Because of its efficacy, NCCN recommends the use of tamoxifen in the reduction of breast cancer risk for women at increased risk (NCCN, 2020).

1.6 Adherence to Management Recommendations

There is little data on how previvors are conducting cancer screening and management or how closely they are following the NCCN screening guidelines. However, in a study by Hesse-Biber and An (2016), researchers looked at surgical decision-making among BRCA-positive individuals. The main factors that went into surgical decision-making included age, parental status, gender of the children, level of

psychological distress from the BRCA result, perceived family support, experienced medical uncertainty, and the level of guilt they felt about the possibility of their children inheriting the mutation. Interestingly, women who experienced more distress related to their BRCA result, those who felt they lacked support from their family, and those who perceived more medical uncertainty were more likely to choose surveillance rather than surgery (Hesse-Biber & An, 2016). While the study looked at factors influencing their management choices, they did not look into how closely surveillance was being followed.

Another study, by Hoskins, Roy, and Greene (2012), analyzed patients' risk perception of their *BRCA1* or *BRCA2* mutation. In their study of 60 participants, 17 had already undergone a risk-reducing mastectomy and eight had an oophorectomy. An additional 34 participants were either considering or concretely planning a risk-reducing mastectomy and 45 were either considering or concretely planning a risk-reducing oophorectomy. The authors concluded that many previvors may initially choose to manage with increased surveillance before electing to proceed with a risk-reducing surgery (Hoskins et al., 2012). Again, this study did not explore what screening these participants were doing in place of risk-reducing surgery, suggesting that further research in this area is needed.

1.7 Psychosocial Needs of Previvors

Individuals with a predisposition to cancer have unique needs compared to the average population. In a study by Dean and Davidson (2018), it was found that these individuals may have increased levels of uncertainty compared to those at average risk for cancer. In fact, researchers found that previvors experience high levels of uncertainty, and increased uncertainty can result in "emotional distress, anxiety, depression, loss of

control, and poor decision making and quality of life" (p. 122). In order to manage this uncertainty, previvors make decisions based on their perceived risk and available information, so it is imperative that these individuals are receiving accurate information and support (Dean & Davidson, 2018).

Furthermore, Mahon (2014) found that, although some online organizations and support groups are available, many of these individuals feel isolated, as if no one else understands what they are going through. They may feel as if they are being labeled or feel "different from those who do not carry a mutation" (Mahon, 2014, p. 22). Many times, support organizations help patients advocate for themselves, an important task as they pursue surveillance and management. Previvors have described their experience with their healthcare providers as overwhelming and exhausting because they have been tasked with teaching their providers about their risk and management instead of receiving the empathy and psychosocial support they need (Dean & Davidson, 2018).

These individuals are even faced with opposition to their decision to select risk-reducing surgery to reduce cancer risk, with the opposing individuals claiming that prophylactic surgery is too extreme. And even if supported in their decision to pursue prophylactic surgery, these individuals face unique challenges as a result of their surgery. Some women, after a risk-reducing mastectomy, have self-image difficulties and lack of security in their identity (Mahon, 2014). After a risk-reducing salpingo-oophorectomy, women may experience altered self-image, increased depression, increased fatigue, sleep deprivation, and sexual dysfunction that can change her desire for intimacy and ultimately affect personal relationships (Alexandre et al., 2017).

Although much of a previvor's uncertainty may arise from the potential to develop cancer, previvors are also concerned about the impact their risk has on other family members. In a study assessing perspectives of young adults at risk to have a BRCA mutation, many participants emphasized the need for information about reproductive issues and family planning (Young et al., 2019). Furthermore, in a study by Dean and Rauscher (2017), it was observed that many women who are previvors use two types of decision-making styles when thinking about family planning: logical and emotional. Logical decision-making involved planning timing for undergoing risk-reducing surgeries and processing the pressure from healthcare providers to receive prophylactic care. Emotional decision-making involved processing her biological time clock, her hopes for the future, guilt associated with children possibly inheriting the mutation, and consideration of pre-implantation genetic diagnosis (Dean & Rauscher, 2017). For example, a woman must weigh the option of breastfeeding her child or reducing her cancer risk with prophylactic mastectomy. In addition, previvors may be worried about the risk to their children and when to communicate that risk (Mahon, 2014).

1.8 Utility of a Specialized Management Clinic

These unique needs of previvors and high-risk breast cancer patients support the necessity of a specialty clinic for previvor management. As described above, management for previvors and high-risk breast cancer patients is complex and variable, thus it is imperative that individuals are managed by healthcare professionals who are knowledgeable about the personalized care required. Studies have shown that obstetrics and gynecology providers (OB/GYN) and family practice providers are not able to give

previvors optimal care, primarily due to a lack of confidence regarding the management recommendations (Dekanek et al., 2019). In fact, in a study of 86 OB/GYN and family practice physicians, only 44% of participants felt somewhat confident in discussing *BRCA* management guidelines, and none of the participants indicated that they felt completely confident (Dekanek et al., 2019).

Other studies have looked at patients' perceived barriers to getting appropriate cancer management. These studies have found that scheduling difficulties are a factor preventing proper adherence to management (Goh & Spigelman, 2020; Young et al., 2019). In addition, confusion surrounding insurance coverage for screening has also been found to be a barrier to accessing these services (Dean et al., 2017). Previvors are in need of healthcare providers who are not only knowledgeable of the management guidelines, but also how to follow through with scheduling and the logistics of following those guidelines.

Although the services included in a previvor clinic will likely vary between clinics, several needs among previvors remain the same. First, because scheduling screenings and appointments is a frequent challenge among previvors, a primary role of this clinic should be to establish a clear appointment plan for patients. A study by Young et al. (2019) of the information needs of previvors found that previvors wanted genetics providers to make referrals to other specialists, such as psychologists, surgeons, or other specialists. Generally, genetic counselors make the initial recommendations for the patient, but rely on the referring provider to make the necessary referrals, potentially causing interruptions in the patient's transition of care. This is largely due to the scope of

practice defined by the National Society of Genetic Counselors, which states that genetic counselors have the authority to:

> "Identify, order, and coordinate genetic laboratory tests and other diagnostic studies as appropriate for the genetic assessment; integrate genetic laboratory test results and other diagnostic studies with personal and family medical history to assess and communicate risk factors for genetic/medical conditions and diseases; and identify and utilize community resources that provide medical, educational, financial, and psychosocial support and advocacy" (National Society of Genetic Counselors, 2021).

Because under the society's scope of practice genetic counselors are unable to refer to outside physicians, this responsibility is placed on either the referring provider or the patient to get connected with these medical specialists.

One potential model for a previvor clinic has been created by the Stefanie Spielman Comprehensive Breast Center in Columbus, Ohio (Senter & Hatfield, 2016). This clinic has high-risk breast cancer patients meet with a cancer genetic counselor first to take a family history and make genetic testing recommendations if warranted. Following the genetic consultation, the patient meets with a nurse practitioner specialized in breast health or a breast, surgical, or medical oncologist who assists in making management recommendations. Many patients receive high-risk breast cancer screening including mammograms, breast MRIs, and clinical evaluation. The clinic model staggers these appointments by six months and alternates visits between the physician and the nurse practitioner. An updated family history is taken at each visit and the cancer genetic counselor may return to see the patient to discuss additional recommendations. Genetic

counselors also serve as a resource for questions regarding genetic testing for family members or other genetics-related considerations. (Senter & Hatfield, 2016). This model addresses the complications of scheduling follow-up appointments for recommended management.

Although many genetic counselors may be unable to serve as full-time staff of a previvor clinic, they play an important role in the care of previvors and should be closely integrated with a specialty clinic. The exact role of the genetic counselor would likely depend on the overall clinic setup as well as the needs of the individual patient. For example, a genetic counseling consult may be requested when a patient is considering family planning decisions, when family history has changed significantly, or when updates to genetic testing have been made. The primary medical staff for a previvor clinic would likely consist of a nurse practitioner and/or physician. The nurse practitioner and/or physician can provide initial screening services and make referrals to necessary specialists including, but not limited to, oncology, plastic surgery, nutritional services, psychological services, and reproductive endocrinology. The establishment of a specialized previvor clinic would allow for unhindered access to resources and support in cancer prevention and management, which has potential to improve outcomes for these individuals (Senter & Hatfield, 2016).

1.9 Rationale of Study

Although there is research into the unique needs of previvors and individuals at an increased risk for cancer, there is little information about previvors' opinions on the establishment of a management clinic in general, and nothing specific for the state of South Carolina. The goal of this study is to determine if patients in Columbia, South

Carolina would benefit from the establishment of a specialized management clinic, and if so, what services these individuals need. It is our belief that this study will bring no harm to the participant but will provide valuable insight into how to best care for this patient population. We will also pull from experiences from a currently established clinic in South Carolina to gather opinions on an already available service.

1.10 Objectives

1. Determine if there is desire and/or need for a previvor clinic in Columbia, South Carolina
2. Determine the utility and efficacy of the Genetics Management Clinic and High-Risk Breast Lifetime Clinic in Greenville, South Carolina
3. Determine what services should be incorporated into such a clinic and how often patients would require these services
4. Assess whether patients who had access to a previvor clinic were better able to adhere to management recommendations compared to those who did not have access

1.11 Hypothesis

It is hypothesized that without clear direction and guidance from knowledgeable health professionals, many previvors may struggle to adhere to NCCN management guidelines. Without proper care, previvors are at risk for worsened health outcomes, so it is expected that this study may improve overall patient satisfaction and care. Furthermore, it is anticipated that patients within the Columbia, South Carolina area will have strong interest for the establishment of a local previvor clinic and that patients who

have attended the specialty clinic in Greenville will experience high patient satisfaction and will attest to the value and benefit of such a clinic.

CHAPTER 2: PREVIVOR AND HIGH-RISK BREAST CANCER PATIENTS' OPINIONS ON A SPECIALIZED MANAGEMENT CLINIC[1]

[1] Tjoelker, M., Dobek, W., Perkins, L., Stapleton, G., & Kim, J. To be submitted to the *Journal of the Advanced Practitioner in Oncology*

2.1 Abstract

Approximately 5-10% of cancers are thought to be hereditary, caused by pathogenic variants in genes associated with inherited cancer syndromes. *Previvors*, individuals who have a higher predisposition to cancer due to genetic or other risk factors, have specific healthcare and psychological needs that may be better served by a specialized management clinic. This study compared the experiences of previvors who had access to a specialized management clinic with those who did not, in order to better understand the unique needs of previvors. This study utilized a mixed methods design including an online survey (N=26) and semi-structured phone interview (N=6). Overall, previvors with access to a specialized management expressed less stress (N=5), less delay in care (N=2), access to a simplified clinical process (N=5), and the information needed to make informed decisions regarding their medical care (N=8). Previvors who did not have access to a specialty clinic described challenges with finding information about their risk, receiving care from general practitioners, and having questions unanswered by healthcare providers, further supporting previous literature that investigated the nuanced care required by previvors. Previvors without access to a management clinic desired a team of specialists familiar with genetics, a forum to ask questions, and a clinic that would ensure their care meets the current recommendations. This study demonstrates the need for specialized management clinics designed with previvors' needs in mind in order to provide these patients with the most appropriate care.

2.2 Introduction

Approximately 5-10% of cancers are thought to be hereditary. Hereditary cancer is caused by mutations in genes associated with hereditary cancer syndromes, and a

pathogenic mutation results in a significantly increased risk for cancer development compared to that of the average population (Senter & Hatfield, 2016). Mutations that cause hereditary cancer syndromes are identified through the use of genetic testing, specifically testing that targets oncogenes and tumor suppressor genes, genes associated with regulation of cell growth and hereditary cancer (Dekanek et al., 2019). Once a pathogenic mutation has been identified, it is recommended that the patient pursue management specific to their cancer risks, often as directed by the guidelines established by the National Comprehensive Cancer Network (NCCN), a society recognized in the medical community to be an authority on cancer care (NCCN, 2020).

There are a large number of genes associated with hereditary cancer. This study encompassed individuals with mutations in *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN*, and *TP53*. While these genes are mainly associated with breast cancer, mutations in them also carry other cancer risks. In addition, these genes have specific management recommendations established by NCCN that aid in patient care (NCCN, 2020).

Genetic testing for hereditary cancer syndromes is recommended for individuals who meet certain criteria as determined by NCCN. One example of this criteria is the Testing Criteria for High-Penetrance Breast and/or Ovarian Cancer Susceptibility Genes, determined by NCCN (NCCN, 2020). If criteria are met, and no familial mutation has been identified, providers should consider comprehensive testing for the patient with a multi-gene panel (NCCN, 2020). NCCN (2020) recommends genetic testing for hereditary breast and ovarian cancer for patients with certain cancer diagnoses or if a family history meets certain criteria.

By pursuing testing for a known familial variant or because of a family history of cancer, individuals are learning of their *previvor* status. The term *previvor* was initially coined by the advocacy group, Facing Our Risk of Cancer Empowered (FORCE), and is used to describe individuals "who have a much greater predisposition to cancer than individuals in the general population but who have not yet developed the disease" (Carvalho et al., 2019, p. 1). This greater susceptibility may be due to the presence of a pathogenic mutation in a hereditary cancer gene or other factors including family history (Getachew-Smith et al., 2019). It has been recognized that individuals falling within the previvor category have "specific psychosocial and healthcare needs…to help them decide how to manage this substantial risk" (Mahon, 2014, p. 21).

Individuals with mutations in hereditary cancer genes have specific management recommendations established by the National Comprehensive Cancer Network, but recommendations differ slightly from gene to gene. Individuals who fall into the previvor category based on family history or other factors can be identified using verified risk models, such as Tyrer-Cuzick, Gail, BRCAPRO, Claus, and BOADICEA. Women with a lifetime risk of breast cancer greater than or equal to 20% as determined by these models are recommended to have additional screening similar to that recommended for individuals with a genetic mutation (Monticciolo et al., 2018). Recommendations include breast awareness, clinical breast exams every six to twelve months, annual breast MRI beginning at age 25, annual mammogram beginning at age 30, and incorporation of risk-reducing strategies, such as the use of tamoxifen as a form of chemoprevention (NCCN, 2020). Because cancer risks vary from gene to gene, it is important to know the patient's carrier status when determining a management plan.

There is little data on how previvors are conducting cancer screening and management or how closely they are following the NCCN screening guidelines. However, in a study by Hesse-Biber and An (2016), researchers looked at surgical decision-making among BRCA-positive individuals. The main factors that went into surgical decision-making included age, parental status, gender of the children, level of psychological distress from the BRCA result, perceived family support, experienced medical uncertainty, and the level of guilt they felt about passing the mutation to their children. Women who experienced more distress related to their BRCA result, those who felt they lacked support from their family, and those who perceived more medical uncertainty were more likely to choose surveillance rather than surgery (Hesse-Biber & An, 2016). Another study, by Hoskins, Roy, and Greene (2012), analyzed patients' risk perception of their *BRCA1* or *BRCA2* mutation. In their study of 60 participants, 17 had already undergone a prophylactic mastectomy and eight had completed an oophorectomy. An additional 34 participants were either considering or concretely planning a risk-reducing mastectomy and 45 were either considering or concretely planning a prophylactic oophorectomy. The authors concluded that many previvors may initially choose to manage with increased surveillance before electing to proceed with prophylactic surgery (Hoskins et al., 2012). Neither study explored previvors' adherence to management recommendations if proceeding with screening surveillance, suggesting that further research in this area is needed.

Individuals with a predisposition to cancer have unique needs compared to the average population. In a study by Dean and Davidson (2018), it was found that these individuals may have increased levels of uncertainty compared to those at average risk

for cancer. In fact, researchers found that previvors experience high levels of uncertainty, and increased uncertainty can result in "emotional distress, anxiety, depression, loss of control, and poor decision making and quality of life" (p. 122). In order to manage this uncertainty, previvors make decisions based on their perceived risk and available information, so it is imperative that these individuals are receiving accurate information and support (Dean & Davidson, 2018). Furthermore, Mahon (2014) found that, although some online organizations and support groups are available, many of these individuals feel isolated, labeled, or feel different from others without a mutation.

Previvors have described their experience with their healthcare providers as overwhelming and exhausting because they have been tasked with teaching their providers about their risk and management instead of receiving the empathy and psychosocial support they need (Dean & Davidson, 2018). These individuals are even faced with opposition to their decision to select risk-reducing surgery to reduce cancer risk, by claims that prophylactic surgery is too extreme. And even if supported in their decision to pursue prophylactic surgery, these individuals face unique challenges as a result of their surgery. Some women, after a risk-reducing mastectomy, have self-image difficulties and lack of security in their identity (Mahon, 2014). After a risk-reducing salpingo-oophorectomy, women may experience altered self-image, increased depression, increased fatigue, sleep deprivation, and sexual dysfunction that can change her desire for intimacy and ultimately affect personal relationships (Alexandre et al., 2017).

Although much of a previvor's uncertainty may arise from the potential to develop cancer, previvors are also concerned about the impact their risk has on other family members. In a study assessing perspectives of young adults at risk to have a

BRCA mutation, many participants emphasized the need for information about reproductive issues and family planning (Young et al., 2019). Individuals who carry a genetic mutation are at risk of their children inheriting this mutation and also have to weigh the option of breastfeeding their child or reducing their cancer risk with risk-reducing mastectomy. In addition, previvors may be worried about the risk to their children and when to communicate about that risk (Mahon, 2014).

These unique needs of previvors and high-risk breast cancer patients support the necessity of a specialty clinic for previvor management. As described above, management for previvors and high-risk breast cancer patients is complex and variable, thus it is imperative that individuals are managed by healthcare professionals who are knowledgeable about the personalized care required. Studies have shown that obstetrics and gynecology (OB/GYN) providers and family practice providers are not able to give previvors optimal care, primarily due to a lack of confidence regarding the management recommendations (Dekanek et al., 2019). In fact, in a study of 86 OB/GYN and family practice physicians, only 44% of participants felt somewhat confident in discussing *BRCA* management guidelines, and none of the participants indicated that they felt completely confident (Dekanek et al., 2019).

Other studies have looked at patients' perceived barriers to getting appropriate cancer management. These studies have found that scheduling difficulties are a factor preventing proper adherence to management (Goh & Spigelman, 2020; Young et al., 2019). In addition, confusion surrounding insurance coverage for screening has been found to be a barrier to accessing these services (Dean et al., 2017). Previvors are in need

of healthcare providers who are not only knowledgeable of what the guidelines are, but also how to follow through with scheduling and logistics of following those guidelines.

Although the services included in a previvor clinic will likely vary from clinic to clinic, several needs among previvors remain the same. First, because scheduling screenings and appointments is a frequent challenge among previvors, a primary role of this clinic should be to establish a clear appointment plan for patients. A study by Young et al. (2019) of the information needs of previvors found that previvors wanted genetics providers to make referrals to other specialists, such as psychologists, surgeons, or other specialists. Generally, genetic counselors make the initial recommendations for the patient, but rely on the referring provider to make the necessary referrals, potentially causing interruptions in the patient's transition of care.

One potential model for a previvor clinic has been created by the Stefanie Spielman Comprehensive Breast Center in Columbus, Ohio (Senter & Hatfield, 2016). This clinic has high-risk breast cancer patients meet with a cancer genetic counselor first to take a family history and make any genetic testing recommendations if warranted. Following the genetic consultation, the patient meets with a nurse practitioner specialized in breast health or a breast, surgical, or medical oncologist who assist in making management recommendations. Many patients receive high-risk breast cancer screening including mammograms, breast MRIs, and clinical evaluation. The clinic model staggers these appointments by six months and alternates visits between the physician and the nurse practitioner. An updated family history is taken at each visit and the cancer genetic counselor may return to see the patient to discuss additional recommendations. Genetic counselors also serve as a resource for questions regarding genetic testing for family

members or other genetics-related considerations (Senter & Hatfield, 2016). The model addresses the issues of scheduling follow-up appointments for management. Clinic organization will vary, as will the role of the genetic counselor's involvement, but patients need access to a provider who can refer to specialists and establish support resources, and a similar model to the one described above would be of great benefit to the patients of Columbia, South Carolina.

The goal of this study was to determine if there was a desire and/or need for a previvor clinic in Columbia, South Carolina and to determine the utility and efficacy of the existing clinics in Greenville, South Carolina—the Genetics Management Clinic and High-Risk Breast Lifetime Clinic. In addition, researchers sought to determine what services ought to be incorporated into such a clinic and how often patients would require these services. Lastly, by comparing experiences of individuals at both hospitals, researchers hoped to assess whether patients who had access to a previvor clinic were better able to adhere to management recommendations compared to those who did not have access. It was hypothesized that, without clear direction and guidance from knowledgeable health professionals, many previvors may struggle to adhere to NCCN management guidelines. Without proper care, previvors are at risk for worsened health outcomes, so it is expected that this study may improve overall patient satisfaction and care. Furthermore, it was anticipated that patients within the Columbia, South Carolina area would have strong interest for the establishment of a local previvor clinic and that patients who have attended the specialty clinic in Greenville experienced high patient satisfaction and will attest to the value and benefit of such a clinic.

2.3 Methods

2.3.1 IRB Approval

This study was approved by the Institutional Review Board, Office of Research Compliance, of the University of South Carolina, Columbia, SC in July 2020.

2.3.2 Participants

Participants included in this study were patients who had a genetic counseling appointment at Prisma Health-Midlands in Columbia, South Carolina or an appointment at the Genetics Management Clinic or High-Risk Breast Lifetime Clinic at Prisma Health Upstate in Greenville, South Carolina. To be included in the study, patients were required to meet the following inclusion criteria: Mutation-positive in a hereditary cancer gene that predisposes to breast cancer and has NCCN management recommendations, including: *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN*, *TP53*, or have a Tyrer-Cuzick or other risk model score of 20% or greater, requiring additional management as dictated by the NCCN Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic guidelines; and never had a diagnosis of cancer (NCCN, 2020).

Prisma Health-Midlands patients who are mutation-positive were recruited retroactively through the clinic's database and prospectively as encountered by the Prisma Health-Midlands genetic counselors. Prisma Health-Midlands patients who are considered high-risk given their Tyrer-Cuzick score were informed of the study prospectively by their genetic counselor. Prisma Health-Upstate patients who have visited the Genetics Management Clinic or High-Risk Breast Lifetime Clinic and met the inclusion criteria were identified through the clinic's records and sent a recruitment letter inviting them to participate in the study.

2.3.3 Online Survey

Participants were mailed a letter inviting them to complete an online survey through Qualtrics^{XM} software. Invitation letters were mailed out to patients of the Genetics Management Clinic and High-Risk Breast Lifetime Clinic in August 2020. Additionally, Prisma Health-Midlands patients who received genetic counseling in the past and were mutation-positive also were sent an invitation letter at this time. Beginning in August 2020, Prisma Health-Midlands patients who received negative genetic testing but were identified to be high risk for breast cancer were given a letter by their genetic counselor, inviting them to participate in the study. Included in the recruitment letter was a link to the participant's clinic-specific survey (Appendix A-C). The survey remained open until November 15, 2020. At the beginning of the survey, participants were asked to provide informed consent by selecting the "I consent" option. Data collection was kept anonymous to protect the privacy of the participants.

Surveys were unique to the clinic the participants were involved with and included a mixture of questions addressing patient demographic information, genetic status, clinic satisfaction, and further suggestions for clinic development and improvement (Appendix D-F). The final question of all three surveys invited participants to list their phone number if interested in participating in a semi-structured phone interview. By providing their phone number, participants consented to being contacted by the principal investigator for this purpose.

2.3.4 Semi-Structured Interviews

Participants who indicated on the survey their willingness to partake in an additional interview were called to complete a semi-structured phone interview regarding

their experiences at their respective clinic (i.e., Prisma Health-Midlands Genetic Counseling, Genetics Management Clinic, or High-Risk Breast Lifetime Clinic). Prior to beginning the interview, participants were asked to give verbal consent to participate in the interview and have their responses recorded. Surveys were transcribed by hand, labeled numerically by the order in which they were completed, and secured on a password-protected computer.

2.3.5 Data Analysis

All data remained deidentified to protect the privacy of the participants. Survey responses to multiple choice questions and ranking activities were analyzed and frequencies were recorded for analysis. Free response questions and qualitative interviews were analyzed for themes using a grounded theory approach, and shared themes were drawn from both the surveys and interviews. This analysis was completed by two independent researchers (M.T. and W.D.), and categories were discussed until three common themes were agreed upon.

2.4 Results

2.4.1 Participation and Demographics

A total of 28 surveys were started, and 26 were completed across the three clinics. Seven surveys were completed by Prisma Health-Midlands patients, ten by Genetics Management Clinic patients, and nine by High-Risk Breast Lifetime Clinic patients. Six semi-structured phone interviews were completed—four by Prisma Health-Midlands patients and two by Genetics Management Clinic patients. An additional eight participants indicated on their survey that they would be willing to complete a semi-structured phone interview but were unable to be contacted to arrange the interviews.

Table 2.1 Demographic Characteristics of Participants

Characteristic	*N* (Percentage)
Clinic	
Prisma Health-Midlands	7 (26.9%)
High-Risk Breast Lifetime Clinic	10 (38.5%)
Genetics Management Clinic	9 (34.6%)
Age (Obtained for Prisma Health-Midlands Only)	
18-25	1 (14.3%)
26-30	1 (14.3%)
31-40	1 (14.3%)
41-50	0 (0%)
51-60	2 (28.6%)
61-70	1 (14.3%)
Over 70	1 (14.3%)
Genetic Status	
Positive	16 (61.5%)
Negative/High-Risk	10 (38.5 %)
Gender*	
Female	26 (100%)
Male	0 (0%)

*Both men and women were eligible for the study, however, all participants reported female gender identity.

2.4.2 Prisma Health-Midlands

A total of nine surveys were started, and seven surveys were completed. Of the seven completed surveys, six participants indicated they had a mutation in a hereditary cancer gene, while the remaining participant had a high lifetime risk for breast cancer based on her family history. Figure 2.1 shows the risk status of Prisma Health-Midlands participants, including genes in which participants had a pathogenic variant and those who tested negative and are high risk based on other factors.

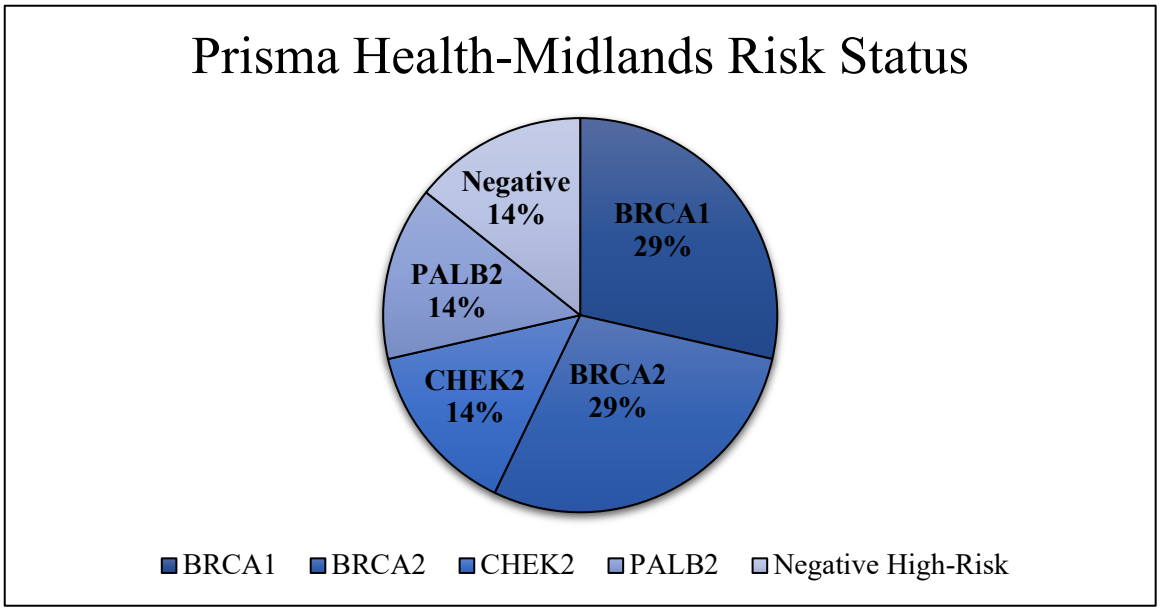

Figure 2.1 Prisma Health-Midlands Risk Status

One participant had a history of cancer, and as a result, the survey was ended after answering that she had a cancer diagnosis. Therefore, the total number of complete, eligible responses for the Prisma Health-Midlands clinic was six surveys.

Participants were asked to rate their attitudes regarding their risk perception and care they currently receive related to their cancer predisposition. These attitudes are summarized in Table 2.2.

Table 2.2 Patient Attitudes Regarding Cancer Risk and Care

Patient Attitudes	
	***N* (Frequency)**
I feel I have adequate knowledge about my cancer risk and am able to manage my personal healthcare to meet the recommendations.	
Strongly agree	1 (16.7%)
Somewhat agree	3 (50%)
Neither agree nor disagree	2 (33.3%)
Somewhat disagree	0 (0%)
Strongly disagree	0 (0%)
I feel that my healthcare providers (primary care physician, obstetrician, gynecologist, etc.) have expert knowledge about the cancer screenings recommended for me.	
Strongly agree	0 (0%)
Somewhat agree	2 (33.3%)
Neither agree nor disagree	2 (33.3%)

Somewhat disagree	1 (16.7%)
Strongly disagree	1 (16.7%)
I have difficulty getting insurance coverage for the recommended cancer screenings.	
Strongly agree	1 (16.7%)
Somewhat agree	0 (0%)
Neither agree nor disagree	3 (50%)
Somewhat disagree	1 (16.7%)
Strongly disagree	1 (16.7%)
Some institutions have set up clinics designed to assist individuals who carry a positive mutation in a hereditary cancer gene or who are at a higher risk for developing breast cancer get the recommended screenings and management. How interested would you be in attending this clinic if it were created at Prisma Health-Midlands?	
Very interested	6 (100%)
Somewhat interested	0 (0%)
Neutral	0 (0%)
Likely not interested	0 (0%)
Not at all interested	0 (0%)
How often would you want to attend this type of clinic? (If other, please specify in the provided blank.)	
Only once	1 (16.7%)
Every 5 years	0 (0%)
Every 2 years	0 (0%)
Every year	1 (16.7%)
Every 6 months	2 (33.3%)
Other— "As often as recommended"	1 (16.7%)
Other— "Every 3 months"	1 (16.7%)

The majority of participants reported feeling very confident or somewhat confident that they had adequate knowledge of their cancer risks and had the ability to manage their healthcare accordingly (*N*=4, 66.7%). However, when asked the degree to which they agree that their healthcare providers had expert knowledge of their cancer risk and the recommended screenings, no participants strongly agreed. One-third (33.3%, *N=2)* somewhat felt that their providers had expert knowledge regarding their risks and management, and one-third (33.3%, *N*=2) of participants did not feel they had this knowledge.

All six participants (100%) indicated that they were very interested in attending a specialized management clinic if one were developed at Prisma Health-Midlands. Participants were then given a list of possible services and asked to rank these services in order of most important to least important for incorporation into a specialized management clinic. Services were ranked by each participant and weighted frequencies were calculated for each category based on participant responses (Appendix G). The results of this ranking question are summarized in Figure 2.2.

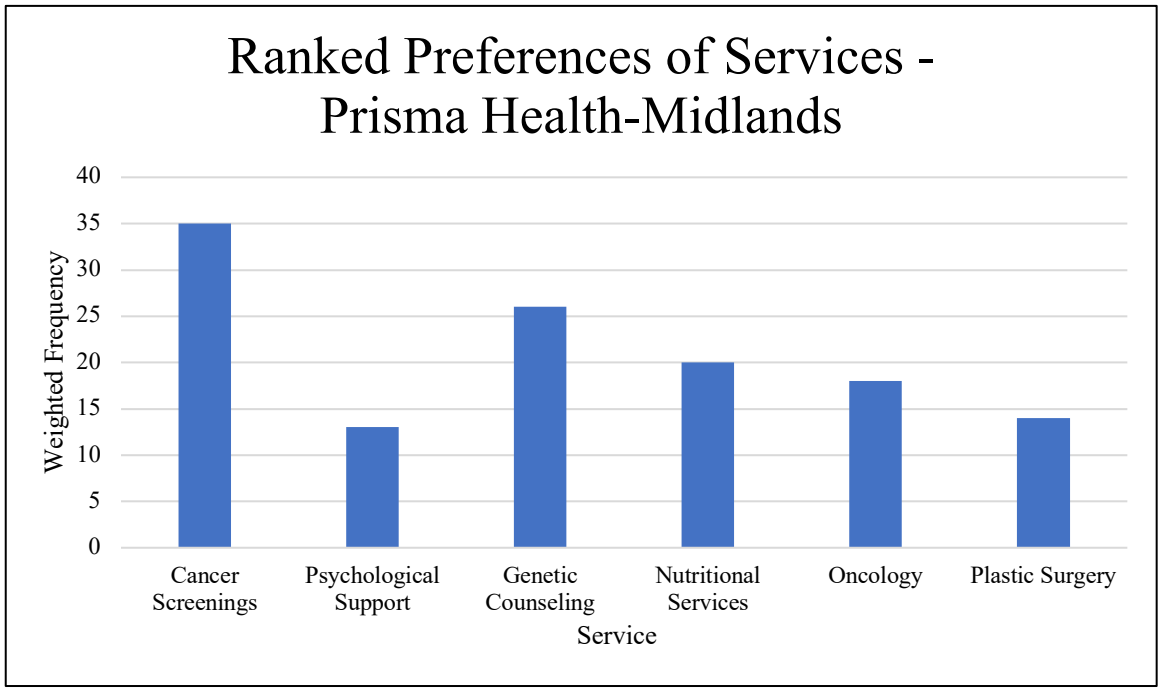

Figure 2.2 Ranked Preferences of Services—Prisma Health-Midlands

The majority of participants ranked access to cancer screenings (i.e., mammogram, breast MRI, etc.) as most important, while psychological support was ranked overall as least important.

2.4.3 Genetics Management Clinic

A total of ten surveys were completed by patients of the Genetics Management Clinic; however, only four of the participants met all of the inclusion criteria for the study. Due to the limited number of responses, answers from participants who completed the survey but had a personal history of cancer were still included.

Participants identified as having pathogenic variants in three hereditary cancer genes: *BRCA1, BRCA2,* and *PALB2,* with the majority of participants (*N*=5, 50%) carrying a *BRCA2* variant. Figure 2.3 shows the breakdown of genetic mutation status by gene.

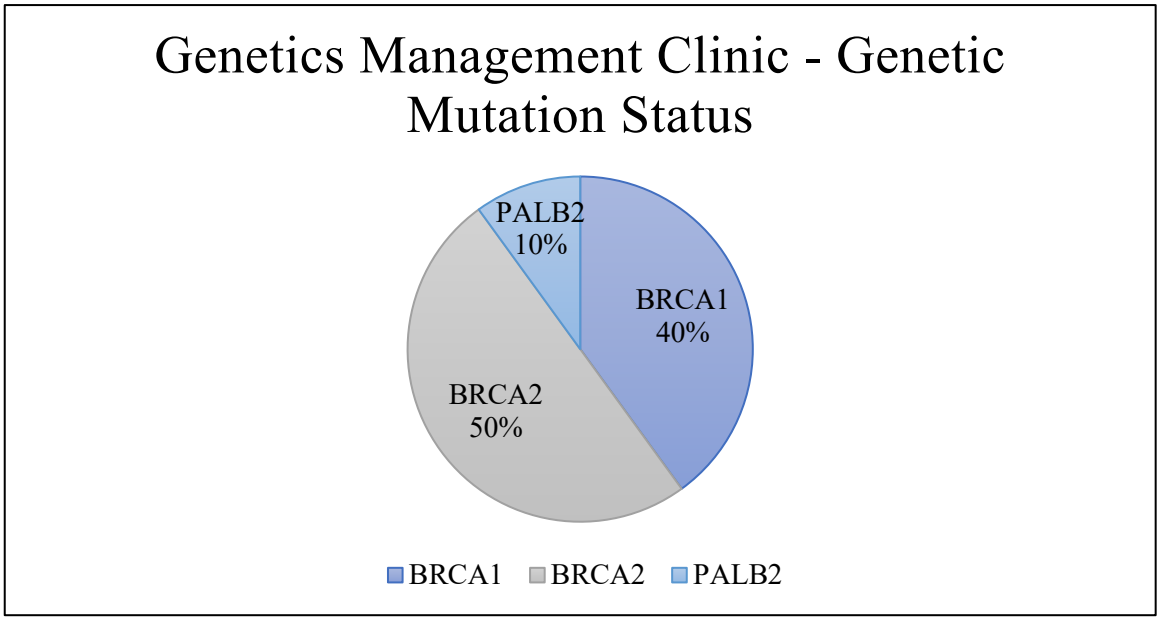

Figure 2.3 Genetics Management Clinic Genetic Mutation Status

After learning of their genetic status, the majority of participants (*N=8,* 80%) have attended the Genetics Management Clinic for longer than 18 months. On average, most participants attended the clinic once per year and felt that this interval between visits was appropriate (*N*=9, 90%). When asked to rate their satisfaction with the clinic on a scale from one to ten (1 being not at all satisfied and 10 being very satisfied), 90%

(*N*=9) rated their satisfaction with the clinic a 10, very satisfied. Additionally, all ten participants indicated that they would recommend the GMC to others (100%).

2.4.4 High-Risk Breast Lifetime Clinic

A total of nine surveys were completed by participants of the High-Risk Breast Lifetime Clinic. 88.9% of participants (*N*=8) have attended the High-Risk Breast Lifetime Clinic for at least 12 months. Four participants indicated that they visit the High-Risk Breast Lifetime Clinic semi-annually (50%), three participants attend annually (37.5%), and one participant no longer attends the clinic (12.5%). Overall, participants expressed high satisfaction with the care they received at the High-Risk Breast Lifetime Clinic as seen in Table 2.3, with all participants ranking their satisfaction greater than or equal to seven on a number scale identifying satisfaction, and the majority (55.5%, *N*=5) ranking satisfaction greater than or equal to nine. Two-thirds (66.7%, *N*=6) expressed that breast cancer screenings had gotten easier since attending the High-Risk Breast Lifetime Clinic.

Table 2.3 High-Risk Breast Lifetime Clinic Patient Satisfaction and Clinic Utility

High-Risk Breast Lifetime Clinic Patient Satisfaction and Clinic Utility	
	***N* (Frequency)**
On a scale from 0-10, with 0 being not at all satisfied and 10 being very satisfied, how satisfied are you with the services provided by the High-Risk Breast Lifetime Clinic?	
0- not at all satisfied	0 (0%)
1	0 (0%)
2	0 (0%)
3	0 (0%)
4	0 (0%)
5	0 (0%)
6	0 (0%)
7	2 (22.2%)
8	2 (22.2%)
9	2 (22.2%)
10- very satisfied	3 (33.3%)

Has getting breast cancer screening (i.e., mammograms and breast MRIs) gotten easier since attending this clinic?	
Yes	6 (66.7%)
No	2 (22.2%
Not sure	1 (11.1%)

2.4.5 Experiences with Genetic Counseling

All participants were able to meet with a genetic counselor either prior to or following genetic testing. Participants expressed high levels of satisfaction with their genetic counseling experience, especially in terms of level of thoroughness and education they received regarding their cancer risk. Participants appreciated the expert genetic knowledge of genetic counselors coupled with their attention to psychosocial concerns of the patient and their relatives. Participants described the process of meeting with a genetic counselor to discuss genetic test results, associated risks, and options for future care. One participant compared her experience seeing a genetic counselor at Prisma Health-Midlands to her sister's experience at her doctor's office:

> Well, I was so pleased with the whole experience with the genetic counseling… And when I compare my experience to my sister who just had a blood test at her OB/GYN and just sent a letter in the mail, she really had no counseling. She filled out a family history, but nobody questioned or interviewed her or anything. Totally different experience. And she was so jealous when I told her my experience and what I went through and how supportive everybody was. (Prisma Health-Midlands Participant 7)

As described by Prisma Health-Midlands Participant 7 and several others, benefits of genetic counseling included psychosocial support, thorough education regarding cancer

risks, and discussion of options to reduce cancer risk (Prisma Health-Midlands Participant 7, Genetics Management Clinic Participant 4 & 5).

2.4.6 Experiences of Participants Without a Specialized Clinic

Without access to a specialized management clinic, participants rely on online support groups, current medical providers, and self-advocacy to get care related to their cancer predisposition. After learning of her *CHEK2* mutation, Prisma Health-Midlands Participant 1 relied on a Facebook group for *CHEK2* mutation carriers to keep her up to date. Members of the Facebook group willingly shared their experiences and research on *CHEK2* variants, but this led to additional questions about associated cancer risks and management options. Prisma Health-Midlands Participant 1 described the dynamic of this group as a "big question and a big waiting game," when expressing the confusion that can surround online support groups. Despite some confusion regarding cancer risks, Prisma Health-Midlands Participant 1 pointed out that a major benefit of the online support group was the allied search for information. She described that the members of the group actively search out research regarding *CHEK2* and willingly share it with the group, although she recognized that information is somewhat limited.

Other participants relied on support from their family members who also carry the same variant. Prisma Health-Midlands Participant 4 described talking with her mother about their risks for breast cancer: She recalled talking with her mother about the different options for risk-reduction but concluded that they would address cancer or management if it came to fruition.

Previvors without a specialized management clinic relied on the experience of their current medical provider (primary care physician, OB/GYN, etc.) for care related to

their increased risk for breast and other cancers. Some participants expressed high levels of satisfaction with their current provider's knowledge regarding their cancer predisposition, while others felt that they had to educate their providers about their increased risk and recommended risk-reduction options. Prisma Health-Midlands Participant 1 described her visits with her nurse practitioner after learning of her positive *CHEK2* variant and explained that her nurse practitioner had been helpful in arranging the necessary screenings. Together, they looked at her genetic testing results and recommendations for management and came up with a plan of action, including scheduling a mammogram and breast MRI. Prisma Health-Midlands Participant 1 also planned to discuss scheduling a colonoscopy in the near future and believed that her nurse practitioner would be able to help facilitate that referral.

Although Prisma Health-Midlands Participant 1 felt that her provider was well-equipped to care for her, not all participants felt the same regarding their providers. Prisma Health-Midlands Participant 3 found that some of her providers did not fully understand her cancer predisposition or the recommendations for screenings or risk-reducing care:

> It's hard to like to tell a doctor like, "hey, I need this done or this done," you know, like when… it's hard to speak up and say that when they know so much more about medical things than you do. (Prisma Health-Midlands Participant 3)

Prisma Health-Midlands Participant 7 described a similar situation in which a provider could not comprehend her desire to reduce her breast cancer risk in any way possible:

> Because after watching family members go through this, I have even asked my doctor—before we even talked about the genetic counseling—I asked her about

> the possibility of having my breasts removed. And she looked at me like, "What?" And I'm like, "Well, you know, that would just kind of eliminate that…" I'm open to anything. Whatever I can do to not have to go through what I've watched other family members go through. (Prisma Health-Midlands Participant 7)

Like Prisma Health-Midlands Participants 3 and 7 described, previvors without a specialized clinic felt they needed to self-advocate for their desire to be proactive to healthcare providers who did not understand their risk, but previvors also found they had to do so with family members who did not understand their desire to reduce cancer risk in any way possible. Prisma Health-Midlands Participant 1 described a conversation with her husband regarding genetic testing and risk reduction in which he described genetic testing as "fearmongering," but she defended her position on testing and risk-reduction because of experiencing the loss of a close friend at age 29 due to breast cancer, claiming that she would do anything she could to be prepared. These encounters with people who do not understand their predisposition, have sparked participants to do everything in their own power to prevent cancer.

When asked what features participants would include in an ideal management clinic, participants highlighted the need for access to specialists familiar with genetics, a provider who will notify of changes to risk or management recommendations, and a place to ask questions. Participants desired a specialist familiar with genetics and able to provide expert care specific to their risks, whether due to a genetic mutation or family history. Prisma Health-Midlands Participant 4 described how the providers at a local women's center performed her breast ultrasound and were familiar with breast health but did not have expert knowledge on the genetic aspects of her care. After leaving her visit

at the women's center, she felt that there were still probably things related to her risk that she didn't know about. She emphasized the difference between a women's center who specializes in women's care and breast cancer screening versus a center that focuses on this care and screening within the context of previvorship. Prisma Health-Midlands Participant 3 echoed this sentiment, saying that she simply wanted a place where the doctors were familiar with the higher risks of previvors.

A second priority for a specialized management clinic was the ability to be notified of changes to cancer risks or management recommendations. Prisma Health-Midlands Participant 4 indicated that this would be a priority for her when thinking about a specialized clinic and emphasized her desire for a healthcare provider that could follow her along and provide updates on changes to care. Similarly, Prisma Health-Midlands Participant 1 valued the idea of having a specific provider to keep patients informed, to help patients stay informed and on top of their care in addition to providing resources. She also emphasized the importance of having new information explained by a medical professional in layman's terms, ensuring their ability to utilize that information.

In addition to using the clinic as a way to stay informed, participants also wanted the clinic to serve as the place to ask questions and get specific recommendations based on their genetic mutation or family history. Prisma Health-Midlands Participant 1 described the confusion that has come up in her *CHEK2* Facebook group regarding associated cancers and risks and described that a specialized management clinic with specific knowledge regarding care for individuals with a *CHEK2* mutation would be extremely beneficial in order to get her questions answered. Prisma Health-Midlands Participant 4 expressed interest in asking questions about risk-reducing surgeries and

other screening options and felt that there was probably information about these options that she was unfamiliar with. Similarly, Prisma Health-Midlands Participant 7 discussed a conversation with her sister about taking tamoxifen and how she was not certain that taking it was still recommended. She also had questions regarding the benefits of a breast MRI and the logistics of getting one.

2.4.7 Experiences of Participants with a Specialized Clinic

Participants from the Genetics Management Clinic and High-Risk Breast Lifetime Clinic expressed that access to a specialized management clinic provided a simplified and efficient management process, streamlining their care. Genetics Management Clinic Participant 4 described the role of the nurse practitioner who heads the clinic as a navigator and facilitator. The nurse practitioner would provide referrals to the necessary specialists, assist in scheduling appointments for consults and imaging, and provided insight on proper care and management. By navigating the process alongside the patient, the nurse practitioner was able to expedite care and ensure the patient received the appropriate services, removing the burden from the patient. High-Risk Breast Lifetime Clinic Participant 7 echoed this feeling, stating that the High-Risk Breast Lifetime Clinic providers had an expert focus on identifying, treating, and curing breast disease, which she found particularly helpful. High-Risk Breast Lifetime Clinic Participant 8 expressed that the ease of visits and concentration on breast cancer risk-reduction and care were the most valuable aspects of the clinic.

In addition to providing care in a simplified manner, participants expressed that this system also prevented delays to care. Genetics Management Clinic Participant 7 mentioned that she was able to get a second surgery quicker because of having access to

the physicians at the Genetics Management Clinic. Genetics Management Clinic Participant 4 discussed how she might have approached clinical care after learning of her *BRCA1* mutation, and how the clinic's approach ensured she saw the proper physicians quickly. Instead of seeing one doctor at a time and approaching care in a step-wise fashion, the clinic made the referrals simultaneously. Having access to a specialized management clinic allowed her care to be expedited compared to if she had coordinated her care plan own her own. She believed that she would've seen all of the recommended physicians eventually, but never would have done so at such a high speed if it had not been for the Genetics Management Clinic.

Study participants emphasized that a major benefit of the Genetics Management Clinic and High-Risk Breast Lifetime Clinic is access to healthcare providers who give detailed education regarding risk and the necessary information to come to an informed decision regarding care and risk-reduction. Genetics Management Clinic Participant 8 described genetic counseling as the most valuable service of the Genetics Management Clinic and how after discussing her mutation in-depth, she felt much better about her plan for care. Genetics Management Clinic Participant 5 expressed similar feelings and explained how receiving thorough education about her genetic mutation and what it meant allowed her to not only be proactive but gave her a sense of control. By receiving proper education about their elevated risk for developing cancer, participants were able to make educated, well-informed decisions relating to their care plan, whether electing to proceed with screening or a risk-reducing surgery.

Furthermore, access to the Genetics Management Clinic or High-Risk Breast Lifetime Clinic reduced participant stress related to receiving care. Several participants

described the sense of relief they experienced after connecting with the clinic. Genetics Management Clinic Participant 5 highlighted this feeling when describing how the clinic recommended where to go, which providers to see, and facilitating scheduling. By leaving this in the hands of the Genetics Management Clinic, she was able to feel more relaxed and less overwhelmed by the process. High-Risk Breast Lifetime Clinic Participant 5 shared that she found "peace of mind knowing the High-Risk Breast Lifetime Clinic is closely monitoring everything." Numerous participants expressed that the process of learning of their increased risk and determining a management plan could be overwhelming, but the clinics help make it manageable (Genetics Management Clinic Participant 5). Genetics Management Clinic Participant 4 describes the overall impact the Genetics Management Clinic had on her, saying:

> I would just say that they're useful. I mean, if it wasn't for the clinic, could I have done all this? Yes, but with them doing it, it was more efficient, it was less stressful for me, it was one less thing I had to research. It just made the whole process just a little bit easier—a lot easier, not a little, a lot. I only have positive things to say. (Genetics Management Clinic Participant 4)

2.5 Discussion

2.5.1 Specialized Management Clinic for Psychosocial Support

Participants who did not have access to a specialized management clinic all indicated that they would be interested in attending such a clinic if one were to be established in the Midlands region. The study sought to identify current practices and resources that these participants utilize, as well as their preferences for the setup of a specialized management clinic in the future. Participants currently utilize online support

groups as well as family members who also are at increased risk for developing cancer as a support resource, relying on these individuals not only for psychological support, but also as a source of information regarding risk and risk-reduction.

Mahon (2014) found that previvors often feel isolated, as if no one else understands what they are going through and "different from those who do not carry a mutation" (Mahon, 2014, p. 22). Study participants, like the participant that joined a Facebook group, expressed similar feelings to that found by Mahon. These findings indicate that a psychosocial support aspect to a specialized clinic may also be beneficial.

Leaning into a support community allowed for not only psychological support, but also further information and education regarding cancer risk and current research. However, despite the benefits gained from the previvor community, Prisma Health-Midlands Participant 1 also expressed concerns with the contrasting information shared within the group, sharing that that some Facebook users had certain perceptions regarding associated cancer risks while others held different perceptions. This experience suggests that support groups may be beneficial for encouragement and learning from other previvors but may not be the most appropriate place for effective education.

In terms of psychological health, numerous participants with access to the Genetics Management Clinic described how the management clinic helped reduce their levels of stress (Genetics Management Clinic Participants 1, 4, & 5, High-Risk Breast Lifetime Clinic Participant 5). Genetics Management Clinic Participant 5 described how the process "can be overwhelming, and they make it manageable." High-Risk Breast Lifetime Clinic Participant 5 shared how the clinic helped provide peace of mind and reduction of anxiety. Genetics Management Clinic Participant 1 felt that she was

confident that she was doing everything to prevent the return of cancer, and Genetics Management Clinic Participant 4 echoed this sentiment, describing how the clinic helped make the process a lot easier and less stressful.

2.5.2 Specialized Management Clinic for Healthcare Needs

Previvors without access to a specialized management clinic often rely on their current healthcare providers (primary care physicians, OB/GYNs, etc.) for care related to their increased cancer risk. Review of the literature suggests that these healthcare providers often lack the confidence regarding management guidelines to effectively care for these patients (Dekanek et. al., 2019). In fact, Dekanek and colleagues (2019) surveyed 86 OB/GYN and family practice physicians and found that only 44% of providers surveyed felt somewhat confident in discussing *BRCA* management guidelines, and no providers felt completely confident in doing so.

Participants in our study were asked to rank their agreement with the following statement: "I feel that my healthcare providers (primary care physician, obstetrician, gynecologist, etc.) have expert knowledge about the cancer screenings recommended for me" (Prisma Health-Midlands Survey). No participants indicated that they strongly agreed with the statement, further supporting previous literature that suggests that healthcare providers may require additional education about hereditary cancer and familial cancer and how to care for these patients. This finding again points to the need for a specialized management clinic, with providers who have expert knowledge in genetics and previvorship.

A study by Dean and Davidson (2018) discussed how previvors cope with uncertainty related to their increased risk for cancer and how they navigate care. One

common theme they identified among previvors was feeling that their experiences with healthcare providers were overwhelming. This feeling was not simply due to the management recommendations associated with the visit but also connected with the fact that previvors are sometimes burdened with the task of educating the provider about their risk and management (Dean & Davidson, 2018). Prisma Health-Midlands Participant 3 experienced this lack of provider education and described how challenging it was to broach the subject with her physician because she felt it was uncomfortable to question the authority of a physician. In this study, one previvor encountered a healthcare provider who did not understand her perception of her cancer risk which left her feeling invalidated when she asked her physician about the possibility of a risk-reducing mastectomy. While there was one participant that felt comfortable with the care given by their healthcare provider, this was not the majority opinion. Therefore, these experiences highlight previvors' need to advocate for themselves in the healthcare community, standing up for the care that they need.

Participants with access to a specialized management clinic (either the Genetics Management Clinic or High-Risk Breast Lifetime Clinic) expressed high levels of satisfaction with their experience at the clinic, especially in regard to the simplicity of the care they received. Although receiving the initial risk assessment and management plan can be quite overwhelming, participants expressed that once established in the clinic, care became much easier. Goh and Spiegelman (2020) found that scheduling difficulties were a major factor prohibiting previvors' proper adherence to the management guidelines. By establishing a specialized management clinic, this barrier was reduced (if not eliminated) with all coordination of appointments for screening or clinical evaluation being handled

by one clinic. Other studies found that insurance coverage for surveillance was a barrier to adherence; however, our participants did not express concerns with insurance coverage, possibly due to the coordination of the management clinic (Dean et. al., 2017).

A specialized management clinic would include psychosocial support, management information, and updates on genetics. All six participants from the Prisma Health-Midlands clinic indicated they would be interested in attending a specialized management clinic for previvors if one were to be established. Although exact logistical details will depend on clinic staffing and location-dependent requirements, participants indicated several important services to be included in a management clinic. These integral features included access to a specialist with expertise in genetics, a system in place to be notified of changes to risk or management, and a place where previvors can ask questions.

Participants described the need for a specialist in genetics and oncology to be involved in their care (Prisma Health-Midlands Participants 3 & 4), as Prisma Health-Midlands Participant 3 sought access to providers who understood risks specific to her genetic mutation. This desire for a specialist's involvement in a management clinic may again relate back to the fact that general practitioners may not be confident in providing care to previvors and, as a result, previvors' care may not be sufficiently attended to (Dekanek et. al., 2019). This likely is of no fault of general practitioners but may stem from inadequate provider education regarding hereditary cancer and familial risks that reduces the confidence of these healthcare workers. A dedicated clinic for these individuals could provide a solution to a lack of access to educated providers without the need to provide mass education.

This benefit was observed in our study with those who have access to a specialized management clinic. The patients were able to be seen for care quickly by multiple different providers, and any delay in care was prevented as much as possible. Genetics Management Clinic Participant 4 explained that she probably would have gone to different providers (breast, gastroenterology, etc.) in a prioritized manner over a period of time, but that it would not have been expedited like it was in the Genetics Management Clinic. Once theoretical plans have been put into action by the management clinic, previvors begin their journey towards cancer prevention and risk-reduction.

In addition to desiring a thorough education from someone who understands their exact mutation, participants also seek a clinic organized in such a way that they will be notified of any important changes to their risk status. As the genetics behind hereditary cancer continue to be researched, cancer risks and management recommendations may be updated. The NCCN publishes updated guidelines for genetic and familial high-risk care on a regular basis, often updating minor details, but occasionally altering cancer risk estimates or recommended screening strategies or risk-reduction methodology (NCCN, 2020). Other research studies or clinical trials may offer specific information for previvors that individuals may not have access to on their own but would value learning about from a trusted healthcare provider. Prisma Health-Midlands Participant 1 described access to research as a major benefit of the *CHEK2* Facebook group. It is possible that access to such research shared by a healthcare provider may be of even greater benefit if coupled with a simplified explanation.

Lastly, participants desired a clinic that gave them the opportunity to ask questions about their cancer risks and recommended management. Logistical questions

about screening, risk-reducing surgeries, and providers familiar with genetics were common concerns of participants. For example, one participant was interested in more information about recommended providers or clinics and where to go for care (Prisma Health-Midlands Participant 1). Although there are many factors that influence a patient's care choices (for example, age, parental status, level of psychological distress, perceived family support, experienced medical uncertainty), patients require a clinic staffed with a provider able to answer their questions and assist in facilitating decision-making with these factors taken into account (Hesse-Biber & An, 2016).

Participants of the Genetics Management Clinic and High-Risk Breast Lifetime Clinic felt that they were provided all information needed to make informed decisions regarding their care. Cancer prevention and risk-reduction comes with many nuances and options, which participants appreciated the opportunity to discuss with a specialized provider. When asked about the most valuable service provided by the clinic, Genetics Management Clinic Participant 5 stated:

> Education about the gene mutations as related to what it means and actions to take that can reduce your risk. I felt I could be proactive in reducing my cancer risks and that gave me some sense of control and a plan. After having my world turned upside down with a cancer diagnosis that was very helpful to me. (Genetics Management Clinic Participant 5)

Providers in the clinic could help patients balance logical and emotional decision-making styles and assist the patient in coming to a decision they are satisfied with (Dean & Rauscher, 2017). For example, the provider could discuss the options of risk-reducing mastectomy or oophorectomy, and the potential effects it could have on patient quality of

life and family dynamics in addition to lowering cancer risks (Alexandre et. al., 2017; Mahon, 2014). By having expert providers provide these services in place of the patient's primary care physician or OB/GYN, general practitioners have reduced burden for counseling patients on risks and management they are not confident of, and patients receive care catered specifically to their needs (Dekanek et. al., 2019).

2.5.3 Role of a Genetic Counselor

Although not specifically seeking to look at patient satisfaction with genetic counseling, this study found that participants were highly satisfied with the level of information provided to them by genetics professional. Many participants first learned of their previvor status from a genetic counselor, and participants expressed the benefits of meeting with a genetic counselor. Participants indicated that they felt supported throughout the testing process and were pleased with the information provided to them by their genetic counselor, stating that it helped them feel more prepared and increased their ability to be proactive regarding their risk.

This high satisfaction with genetic counselors echoes previous research on patient satisfaction with genetic counseling (Sagi et. al., 1998). After genetic counseling, patients better understood the link between genetics and cancer, received answers to their questions, and felt confident in their plan for follow-up (Sagi et. al., 1998). Genetic counselors' ability to break down complex scientific material in layman's terms, coupled with a focus on psychosocial considerations associated with hereditary cancer testing, allows for increased patient satisfaction. Although participants were highly satisfied with their initial genetic counseling visit, it is evident from the survey responses that patients would like additional follow-up regarding their high-risk status, whether due to family

history or a genetic mutation. This supports the role of a genetic counselor in a specialized management clinic.

2.6 Study Limitations

A major limitation of this study is the lack of participants who responded to the survey invitations and semi-structured phone interviews. Possible explanations for this may be due to the nature of sending research study invitations by mail, especially considering participants had no notification prior to receiving a letter that they would be contacted. Furthermore, it is possible that patients who are several years out from their initial appointment with the genetic counselor would be less interested in participating in the study if they already have a management plan in place. Additionally, the majority of participants with hereditary cancer mutations carried a variant in either *BRCA1* or *BRCA2*, the two genes in the study with the highest lifetime risk for breast cancer. It is possible that this sample is biased towards desiring a clinic due to their substantially increased risk, as individuals of the clinics with variants in genes with lower risk chose not to participate. It is however possible, that this finding could be due to mutations in these genes being the most common finding on hereditary breast cancer panels and clinical testing for these genes having been available for longer, implying more patients may have been identified with *BRCA1* or *BRCA2* variants.

2.7 Future Research

Further research is needed in the area of previvorship and the establishment and utility of specialized management clinics, particularly studying patient adherence to clinical guidelines once a patient is seen in a management clinic, exploring patient health outcomes of previvors within these clinics, and further exploration of the clinic model for

previvors with hereditary cancer syndromes associated with cancers other than breast cancer.

CHAPTER 3: CONCLUSIONS

This study explored patient satisfaction and approach to care for previvors with access to a specialized management clinic and for those without access. Overall, all participants without access to a management clinic desired the establishment of one in the Midlands region, particularly with the hope of connecting with a specialist in genetics, being notified of changes to cancer risks or management recommendations, and having an opportunity to get questions answered. Participants described their current framework of care which largely included online support groups, guidance from their general practitioner, and the need for self-advocacy within both the healthcare realm and with their family, friends, and society. In contrast, participants who had access to a management clinic, such as the Genetics Management Clinic or High-Risk Breast Lifetime Clinic expressed high levels of satisfaction with their care. These participants cited a simplified and efficient process, prevention of delay to care, access to all information needed to make informed decisions, and reduced stress as major benefits of the management clinic. Specialized management clinics are not only helpful in guiding patients and allowing unhindered access to resources and support, but it improves the overall psychological health of the patient, which may lead to better health outcomes. These findings further point to the necessity of specialized management clinics, specific to the needs of previvors in order to improve patient care and overall wellbeing.

REFERENCES

Alexandre, M., Black, J., Whicker, M., Minkin, M. J., & Ratner, E. (2017). The management of sexuality, intimacy, and menopause symptoms (SIMS) after prophylactic bilateral salpingo-oophorectomy: How to maintain sexual health in "previvors." *Maturitas*, *105*, 46–51. doi: 10.1016/j.maturitas.2017.06.003

Carvalho, J. P., Baracat, E. C., & Carvalho, F. M. (2019). Ovarian cancer previvors: How to manage these patients? *Clinics*, *74*. doi: 10.6061/clinics/2019/e1343

Dean, M., & Davidson, L. G. (2018). Previvors' uncertainty management strategies for hereditary breast and ovarian cancer. *Health Communication*, *33*(2), 122–130. doi: 10.1080/10410236.2016.1250187

Dean, M., & Rauscher, E. A. (2017). "It was an emotional Baby": Previvors' family planning decision-making styles about hereditary breast and ovarian cancer risk. *Journal of Genetic Counseling, 26*(6), 1301-1313. doi:10.1007/s10897-017-0069-8

Dean, M., Scherr, C. L., Clements, M., Koruo, R., Martinez, J., & Ross, A. (2017). "When information is not enough": A model for understanding BRCA-positive previvors' information needs regarding hereditary breast and ovarian cancer risk. *Patient Education and Counseling*, *100*(9), 1738–1743. doi: 10.1016/j.pec.2017.03.013

Dekanek, E. W., Thull, D. L., Massart, M., Grubs, R. E., Rajkovic, A., & Mai, P. L. (2019). Knowledge and opinions regarding BRCA1 and BRCA2 genetic testing among primary care physicians. *Journal of Genetic Counseling*, *29*(1), 122–130. doi: 10.1002/jgc4.1189

Getachew-Smith, H., Ross, A. A., Scherr, C. L., Dean, M., & Clements, M. L. (2019). Previving: How unaffected women with a BRCA1/2 mutation navigate previvor identity. *Health Communication*, 1–10. doi: 10.1080/10410236.2019.1625002

Goh, L. H., & Spigelman, A. D. (2020). Assessing the adherence to guidelines in Lynch syndrome patients: A pilot study. *ANZ Journal of Surgery*. doi: 10.1111/ans.15690

Hesse-Biber, S., & An, C. (2016). Genetic testing and post-testing decision making among BRCA-positive mutation women: A psychosocial approach. *Journal of Genetic Counseling*, *25*(5), 978–992. doi: 10.1007/s10897-015-9929-2

Hoskins, L. M., Roy, K. M., & Greene, M. H. (2012). Toward a new understanding of risk perception among young female BRCA1/2 "previvors". *Families, Systems, & Health*, *30*(1), 32–46. doi: 10.1037/a0027276

Mahon, S. M. (2014). Providing care for previvors: Implications for oncology nurses. *Clinical Journal of Oncology Nursing*, *18*(1), 21–24. doi: 10.1188/14.cjon.21-24

Monticciolo, D. L., Newell, M. S., Moy, L., Niell, B., Monsees, B., & Sickles, E. A. (2018). Breast cancer screening in women at higher-than-average risk: Recommendations from the ACR. *Journal of the American College of Radiology*, *15*(3), 408–414. doi: 10.1016/j.jacr.2017.11.034

National Comprehensive Cancer Network (2020, May 29). Breast cancer risk reduction. Version 1.2021. Retrieved March 30, 2021, from https://www.nccn.org/professionals/physician_gls/pdf/breast_risk.pdf

National Comprehensive Cancer Network (2020, November 20). Genetic/familial high-risk assessment: Breast, ovarian, and pancreatic. Version 1.2020. Retrieved May 13, 2020, from https://www.nccn.org/professionals/physician_gls/pdf/genetics_bop.pdf

National Comprehensive Cancer Network (2019, December 13). Genetic/familial high-risk assessment: Colorectal. Version 3.2019. Retrieved May 14, 2020, from https://www.nccn.org/professionals/physician_gls/pdf/genetics_colon.pdf

National Comprehensive Cancer Network (2020, May 13). NCCN clinical practice guidelines in oncology: Gastric cancer. Version 2.2020. Retrieved May 14, 2020, from https://www.nccn.org/professionals/physician_gls/pdf/gastric.pdf

National Society of Genetic Counselors. *Model Legislative Provisions*. https://www.nsgc.org/p/cm/ld/fid=18#scope.

Petrucelli, N., Daly, M. B., & Pal, T. (2016, December 15). BRCA1- and BRCA2-associated hereditary breast and ovarian cancer. Retrieved May 14, 2020, from https://www.ncbi.nlm.nih.gov/books/NBK1247/

Sagi, M., Kaduri, L., Zlotogora, J., & Peretz, T. (1998). The effect of Genetic counseling on knowledge and perceptions regarding risks for breast cancer. *Journal of Genetic Counseling*, *7*(5), 417–434. https://doi.org/10.1023/a:1022880831996

Senter, L., & Hatfield, R. (2016). Nurse practitioners & genetic counselors: Collaborative roles in a complex system. *The Nurse Practitioner*, *41*(7), 43–49. doi: 10.1097/01.npr.0000470355.00838.a2

Young, A. L., Butow, P. N., Tucker, K. M., Williams, R., Healey, E., & Wakefield, C. E. (2019). Health professional and at-risk BRCA young adult perspectives about information needs: What does Gen Y need to know? *Journal of Genetic Counseling*, *28*(6), 1154–1165. doi: 10.1002/jgc4.1

APPENDIX A: RECRUITMENT LETTER FOR

PRISMA HEALTH-MIDLANDS PARTICIPANTS

Dear Patient,

You are receiving this letter because you underwent cancer genetic counseling with Prisma Health-USC Genetic Counseling, and we are interested in creating a clinic that could assist in your healthcare. We are looking into the development of a clinic that would allow individuals with an increased risk of cancer to be seen by one healthcare practitioner who would oversee all the screening required because of that risk. To assess patient interest in this service, we are asking former patients to complete a survey. Through the information collected, we hope to provide better care for our patients.

The study involves completing an online survey. At the completion of the survey, you will be given the option for a follow-up telephone interview. The survey is available online through Qualtrics and should take no more than 15 minutes to complete. The survey involves questions about your understanding of your predisposition for cancer, how you are currently receiving care, and your level of interest in the establishment of a new clinic for your cancer management. Participation is completely confidential. Data from the study will be stored in a secure location, will not be placed in your health record nor linked to your name, and will be destroyed at the completion of the study. The phone interviews will be recorded for analysis by the research team, but all information that you provide will be kept confidential. The results of the study may be published or presented; however, your identity will remain anonymous. This study is completely voluntary, and there are no negative consequences should you withdraw from the study.

This study is a University of South Carolina Master in Genetic Counseling student thesis project. Maddie Tjoelker is completing her master's degree and is conducting research on how individuals with a family history of cancer access screening and management.

If you are interested in taking this survey, please enter the following link into your internet browser on your computer, smart phone, or tablet. You may also scan the QR link to access the survey.

Link: https://uofsc.co1.qualtrics.com/jfe/form/SV_3PD56T5ogQ2101

Your involvement in this study will help healthcare providers best serve their patients and give easier access to much-needed services. If you have any questions regarding the study or trouble accessing the survey, please reach out to myself or to Maddie Tjoelker. Thank you for your consideration.

Sincerely,

[GENETIC COUNSELOR], MS, CGC	Maddie Tjoelker
[PHONE NUMBER]	(803) 386-7302
[EMAIL ADDRESS]	Madeleine.Tjoelker@uscmed.sc.edu

APPENDIX B: RECRUITMENT LETTER FOR PRISMA HEALTH-UPSTATE GENETICS MANAGEMENT CLINIC PARTICIPANTS

Dear Patient,

You are receiving this letter because you have been seen at the Prisma Health Genetics Management Clinic, and we are interested in hearing your feedback. To assess patient satisfaction in this service, we are asking patients to complete a survey. Through the information collected, we hope to provide better care for our patients.

The study involves completing an online survey. At the completion of the survey, you will be given the option for a follow-up telephone interview. The survey is available online through Qualtrics and should take no more than 15 minutes to complete. The survey involves questions about your understanding of your predisposition for cancer, how you are currently receiving care, and your level of satisfaction in the care you have received at the Genetics Management Clinic. Participation is completely confidential. Data from the study will be stored in a secure location, will not be placed in your health record nor linked to your name, and will be destroyed at the completion of the study. The phone interviews will be recorded for analysis by the research team, but all information that you provide will be kept confidential. The results of the study may be published or presented; however, your identity will remain anonymous. This study is completely voluntary, and there are no negative consequences should you withdraw from the study.

This study is a University of South Carolina Master in Genetic Counseling student thesis project. Maddie Tjoelker is completing her master's degree and is conducting research on how individuals with a family history of cancer access screening and management.

If you are interested in taking this survey, please enter the following link into your internet browser on your computer, smart phone, or tablet. You may also scan the QR link to access the survey.

Link: https://uofsc.co1.qualtrics.com/jfe/form/SV_8kzMhuAwTTLyQ9D

Your involvement in this study will help healthcare providers best serve their patients and give easier access to much-needed services. If you have any questions regarding the study or trouble accessing the survey, please reach out to myself or to Maddie Tjoelker. Thank you for your consideration.

Sincerely,

LeAnn Perkins, MSN, FNP-BC
(864) 455-1346

Maddie Tjoelker
(803) 386-7302
Madeleine.Tjoelker@uscmed.sc.edu

APPENDIX C: RECRUITMENT LETTER FOR PRISMA HEALTH-UPSTATE HIGH-RISK BREAST LIFETIME CLINIC PARTICIPANTS

Dear Patient,

You are receiving this letter because you have been seen at the Prisma Health High-Risk Breast Lifetime Clinic, and we are interested in hearing your feedback. To assess patient satisfaction in this service, we are asking patients to complete a survey. Through the information collected, we hope to provide better care for our patients.

The study involves completing an online survey. At the completion of the survey, you will be given the option for a follow-up telephone interview. The survey is available online through Qualtrics and should take no more than 15 minutes to complete. The survey involves questions about your understanding of your predisposition for cancer, how you are currently receiving care, and your level of satisfaction in the care you have received at the High-Risk Breast Lifetime Clinic. Participation is completely confidential. Data from the study will be stored in a secure location, will not be placed in your health record nor linked to your name, and will be destroyed at the completion of the study. The phone interviews will be recorded for analysis by the research team, but all information that you provide will be kept confidential. The results of the study may be published or presented; however, your identity will remain anonymous. This study is completely voluntary, and there are no negative consequences should you withdraw from the study.

This study is a University of South Carolina Master in Genetic Counseling student thesis project. Maddie Tjoelker is completing her master's degree and is conducting research on how individuals with a family history of cancer access screening and management.

If you are interested in taking this survey, please enter the following link into your internet browser on your computer, smart phone, or tablet. You may also scan the QR link to access the survey.

Link: https://uofsc.co1.qualtrics.com/jfe/form/SV_0pPw6LTin4WbKDP

Your involvement in this study will help healthcare providers best serve their patients and give easier access to much-needed services. If you have any questions regarding the study or trouble accessing the survey, please reach out to myself or to Maddie Tjoelker. Thank you for your consideration.

Sincerely,

LeAnn Perkins, MSN, FNP-BC
(864) 455-1346

Maddie Tjoelker
(803) 386-7302
Madeleine.Tjoelker@uscmed.sc.edu

APPENDIX D: SURVEY QUESTIONS FOR

PRISMA HEALTH-MIDLANDS PARTICIPANTS

Thank you for your participation in our study. All responses will be kept confidential and anonymous. In order to begin the survey, please indicate whether you consent to participating in our study.

a. I consent
b. I do not consent

1. Please select your gender
    a. Male
    b. Female
    c. Prefer not to answer

2. Please select your age from the following ranges:
    a. 18-25
    b. 26-30
    c. 31-40
    d. 41-50
    e. 51-60
    f. 61-70
    g. Over 70

3. Do you carry a positive mutation in a hereditary cancer gene?
    a. Yes
    b. No

<u>FOR MUTATION-POSITIVE PARTICIPANTS</u>

4. If yes, which gene?

5. Have you been diagnosed with cancer?
    a. Yes
    b. No

Please rate the following statements:

6. I feel I have adequate knowledge about my cancer risk and am able to manage my personal healthcare to meet the recommendations.
    a. Strongly disagree
    b. Somewhat disagree

c. Neither agree nor disagree
d. Somewhat agree
e. Strongly agree

Comments on Question 6:

7. I feel that my healthcare providers (primary care physician, obstetrician, gynecologist, etc.) have expert knowledge about the cancer screenings recommended for me.

a. Strongly disagree
b. Somewhat disagree
c. Neither agree nor disagree
d. Somewhat agree
e. Strongly agree

Comments on Question 7:

8. I have difficulty getting insurance coverage for the recommended cancer screenings.

a. Strongly disagree
b. Somewhat disagree
c. Neither agree nor disagree
d. Somewhat agree
e. Strongly agree

Comments on Question 8:

9. Some institutions have set up clinics designed to assist individuals who carry a positive mutation in a hereditary cancer gene get the recommended screenings and management. How interested would you be in attending this clinic if it were created at Prisma Health Richland?

a. Not at all interested
b. Likely not interested
c. Neutral
d. Somewhat interested
e. Very interested

10. Click and drag the following selections to rank the following services based on how important their incorporation into such a clinic is to you, with 1 being most important and 6 being least important.

- Cancer screenings (mammograms, breast MRI, etc.)
- Psychosocial support
- Genetic counseling
- Nutritional services
- Oncology
- Plastic surgery

11. Are there any additional services you would like to be included in this clinic?

12. How often would you want to attend this type of clinic?

a. Only once

b. Every 5 years
c. Every 2 years
d. Every year
e. Every 6 months
f. Other (please specify)

13. Based on this conversation, what other thoughts do you have regarding the establishment of this type of clinic?

FOR HIGH-RISK PARTICIPANTS

4. Have you been diagnosed with cancer?
a. Yes
b. No

5. Have you ever been told that you are at high risk for breast cancer?
a. Yes
b. No
c. Not sure

6. Have you ever been told to do more breast cancer screening than an annual mammogram (such as breast MRI annually)?
a. Yes
b. No
c. Not sure

Please rate the following statements:

7. I feel I have adequate knowledge about my cancer risk and am able to manage my personal healthcare to meet the recommendations.
a. Strongly disagree
b. Somewhat disagree
c. Neither agree nor disagree
d. Somewhat agree
e. Strongly agree

Comments on Question 7:

8. I feel that my healthcare providers (primary care physician, obstetrician, gynecologist, etc.) have expert knowledge
a. Strongly disagree
b. Somewhat disagree
c. Neither agree nor disagree
d. Somewhat agree
e. Strongly agree

Comments on Question 8:

9. I have difficulty getting insurance coverage for the recommended cancer screenings.
a. Strongly disagree

b. Somewhat disagree
c. Neither agree nor disagree
d. Somewhat agree
e. Strongly agree

Comments on Question 9:

10. Some institutions have set up clinics designed to help individuals who are at a higher risk for developing breast cancer get the recommended screenings. How interested would you be in attending this clinic if it were created at Prisma Health Richland?

a. Not at all interested
b. Likely not interested
c. Neutral
d. Somewhat interested
e. Very interested

Comments on Question 10:

11. Click and drag the following selections to rank the following services based on how important their incorporation into such a clinic is to you, with 1 being most important and 6 being least important.

- Cancer screenings (mammograms, breast MRI, etc.)
- Psychosocial support
- Genetic counseling
- Nutritional services
- Oncology
- Plastic surgery

12. Are there any other services that you would want to be offered at this clinic?

13. How often would you want to attend this type of clinic?

a. Only once
b. Every 5 years
c. Every 2 years
d. Every year
e. Every 6 months
f. Other (please specify)

14. Based on this conversation, what other thoughts do you have regarding the establishment of this type of clinic?

FOR BOTH GROUPS: *[At the completion of the survey]* We want to hear more about your personal experience getting care and your level of interest in the establishment of this new clinic. If you would be willing to complete a follow-up telephone interview with Maddie Tjoelker, the primary investigator on this project, please fill in your telephone number below, otherwise enter N/A to complete the survey.

APPENDIX E: SURVEY QUESTIONS FOR PRISMA HEALTH-UPSTATE GENETICS MANAGEMENT CLINIC PARTICIPANTS

Thank you for your participation in our study. All responses will be kept confidential and anonymous. In order to begin the survey, please indicate whether you consent to participating in our study.

a. I consent
b. I do not consent

1. Do you carry a positive mutation in a hereditary cancer gene?
    a. Yes
    b. No

2. If yes, what gene?
    a. ATM
    b. BRCA1
    c. BRCA2
    d. CDH1
    e. CHEK2
    f. PALB2
    g. PTEN
    h. TP53

3. When did you have genetic testing?
    a. Less than 6 months ago
    b. 6-12 months ago
    c. 12-18 months ago
    d. 18-24 months ago
    e. Over 24 months ago

4. When did you first begin attending the Genetics Management Clinic?
    a. Less than 6 months ago
    b. 6-12 months ago
    c. 12-18 months ago
    d. 18-24 months ago
    e. Over 24 months ago

5. Have you been diagnosed with cancer (not including non-melanoma skin cancer)?

a. Yes
b. No

6. On a scale from 1-10, with 1 being not at all satisfied and 10 being very satisfied, how satisfied are you with the services from the Genetics Management Clinic?

7. Has getting screenings for your cancer risk gotten easier since going to the Genetics Management Clinic?
    a. Yes
    b. No
    c. Not sure

    Comments on Question 7:

8. How often do you attend the Genetics Management Clinic?
    a. Less than once a year
    b. Once a year
    c. More than once a year

9. Do you feel this is too often or not enough?

10. What do you think is the most valuable service provided by the Genetics Management Clinic?

11. Would you recommend this clinic to others?

12. Is there anything that you wish would be added to the offerings of this clinic? (e.g., psychology, nutrition, etc.)

[At completion of the survey] We want to hear more about your personal experience getting care at the Genetics Management Clinic. If you would be willing to complete a follow-up telephone interview with Maddie Tjoelker, the primary investigator on this project, please fill in your telephone number below, otherwise enter N/A to complete the survey.

APPENDIX F: SURVEY QUESTIONS FOR PRISMA HEALTH-UPSTATE HIGH-RISK BREAST LIFETIME CLINIC PARTICIPANTS

Thank you for your participation in our study. All responses will be kept confidential and anonymous. In order to begin the survey, please indicate whether you consent to participating in our study.

a. I consent
b. I do not consent

1. When did you first begin attending the High-Risk Breast Lifetime Clinic?
    a. Less than 6 months ago
    b. 6-12 months ago
    c. 12-18 months ago
    d. 18-24 months ago
    e. More than 24 month ago

2. What made you decide to attend the High-Risk Breast Lifetime Clinic?

3. How often do you attend this clinic?
    a. Every 3 months
    b. Every 6 months
    c. Every 12 months
    d. Every 18 months
    e. Every 24 months
    f. Other (please specify)

4. On a scale from 1-10, with 1 being not at all satisfied and 10 being very satisfied, how satisfied are you with the services provided by the High-Risk Breast Lifetime Clinic?

5. Has getting mammograms and breast MRIs gotten easier since attending this clinic?
    a. Yes
    b. No
    c. Not sure

    Comments on Question 5:

6. What do you think is the most valuable service provided by the High-Risk Breast Lifetime Clinic?

7. Is there anything that you wish would be added to the offerings of this clinic?

[At completion of the survey] We are interested in hearing more about your personal experience with the High-Risk Breast Lifetime Clinic. If you would be willing to complete a follow-up telephone interview with Maddie Tjoelker, the primary investigator on this project, please fill in your telephone number below, otherwise enter N/A to complete the survey.

APPENDIX G: PRISMA HEALTH-MIDLANDS SERVICE PREFERENCES

Table G.1 Service Preferences Frequencies

FREQUENCIES						
SERVICE	**RANK**					
	1	**2**	**3**	**4**	**5**	**6**
Cancer Screenings	5	1	0	0	0	0
Psychological Support	0	0	1	1	2	2
Genetic Counseling	1	3	1	0	0	1
Nutritional Services	0	1	2	2	0	1
Oncology	0	1	1	2	1	1
Plastic Surgery	0	0	1	1	3	1

Table G.2 Ranked Service Preferences

OVERALL RANKING		
	Total	**Rank**
Cancer Screenings	35	1
Psychological Support	14	6
Genetic Counseling	26	2
Nutritional Services	20	3
Oncology	18	4
Plastic Surgery	14	5

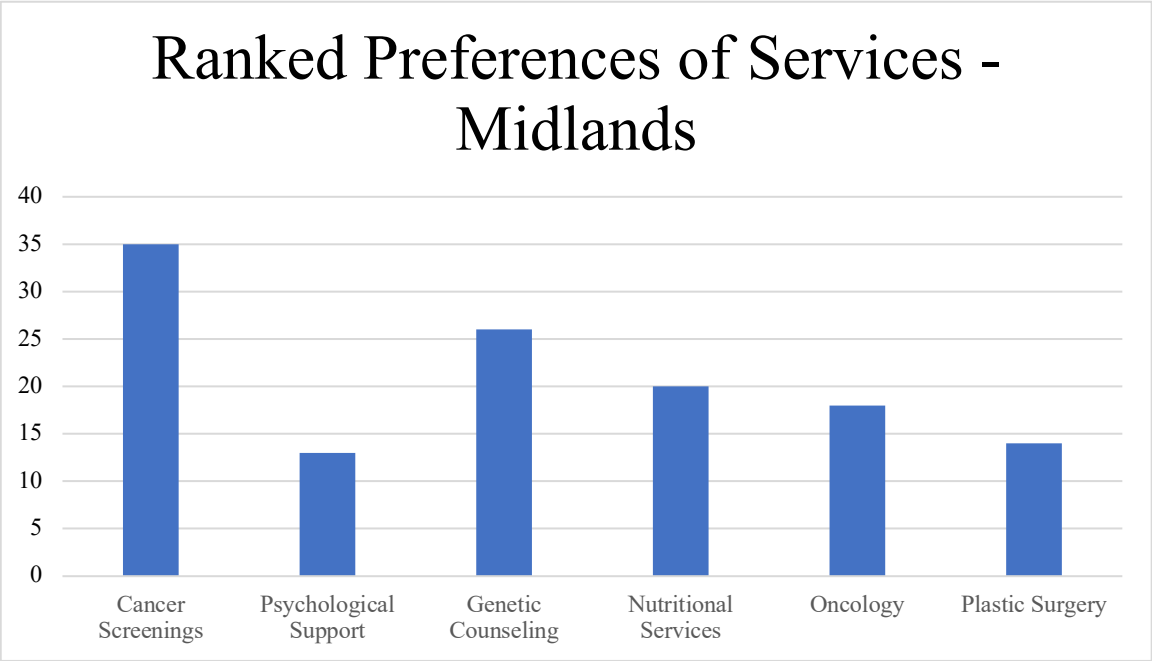

Figure G.1 Ranked Preferences of Services-Midlands

APPENDIX H: GENETICS MANAGEMENT CLINIC PARTICIPANT DATA

Table H.1 Genetics Management Clinic Participant Data

Clinical Information and Attendance		
	N	**Frequency**
When did you have genetic testing?		
Less than 6 months ago	0	0%
6-12 months ago	1	10%
12-18 months ago	1	10%
18-24 months ago	3	30%
Over 24 months ago	5	50%
When did you first begin attending the Genetics Management Clinic?		
Less than 6 months ago	0	0%
6-12 months ago	1	10%
12-18 months ago	1	10%
18-24 months ago	4	40%
Over 24 months ago	4	40%
Have you been diagnosed with cancer? (Not including non-melanoma skin cancer)		
Yes	6	60%
No	4	40%
Has getting screenings for your cancer risk gotten easier since going to the Genetics Management Clinic?		
Yes	5	50%
No	0	0%
Not sure	5	50%
How often do you attend the Genetics Management Clinic?		
Less than once a year	1	10%
Once a year	9	90%
More than once a year	0	0%

APPENDIX I: HIGH-RISK BREAST LIFETIME CLINIC PARTICIPANT DATA

Table I.1 High-Risk Breast Lifetime Clinic Participant Data

Clinical Information and Attendance		
	N	**Frequency**
When did you first begin attending the High-Risk Breast Lifetime Clinic?		
Less than 6 months ago	0	0%
6-12 months ago	1	11.1%
12-18 months ago	2	22.2%
18-24 months ago	1	11.1%
Over 24 months ago	5	55.6%
How often do you attend the High-Risk Breast Lifetime Clinic?		
Every 6 months	4	50%
Every 12 months	3	37.5%
Other—No longer attend	1	12.5%
On a scale from 0-10, with 0 being not at all satisfied and 10 being very satisfied, how satisfied are you with the services provided by the High-Risk Breast Lifetime Clinic?		
1	0	0%
2	0	0%
3	0	0%
4	0	0%
5	0	0%
6	0	0%
7	2	22.2%
8	2	22.2%
9	2	22.2%
10	3	33.3%
Has getting breast cancer screening (i.e., mammograms and breast MRIs) gotten easier since attending this clinic?		
Yes	6	66.7%
No	2	22.2%
Not sure	1	11.1%